1ST MENA REGION & AFRICAN INTERNATIONAL EB CONGRESS 2024 IN EGYPT

25th-28th April Cairo, Egypt

Hosted by Yasmine El Samra Foundation, Debra Egypt
Endorsed by Debra international, AfSHG, NRC, HGGRI, iDS

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WELCOME TO EGYPT

Egypt, officially The Arab Republic of Egypt is the cradle of civilisation, known with its famous and rich history through successive epochs with seven listed world heritage sites all historic except Whale Valley (Wadi Hitan) a natural heritage.Egypt has a unique geographical position at the heart of the Mena Region in NortheastAfrica, at the crossroads of Europe and Asia, on the Mediterranean and Red Sea, and is connected to Sub Saharan Africa through the Nile Valley.

Arabic is the official language of Egypt and Islam is its official religion. Cairo is the official capital of Egypt with a total population in excess 20 million people. Over 100 million inhabitants are distributed mainly around urban areas.

Cairo or Al Qahirah is the capital of Egypt. The Greater Cairo Region is the largest city in the Middle East and Africa, and its metropolitan area is the 16th largest in the world. The city embraces ancient and new, East and West.The Pyramids of Giza built over 4500 years ago, and the old Cairo region over 2000 years ago were the beginning of an ancient canal between the River Nile and the Red Sea. The river is a location still marked by several significant historic sites, many of which date back to the Greco-Roman period.

The city embraces architectural monuments dating from Arab and Ottoman times as well as a large functioning bazaar and an extensive, semi-walled medieval city endowed with more than 400 registered historic monuments including mosques, mausoleums, and massive stone gates dating to 130 CE. Cairo is rich in many shopping areas and chillout spaces, restaurants, coffee shops, cinemas, sports centers, shopping malls, and luxurious hotels

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EB Congress-Committees

Congress Co-Chairs

Prof. Mohamed El Darouti: *Professor of Dermatology & Dermatopathology, Cairo University, Egypt*

Prof. Ghada El-Kamah: *Professor of Clinical Genetics, Coordinator of The Hereditary Blood Disorders (HBD) and Genodermatoses clinics and research team, NRC, Egypt*

Scientific Committee Co-Chairs

Prof. Ghada El-Kamah: *Professor of Clinical Genetics, Coordinator of The Hereditary Blood Disorders (HBD) and Genodermatoses clinics and research team, NRC, Egypt*

Peter Marinkovich

Associate Professor of Dermatology, Faculty member of the Program in Epithelial Biology and the Stanford Cncer Biology Program, Director of thestanford Bullous Disease and Psoriasis Clinics as well as an attending dermatologist at the VA Palo Alto Medical Center, USA

EB Congress Organizing committee

Emad El Gamal
Al-Azhar University

Ritu Jain
Debra International

Ghada El-Kamah
National Research Centre

Hanaa Elsadat
DEBRA Egypt

Heba Ahmed
National Research Centre

Mohamed Badei
National Research Centre

Rasha Elhossini
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Cairo University

Hagar El Sayed

Cairo University

Reham W. Doss

Beni Suef University

Heba Anis

National Research Centre

Amira Abulfotooh

Alexandria University

Welcome Address

The Congress starts on the 25th April 2024 with a full-day session on genodermatoses aimed at including geneticists and dermatologists within the DEBRA community to raise awareness about incidence, and updates on management and research of orphan disorders, particularly EB, especially in the MENA region and Africa where consanguineous marriage is a still a tradition with an expected high incidence of these disorders.

The Scientific Committee is co-chaired by Professors Peter Marinkovich and Ghada El-Kamah together with an eminent panel of expert dermatologists, geneticists, biologists, surgeons, and industrial representatives to discuss updates on research and appropriate therapy, minimisation of adverse events, and strategies to improve patient quality of life.

Educational objectives:

- Review practice-changing data on the efficacy and safety of new and emerging therapies, and determining which treatments are appropriate for which patients.
- Efficacy data from practice-changing clinical trials on new and emerging therapies.
- Outline adverse events associated with novel therapies and approaches to mitigate these effects.
- Patients advocacy able to discuss their needs and whether fulfilled through novel approaches.

Ultimately the meeting aims at establishing a network bridging gaps both on patient advocacy and on technology transfer levels to the MENA region and Africa.

EB Congress Program

Evening prior to Day 1 - Welcome Reception				
Day 1 - Thursday 25th April - Genodermatoses				
PLENARY				
09:00-09:40		**Opening session: Welcome**		**Chair(s)**
09:00-09:15	**0h 35**	Opening remarks by Congress Chairs	Mohamed El-Darouti (Cairo University, Egypt) and Ghada El-Kamah (National Research Centre, Egypt)	Emad El-Gamal, Mohamed El-Darouti, and Ghada El Kamah
09:15-09:20		Opening remarks by Genodermatoses day Chair	Emad El-Gamal (Al-Azhar University, Egypt)	
09:20-09:25		Opening remarks by DEBRA Egypt Chair	Hanaa El Sadat (DEBRA Egypt, Egypt)	
09:25-09:30		Opening remarks by DEBRA International President	Ritu Jain (DEBRA International, Singapore)	
09:30-09:35		Opening remarks by Egyptian MP for the House of Representatives Human Rights Committee	Amal Salama (Egyptian Government, Egypt)	
09:40-10:40		**Session I: The situation**		**Chair(s)**
09:40-10:10	**1h**	New systematic therapeutic options for the treatment of dystrophic EB	Mohamed El-Darouti (Cairo University, Egypt)	Galal El-Anany and Samia Esmat
10:10-10:40		Genodermatoses challenge	Ghada El-Kamah (National Research Centre, Egypt)	
10:40-11:00		**Break**		
11:00-13:00		**Session II A: Genodermatoses - from orphan to priority**		**Chair(s)**
11:00-11:20	**2h**	Prenatal diagnoses experience in genodermatoses	Khaled Gaber (National Research Centre, Egypt)	Ghada El-Hoseiny, Hisham Fayek, Lamia Mohsen, Hanan Hosny Afifi, Mona Farag, Mona Essawy, and Tarek El-Badry
11:20-11:40		Molecular profiling of some genodermatoses	Khalda Amr (National Research Centre, Egypt)	
11:40-12:00		Genetics in Vitilligo	Rasha El Barbary (Al-Azhar University, Egypt)	
12:00-12:10		Genodermatoses - a view from Saudi Arabia	Khalid Al Aboud (King Faisal Hospital, Saudi Arabia)	
12:10-12:20		Study of Molecular and Clinical Spectrum of Some Egyptian Cases with Premature Grey hair	Nesrine Gomaa (Tanta University, Egypt)	
12:20-12:50		ED experience - clinical, molecular and dental care	Heba Ahmad, Eman Rabie, and Inas El Sayed (National Research Centre, Egypt)	
12:50-13:00		Gingival manifestations in oral genodermatoses	Heba Gamal El Din (National Research Centre, Egypt)	
13:00-14:20		**Session II B: Genodermatoses- Precision Medicine era**		**Chair(s)**
13:00-13:20	**1 h 20**	Photo-Anthropometry in Clinical Genetics	Khaled Helmi El-Wakeel (National Research Centre)	Emad El-Gamal, Hesham Zaher,

13:20-13:40		Medication Use Evaluation: TDM vs MDT	Sherif Kamal (EHA board for medication management and pharmacy affairs)	Mohammad El-Komi, Samia Esmat
13:40-14:00		Biomarkers in psoriasis	Hoda Yousry (Suez Canal University, Egypt)	
14:00-14:20		Pharmacogenomics in LMIC setting	Mohamad Nagy (Children's Cancer Hospital, Egypt)	
14:20-14:50		**Break**		
14:50-16:30		**Session III : Challenging cases and management**		**Chair(s)**
14:50-15:20	**1 h 40**	Genodermatoses among Burundian	Adel Botros Zaghloul (Al Haud Al-Marsoud Hospital, Egypt)	Ahmed Sadek, Amira Eid, Medhat El-Mofty, Mohamad El-Darouti, Mostafa Ibrahim
		Challenging cases in genodermatoses I	Ahmed Sadek (Al-Haud Al-Marsoud Hospital, Egypt)	
15:20-15:50		Challenging cases in genodermatoses II	Mohammed El-Darouti (Cairo University, Egypt)	
15:50-16:10		The use of non-coverage cost effective technique for management of epidermolysis bullosa hands in Egypt: a series of 16 cases	Adel Hussein Amr (Ain Shams University, Egypt)	
16:10-16:30		Fifteen years of oral rehabilitation of ectodermal dysplasia	Mohamad Abdel-Kader (National Research Centre, Egypt)	
16:30-18:30		**Session IV: Global perspective**		**Chair(s)**
16:30-16:50	**2h**	Pigmentary Lesion in EB patients	Hagar El Sayed (Cairo University, Egypt)	Eman Kamel, Ghada Abd El-Badea, Hamza Abd El-Raouf, Hanan Saleh, Noha Zakareya, Sahar El-Sayed
16:50-17:10		A better life for EB patients	Mona Mostafa (Cairo University, Egypt)	
17:10-17:20		Effect of different concentrations of human platelet rich plasma contrasted with fetal bovine serum on periodontal ligament stem cells	Maha Abdelfattah (National Research Centre, Egypt)	
17:20-17:30		Global science, genetic diseases and rare diseases in social networks. Perspective from the ethics of responsibility. Leading case 2023-2024	Liliana Virginia Ziadé (Lanús University; Museum Social University, QSN, Argentina)	
17:30-17:50		Genomic management of a polygenic disorder	Reham Doss (Beni Suef University, Egypt)	
17:50-18:10		Adapting the blended learning distributed classroom model for training multiple learner groups in genomic medicine	Pertunia Mutheiwana (H3ABioNet, South Africa)	
18:10-18:30		Wrapping	Mohamed El-Darouti (Cairo University, Egypt), Emad El-Gamal (Al-Azhar University, Egypt), and Ghada El-Kamah (National Research Centre, Egypt)	
18:30		**Close**		

Welcome reception - DEBRA International Roundtable of Companies (by invitation only)

Day 2 - Friday 26th April - Advocacy in EB

PLENARY				
08:30-09:00		**Opening session: Welcome**		**Chair(s)**
08:30-08:40	**0 h 30**	Opening remarks by Congress Chairs	Mohamed El-Darouti (Cairo University, Egypt) and Ghada El-Kamah (National Research Centre, Egypt)	Mohamed El-Darouti (Cairo University, Egypt) and Ghada El-Kamah (National Research Center, Egypt)
08:40-08:50		Opening remarks by DEBRA Egypt Chair	Hanaa El-Sadat (DEBRA Egypt, Egypt)	
08:50-09:00		Opening remarks by DEBRA International Chair	Ritu Jain (DEBRA International, Singapore)	
09:00-10:30		**Session I: Patients (and communities) as partners in research**		**Chair(s)**
		Panel session: The patient researcher: partnering with patients at various stages of therapeutic development and access		
	1 h 30	BUR-EB - the importance of understanding the socio-economic burden of the disease	Sophie Strobl (Sigmund Freud University, Austria / University of Freiburg, Germany)	Ritu Jain
		The COSEB initiative - developing a core outcome set for EB	Dimitra Kiritsi (Aristotle University of Thessaloniki, Greece)	
		Incorporating patient-reported outcome measures (PROMs) as proof of treatment efficacy	Dedee Murrell (University of New South Wales, Australia)	
		Patient driven evidence generation for advocacy - the Brazil project	Emeline Baillargeault (URGO Medical, France)	
10:30-11:00		**Break**		
11:00-12:30		**Session II: The patient advocate - a vital player in health care**		**Chair(s)**
		Panel session: Collaborating with patients: building on lived experiences for improved health outcomes		
	1 h 30	The role of patient advocates in health care	Ritu Jain (DEBRA International, Singapore)	Olivia Mullins
		The voice of EB patients - experts in their own condition	Andreas Miller (DEBRA Germany, Germany)	
		The voice of EB patients - experts in their own condition	Mehar Singh (DEBRA International, India)	
		Involving the patient voice in research - a research perspective	Ignacia Fuentes (DEBRA Chile, Chile)	
		Involving the patient voice in health care - a clinical perspective	Kalsoom Begum (University Hospitals Birmingham, UK)	
12:30-13:30		**Lunch**		
13:30-16:45		**Session III: Increasing the reach of DEBRA and awareness of EB worldwide**		**Chair(s)**
13:30-14:30		**Panel session: Best practices and challenges of patient advocacy organisations (PAOs) - what can we learn from each other?**		
	1h	DEBRA Australia	Zlatko Kopecki (DEBRA Australia, Australia)	Hanaa El-Sadat
		DEBRA Brasil	Priscila Keiko Matsumoto Martin (DEBRA Brasil, Brazil)	

		DEBRA Canada	Ryan Hultman (DEBRA Canada, Canada)	
		DEBRA Norge	Zlatko Orucevic (DEBRA Norge, Norway)	
14:30-15:30		**Panel session: Forming a patient advocacy group - how to go about it and what makes one successful?**		
	1h	DEBRA Pakistan and the role of culture	Faiza Ambreen (DEBRA Pakistan, Pakistan)	Toni Roberts
		DEBRA Croatia as the hub for expertise in the region	Vlasta Zmazek (DEBRA Croatia, Croatia)	
		DEBRA Egypt - "Finding a purpose"	Hanaa El-Sadat (DEBRA Egypt, Egypt)	
		DEBRA South Africa - "From patient to advocate"	Toni Roberts (DEBRA South Africa, South Africa)	
15:30-16:00		**Break**		
16:00-16:30	**0 h 45**	Shaping the patient leaders of tomorrow - DEBRA International Youth Council	Toni Roberts (DEBRA South Africa, South Africa) and Ira Jain (DEBRA Singapore, Singapore)	Olivia Mullins
16:30-16:45		EB Awareness Week 2024 - an international campaign	Megan Foster Flaherty (DEBRA International, UK)	
16:45-18:30		**Session IV: Creating contacts for life**		**Chair(s)**
16:45-18:30	**1 h 45**	Small groups networking session for clinicians, HCPs, advocates, patients, caregivers		Ryan Hultman, Olivia Mullins, Toni Roberts, and Megan Foster Flaherty
		Close		
		Gala Dinner I		
		Day 3 - Saturday 27th April - Research and Healthcare in EB		
		PLENARY		
09:00-12:00		**Session I: Updates in EB research I**		**Chair(s)**
09:00-09:30	**1 h 30**	New topical therapeutic options for the treatment of dystrophic epidermolysis bullosa	Mohamed El-Darouti (Cairo University)	Ghada El-Kamah & Zbigniew Ruszczal
09:30-10:00		Research update on genetics of EB; cell, gene, and protein therapies	Ignacia Fuentes (DEBRA Chile, Chile)	
10:00-10:30		Epidermolysis Bullosa: the newest news	Zbigniew Ruszczal (Sheikh Khalifa Medical City, UAE)	
10:30-11:00		**Break**		
11:00-11:30	**1 h**	Research update on symptom prevention and relief	Dimitra Kiritsi (Aristotle University of Thessaloniki, Greece)	Andrew South and Zlatko Kopecki
11:30-12:00		Research update on the EB development pipeline	Dedee Murrell (University of New South Wales, Australia)	
12:00-12:50		**Session II: Updates in EB research II**		**Chair(s)**

12:00-12:10		Evaluating ELK-003 Eye Drops for Ocular Manifestations in Dystrophic and Junctional Epidermolysis Bullosa Patients in Chile: A Pilot Study	Ignacia Fuentes (DEBRA Chile, Chile)	
12:10-12:20		An RNA marker of cancer in recessive dystrophic epidermolysis bullosa (RDEB)	Albert Mellick (University of New South Wales, Australia)	
12:20-12:30	**0 h 50**	Gene therapy for restoring collagen VII expression in Recessive Dystrophic Epidermolysis Bullosa	Julia Carnaz Benincasa (Albert Einstein Hospital - Education and Research Center, Brazil)	Andrew South and Zlatko Kopecki
12:30-12:40		Do EB cells react differently from healthy keratinocytes to biofilm presence in EB wounds?	Hadeer Ibrahim (University of Birmingham, UK)	
12:40-12:50		Development of bacteria activated stimuli response dressing for management of infection in Epidermolysis Bullosa	Zlatko Kopecki (University of South Australia, Australia)	
12:50-14:00		**Lunch**		
14:00-16:00		**Session III: Health care in EB**		**Chair(s)**
14:00-14:30		Keynote presentation: Supporting EB patients psychologically transitioning from paediatric to adult care	Sarah Elshakankiry (Malachite Institute for Behavioral Health, Egypt)	
14:30-15:00		Keynote presentation: Clinical and social management - the role of the EB CNS	Kalsoom Begum (University Hospitals Birmingham, UK)	
15:00-15:30	**2 h**	EB Clinet - an international network to support care and retain expertise	Sophie Kitzmüller (EB Haus, Austria)	Dedee Murrell and Zlatko Kopecki
15:30-15:40		Consensus-based guidelines for the provision of palliative and end-of-life care for people living with epidermolysis bullosa	Toni Roberts (DEBRA South Africa, South Africa)	
15:40-15:50		Managing cases of Epidermolysis bullosa in a resource-limited setting: An experience from Nepal	Niraj Parajuli (National Academy of Medical Sciences, Nepal)	
15:50-16:00		Nutritional deficiency in patients with congenital epidermolysis bullosa	Elena Belonogova (Butterfly Children Foundation, Russia)	
16:00-16:30		**Break**		
16:30-17:30		**Session IV: Health care in rare diseases**		**Chair(s)**
16:30-17:00	**1 h**	The importance of genetic counselling in rare diseases and cultural challenges faced	Nabeelah Peerbhai (University of Cape Town, South Africa)	Huda Rasheed and Yahia Zakaria Gad
17:00-17:30		Long term care and holistic approaches to care for rare disease and EB patients	Anica Ježić (Government of Croatia, Croatia)	
17:30		**Close**		
19:30		**Gala Dinner II - Grand Egyptian Museum**		
Day 4 - Sunday 28th April – Panel sessions				
PLENARY				

08:00-09:30		**Panel session I: How various initiatives can be consolidated towards positive healthcare outcomes for those with rare disorders**		**Chair(s)**
	1 h 30	Connecting the MENA region with global rare disease initiatives	Alexandra Heumber (RDI, Switzerland)	Ritu Jain
		Ethical engagement with pharmaceutical organisations	TBC	
		Connecting diagnosis to care	Shirlene Badger (Illumina, UK)	
		The importance of knowledge retention, education, and connection in the healthcare professionals' community	Sophie Kitzmüller (EB Haus, Austria)	
09:30-10:00		**Break**		
10:00-11:00		**Panel session II: Overcoming challenges to development and access to medicines and devices in the MENA region and internationally**		**Chair(s)**
	1 h	Value based Healthcare Integrated model for rare diseases	Sherif Kamal (EHA board for medication management and pharmacy affairs)	Hussein El-Sabagh,Mahmoud Bahgat, Mostafa El-Awady, and Ghada El-Kamah
		EDA role to ensure the ease of access and availability of medicines for rare diseases	Nourhan Tahoun (Egyptian Drug Authority, Egypt)	
		The National Consultancy Centre: A proposed initiative by the Drug Research Council to overcome challenges of pharmaceutical industry in Egypt	Maha Nasr (Drug Research Council, Ain Shams University, Egypt)	
		Development and access of advanced biologics in emerging countries: a real case study	Shaheer Bardissi (Minapharm pharmaceutical)	
11:00-11:30		**Break**		
11:30-13:00		**Panel session III: The patient journey and care approaches in different countries that meet the needs of patients and physicians in the MENA region**		**Chair(s)**
	1 h 30	Managing EB with limited resources, funding, and care teams - a South African experience	Carol Hlela (Red Cross Children's Hospital, South Africa)	Mohamed El-Darouti
		Establishing an EB House - a MENA region experience	Shaden Abdel Hadi (Sheikh Khalifa Medical City, Abu Dhabi, UAE)	
		Mutational analysis and management strategies in epidermolysis bullosa - an Indian experience	Rahul Mahajan (Postgraduate Institute of Medical Education and Research, India)	
		EB management updates - a MENA region experience	Khadija Sellami (Hedi Chaker Hospital, Tunisia)	
13:00-13:30		**Session IV: Poster prizes and closing remarks**		**Chair(s)**
13:00-13:15	**0 h 30**	Poster prizes	Mohamed El-Darouti (National Research Centre) and Ghada El-Kamah (National Research Centre)	Mohamed El-Darouti and Ghada El-Kamah
13:15-13:30		Closing remarks by Congress Chairs and DEBRA Egypt Chair	Mohamed El-Darouti (National Research Centre), Ghada El-Kamah (National Research Centre), and Hanaa El-Sadat (DEBRA Egypt)	
Close				

Speakers

International Speakers

Albert Mellick: University of New South Wales

Albert has been to develop techniques to investigate the role of non-cancer (or host derived stromal) cells in the development and spread of cancer. This includes study of bone marrow derived inflammatory and vascular stem cells. He has established strong (national & international) collaborative linkages, and he is panel member for the National Health & Medical and Australian Research Councils.

Alexandra Heumber Perry: Rare Diseases International

Alexandra is the Chief Executive Officer of Rare Diseases International, the global alliance of Persons Living with a Rare Disease. As CEO of RDI, Alex drives organizational strategy, rare disease raising awareness, access to diagnostics, treatments and care, and advocacy for the rights of patients around the world. In her presentation, Alex highlights how patient organizations in the MENA region can work towards and in partnership with RDI for improving healthcare policies for Persons Living with a Rare Diseases.

Andreas Miller: DEBRA Germany

Meet Andreas Miller, born with junctional epidermolysis bullosa. His early years were marked by harrowing experiences due to the lack of adequate EB care in Germany. These challenges later ignited a conviction for concrete and possible change and his motivation to actively advocate for the rights of EB patients. Andreas, who studied sociology, promotes hope, and fosters collaboration with fellow patients, researchers, and industry. For several years now, Andreas has served as Vice Chairman of DEBRA Germany, tirelessly advocating for improved resources and support for EB patients. His motivation stems from his conviction for tangible change and his faith in DEBRA Germany's capacity to effect positive transformation. He cherishes direct exchanges with patients, believing in support, empowerment, and empathy through shared experiences. Andreas firmly believes that everyone with EB has the right to lead a good life.

Andrew South: Thomas Jefferson University

Professor Andrew South is genetics graduate from the University of Leeds, who obtained his PhD from the University of London in 1999 whilst working on the human genome project with Dean Nizetic. Following this from 1999-2002 he was a postdoctoral research fellow at St John's Institute of Dermatology, St Thomas' Hospital in London with John McGrath. He then worked as a research associate with Ian Hart and subsequently as a lecturer in Irene Leigh's department at Bart's and the London from 2002 to 2007. He then moved his lab to the University of Dundee, based at Ninewells Hospital in the Division of Cancer Research, where he was until he moved to the United States in 2014. His laboratory interests centre on determining and dissecting basic mechanisms implicated in the underlying pathogenesis of cutaneous squamous cell carcinoma in patients with recessive dystrophic epidermolysis bullosa.

Carol Hlela: Red Cross Children's Hospital

Prof Hlela is a dermatologist, former Dermatology Head of Department at Red Cross Memorial Children's Hospital, and a researcher currently doing her fellowship in the United States since 2019. She grew up in KwaMashu Township, just outside Durban in KwaZulu Natal.

Dedee Murrell: University of New South Wales

Professor Dedee Murrell completed medical training at Cambridge and Oxford Universities, 3 years of internal medicine in the UK and USA, dermatology training at UNC-Chapel Hill, fellowships in dermatopharmacology at Duke University, blistering diseases, and cell biology at New York University, and then became a clinical scholar in psoriasis and epidermolysis bullosa at Rockefeller University with Prof James Krueger, completing a doctorate on the pathogenesis of blistering disorders. She developed the Australian EB diagnostic lab, MDTs for EB in Sydney, EB registry and pioneered the National EB Dressing scheme with DebRA Australia. She has developed and validated outcome measures for EB (QOLEB and EBDASI) pemphigus, pemphigoid and EB and designs and conducts clinical trials of new therapies for blistering diseases and inflammatory dermatoses. She has edited 6 books on blistering diseases, including the textbook, Blistering Diseases. She was President of the ASDR 2019-21 and Co-Chair of the EB research conference in Osaka, Japan in 2023. She has delivered invited lectures in 50 countries, including many in the Middle East.

Dimitra Kiritsi: Aristotle University of Thessaloniki

Professor and Consultant dermatologist & Group leader, Department of Dermatology, Medical Center – University of Freiburg. M.D., PhD in Experimental Dermatology, University of Freiburg. Consultant and head of the Immunofluorescence laboratory and the "Clinical Trial Center-Fragile Skin", Department of Dermatology, Medical Center - University of Freiburg, since 2015. Professor of Dermatology, Aristotle University of Thessaloniki, Greece, since 04/2023. Fellow of the European Board of Dermatology and Venerology. Associate Editor for Frontiers in Genetics and Frontiers in Pediatrics. Has more than 110 publications in high impact factor journals.

Elena Belonogova: Butterfly Children Foundation, Russia

a doctor at the Pirogov Clinic of High Medical Technologies at St Petersburg University. She has received letters of appreciation from members of the State Duma and the Civic Chamber of the Russian Federation for her efforts in improving the system of medical and social care for patients with orphan skin diseases.

Emeline Baillargeault: URGO Medical

Emeline has worked in the wound care and healing sector since 2012, she obtained a university degree in wounds and healing in 2019 and then decided to specialize in professional relations, medical training and awareness of chronic wounds, diabetes and to epidermolysis bullosa. She is also an active member of the Urgo Foundation, managing projects in around fifteen countries to support and train healthcare professionals to help them better heal their patients. Passionate and committed to EB patients and their families, in 2023 she promotes the partnership with Debra International and becomes the ambassador of Urgo Medical in all their joint projects.

Faiza Ambreen: DEBRA Pakistan

Faiza is a Global Health expert with over two years of experience in evidence generation and health policy analysis. She holds and MSc in Global Health Policy form the London School of Economics. Currently, she is serving as a Research Consultant at the Global Surgery Policy Unit within the Department of Health Policy at LSE. She conducted pioneering research by using consensus-building approach for prioritizing unmet needs for people living with rare diseases in Pakistan and developed a conceptual policy framework to inform national strategy. As a parent of child with EB, she passionately advocates for people with rare diseases. As a director of DEBRA Pakistan, she has spearheaded various EB awareness campaigns in collaboration with DEBRA International. Through her work, she aims to enhance access to pharmacological products for rare diseases in LMICs, ensuring that geographic and socioeconomic barriers do not hinder access to life-saving treatments.

Ignacia Fuentes: DEBRA Chile

Ignacia is based in Santiago, Chile and joined DEBRA International in September 2018. Ignacia studied biochemistry in Chile, did her PhD in genetics in Germany, and returned to Chile to work in EB in 2013. Ignacia loves travelling. Thus, she has been a visiting scientist to learn about EB in many EB centres worldwide, such as the EB-Haus Austria, Thomas Jefferson University USA, and others. Ignacia is currently the Research Director of DEBRA Chile and leads a great team of people working for EB patients in different aspects of the disease, such as understanding the genetics and physiology of EB, participating in clinical trials on EB, and providing molecular diagnosis to EB patients in Chile. She is also an Assistant Professor at the Pontificia Universidad Católica de Chile. Although Ignacia has no personal connection to EB, she is committed to support with her knowledge and experience on EB research, DEBRA Chile, and EB patients worldwide.

Ira Jain: DEBRA Singapore / Youth Council

Ira is a management consultant in Financial Services Advisory who lives with EB Simplex in Singapore. She has been a member of DEBRA Singapore since its inception in 2015. As a current executive committee member, her responsibilities include grant management, event organisation, and publicity & communications. Ira is also a founding member & Vice President of the Debra International Youth Council, committed to helping meet the often unseen needs of youths with EB. She graduated from University College London with a degree in Chemistry in 2021, and is passionate about scientific research, sustainability, and making a difference. On weekends, you can find her reading a good book or watching a game of ice hockey. Go Leafs!

Julia Carnaz Benincasa: Albert Einstein Hospital - Education and Research Center

Biologist graduated from UFSCar/São Carlos, with a master's degree in Cellular and Structural Biology from UNICAMP/Campinas, and a Ph.D. from UNIFESP-EPM/São Paulo, focusing on Molecular Biology in the field of Neurosciences/Bioengineering. She was driven by the dream of working with regenerative therapies and contributing to the development of protocols that bring significant improvements to patients' lives, especially in areas where conventional pharmaceutical methods have not reached yet. Currently, she works as a postdoctoral researcher at the Albert Einstein Israelite Institute of Education and Research, with Dr. Ricardo Weinrich's group, focusing on gene therapy in the study and development of a treatment for Epidermolysis Bullosa. Over the past 10 years, both in Brazil and the United States, she can wo rk on Research and Development (R&D) projects, conducting data collection, molecular and cellular assay analyses, mammalian cell culture, DNA/RNA extraction and analysis, protein studies, molecular cloning, and genomic sequencing analysis and interpretation. As a researcher, her main responsibilities include project management, data analysis, and writing scientific articles and technical reports. Working in the field of regenerative therapies has developed a critical perception in identifying problems, planning, and executing experiments, as well as qualitatively and quantitatively analyzing various types of results.

Khadija Sellami: University of Sfax

Khadija is associate professor at the Dermatology venereology Department, Hedi Chaker University Hospital of Sfax, University of Sfax (south-east of Tunisia) Deputy secretary general of the Tunisian association of Pediatric dermatology. Qualifications in Pediatric dermatology, Sexology, Wound healing, Simulation in healthcare and Cosmetic dermatology. She Has special interest in pediatric dermatology and genodermatoses especially Epidermolysis bullousa and ichthyoses. She Has actively contributed to the creation of the EB regional committee in Sfax, which gathers different specialists involved upon EB patient needs to offer a multidisciplinary care.

Khaled Al Aboud: King Faisal Hospital

Khalid Al Aboud, holds Bachelor of Medicine and Bachelor of Surgery (MBBS) from King Abdulaziz University, Saudi Arabia. Recently he finished his fellowship in dermatopathology from Wake Forest University, Winston-Salem, North Carolina, USA. Khalid Al Aboud who is currently the head of public health department in King Faisal Hospital, Makkah, Saudi Arabia, has diverse medicals interests and has several publications in Sexually Transmitted Infections, dermatology, pathology, and public health. In 2008, he received Zakon prize from the Society of History of Dermatology. He conducted national researches on several public health problems including leprosy, leishmania and skin cancers. He is on the Editorial and Advisory Boards of several peer-reviewed scientific journals.

Megan Foster Flaherty: DEBRA International

Megan is based in the UK and has recently joined DEBRA International in December 2023 as the Operations and Communications Coordinator, helping Liv with the managing the day-to-day operations of the organization, and managing the communications such as the newsletter, social media, and campaigns. She has previously worked in another small, rare disease charity after graduating from university with a degree in journalism. She has worked across the charity sector in different roles including event and social media management, as well as admin. Despite having no personal link to EB, Megan is very passionate about supporting small charities who help families with rare diseases and who provide support, research, advocacy, and education about rare diseases after losing her youngest sister to a life limiting condition.

Mehar Singh: DEBRA International

My name is Mehar, I am 17 years old, and I have RDEB. Talking a bit about myself, from a young age, I've immersed myself in the world of drones, driven by an insatiable curiosity for technology and innovation. Despite the physical limitations, my determination to explore the skies has only grown stronger. Beyond my love for drones, I am deeply passionate about spreading awareness about EB and advocating for patients like myself. I initiated the DEBRA International Youth Council, a platform dedicated to raising awareness about EB and advocating for improved treatments and support for those affected by the condition. Through my advocacy work, I strive to shed light on the challenges faced by individuals living with rare diseases and to foster a community of support and understanding.

Nabeelah Peerbhai: University of Cape Town

Nabeelah is currently a genetic counselling intern at the University of Cape Town in South Africa where She's also involved in variant interpretation for the Neuromuscular Disease Africa Initiative and where she completed her MMedSci degree in Genetic Counselling. Her research explored the perceived experiences of Sickle Cell Disease Patients in the Emergency Centre's of South African hospitals. Prior to this, she completed my honours in Human Genetics at the University of Witwatersrand and an undergraduate degree in Biochemistry and Psychology from the University of Johannesburg. In addition to her academic roles and qualifications, she is the director of the Sickle Cell South Africa Non Profit Company and actively involved in patient advocacy with Rare Diseases South Africa.

Niraj Parajuli: National Academy of Medical Sciences of Nepal

Doctor of Medicine, Senior consultant dermatologist and assistant professor at National Academy of medical Sciences of Nepal. He is a clinical dermatologist with more than 12 years of experience. He is interested in neglected tropical diseases, rare skin diseases, blistering diseases, teledermatology and use of AI in dermatology.

Olivia Mullins: DEBRA International

Liv is Executive Director for DEBRA International based in the UK having joined the organisation in 2016. In her role, she is responsible for overseeing the strategic and operational efficiencies of the organisation's programmes and staff. She has previously worked in project and account management, and the education sectors within the UK and Spain. Liv studied Spanish and Italian at university with a focus on translation. Despite having no personal link to EB, Liv was driven to join DEBRA having seen how support, like DEBRA's, has helped her father cope with a severe and debilitating skin condition. A self-professed travel enthusiast, she's always looking for the next adventure

Pertunia Mutheiwana: H3ABioNet

Pertunia is a Training Curriculum Administrator at the Computational Biology Division in the Health Sciences Faculty at the University of Cape Town (UCT). She offers training and curriculum coordination, facilitation, and administration support for H3ABioNet, a Pan African Bioinformatics Network for the Human Heredity and Health in Africa (H3Africa) consortium. Pertunia coordinated the 2022 and 2023 African Genomic Medicine Training Initiative (AGMT) course, an H3ABioNet-funded course that aimed to develop relevant competencies in genomic medicine and provide basic genomics and genetics education to healthcare professionals in Africa. Additionally, Pertunia is a qualified award-winning teacher with a master's degree in education and has been accepted to study for a PhD at UCT under the Accelerated Transformation of the Academic Programme (ATAP). Furthermore, Pertunia is interested in education and training, particularly in curriculum development, educational technology, and educational psychology.

Priscila Matsumoto Martin: DEBRA Brazil/Albert Einstein Hospital

Priscila Keiko Matsumoto Martin is a researcher with a background in Medical Biological Sciences from the Federal University of São Paulo (2008). She completed her master's (2011) and doctoral (2016) degrees in Biological Sciences (Molecular Biology) at the same institution. Currently, she works at the Research Institute of Albert Einstein Hospital, focusing on gene therapy projects using base editing for recessive dystrophic epidermolysis bullosa and sickle cell anemia. Additionally, she serves as the Director of Research at Debra Brazil. Priscila has extensive experience in biochemistry, molecular and cellular biology, and genetics, with a particular emphasis on gene and cell therapy for monogenic diseases.

Rahul Mahajan: PGIMER

Rahul currently works as Additional Professor in the Department of Dermatology, Venereology and Leprology, Postgraduate Institute of Medical Education and Research (PGIMER), Chandigarh, India. He has published more than 150 research papers in indexed journals, and in recent years, his work includes clinical and translational research on epidermolysis bullosa and congenital ichthyosis, pediatric atopic eczema, pediatric psoriasis, pediatric alopecia areata, and infantile hemangioma. He is the Lead investigator in 10 research projects. He has utilized these projects to set up diagnostic next-generation sequencing facilities for EB and congenital ichthyosis at his institute in India, and initiated pilot clinical trials on investigational drugs in these disorders. For his work in the field of Dermatology, he was awarded the Fellowship to Royal College of Physicians, Edinburgh in 2022, and the membership to the Indian “National Academy of Medical Sciences” in the year 2016. He is the Deputy Editor of the Indian Dermatology Online Journal.

Ritu Jain: DEBRA International

Ritu is President of DEBRA International, and the board member of various rare disease organisations including APARDO, RDI, IRDiRC, and GlobalSkin. Ritu is wife and mum to brave EB warriors based in Singapore. She joined DEBRA International in 2016, a year after founding DEBRA Singapore. Her firm commitment to making a difference to under-served EB patients in South and South-East Asia drives her service as an EB Without Borders Ambassador. Ritu also serves on Professor of sociolinguist at the Nanyang Technological University where she teaches graduate and undergraduate courses. She is equally passionate about her research on the impact of Language Policies on immigrant minorities in multilingual sites and publishes on the topic. In her 'spare time', Ritu loves to run and still dreams of completing a full marathon one day!

Ryan Hultman: DEBRA Canada

Ryan is currently the Vice President of DEBRA Canada and has served on the board for 12 years. He has a 13-year-old with RDEB. He lives in Vancouver, Canada and when not working as the owner of a residential design company and EB caregiver, works diligently to make sure each daughter has the same experience and opportunities, including the great outdoors. The recent highlight was taking his child with RDEB on a 5-day remote wilderness canoe circuit with portages. For the 11th straight year, he runs a DEBRA Canada fundraiser at the top of Grouse Mountain called Bella's Ball. A broomball event (like hockey, no skates and on ice) where able bodied people must run about the ice to get a fun and often painful view of what it is like to live with EB. Like the game, the reality of EB for Ryan and family is to be an active participant and not sit back and watch the game. The event seeks out EB sufferers from across Canada to attend as a belle or beau of the ball. Providing a weekend away and avenue to build an ever-stronger national EB community.

Shaden Abdel Hadi: Khalifa University

Consultant of Pediatric Dermatology, European Board and Arab Board Certified in Dermatology & Venereology, Belgian Board Certified in Pediatric Dermatology (including clinical training at Seattle Children's Hospital, Seattle, USA). She holds the Belgian Board in Human Medical Genetics from the Belgian Society of Human Genetics BeSHG (including training at Guys & St. Thomas Genetic Skin Diseases Center in London, UK). Moreover, she is a Fellow of the Royal College of Physicians of Edinburgh (FRCP Edin). Additionally, she is a Fellow of the American Academy of Dermatology (FAAD), the European Academy of Dermatology and Venereology (FEADV) and the European Society of Pediatric Dermatology (FESPD). Dr. Shaden is a Member of the Editorial Board of the Dermatologic Therapy, a John Wiley & Sons Ltd. journal. She is also the Director of the ACGMEi accredited Dermatology Residency Program at Sheikh Khalifa Medical City, Abu Dhabi. Dr. Shaden holds the distinction of being the first fully licensed Pediatric Dermatologist in Abu Dhabi. She has been highly active in academics since 2012 including active teaching and supervision in her specialties in both Belgium and the UAE. Dr. Shaden Abdelhadi is an Assistant Professor of Dermatology at College of Medicine and Health Sciences, Khalifa University in Abu Dhabi. In 2021, she was appointed as a founding committee member and section chair, National Institute of Health Sciences (NIHS) for the Emirati Board of Dermatology. Moreover, she is the founder and head of the Multidisciplinary Epidermolysis Bullosa Center at SKMC.

Shirlene Badger: Illumina

Shirlene Badger is Patient Advocacy lead for EMEA at Illumina, Inc. In this role, she leads the Patient Advocacy strategy and engagement across the fields of rare and undiagnosed genetic disease, reproductive health and oncology. Trained as a medical sociologist, she spent over fifteen years leading academic research initiatives at the University of Cambridge and elsewhere that sought to elevate the impact of patient voice in the development and implementation of novel and experimental medical technologies.

Sophie Kitzmüller: EB Haus

Working for and with EB patients for 13 years in different roles, Dr Sophie Kitzmüller established a deep understanding of the needs and strengths of patients, families and HCPs, patient advocacy and industry relations. She started her career as a research associate in the lab of the expertise center EB house Austria. 3 years of intense research led to the offering of a PhD candidate position at the University of Salzburg, where she earned her PhD title 3.5 years later. With newfound interest in clinical research, she transitioned back to the EB house Austria as clinical trial coordinator. Experience and dedication lead to the transition to clinical trial management and in 2019 to becoming head of the EB academy and EB Clinet, an international network for HCPs working with EB patients.

Sophie Strobl: Sigmund Freud University Vienna

Sophie is a dedicated researcher and lecturer at the Department of Psychology, Sigmund Freud University Vienna, Austria. Specializing in health psychology and psychological testing, Sophie's work focuses on improving the quality of life in chronic and rare diseases through interdisciplinary approaches bridging medicine and psychology. Currently pursuing her Ph.D. at the University of Freiburg, Sophie's research centers on "quality of life in Epidermolysis bullosa." She has been part of the research project "Epidermolysis bullosa: Burdens and helpful factors for quality of life," led by Dr. Gudrun Salamon for four years and is part of the BUR-EB Study group. Sophie's commitment to advancing knowledge in her field is reflected in her latest publication in the British Journal of Dermatology, where she investigated the psychometric properties of the Instrument for Scoring Clinical Outcomes of Research for Epidermolysis Bullosa patient score (iscorEB-p), a patient-reported outcome measure.

Toni Roberts: DEBRA South Africa

A dedicated advocate for rare disease patients, Toni co-founded DEBRA South Africa after years of connecting and supporting individuals affected by Epidermolysis Bullosa (EB) around the country. Armed with her degrees in Psychology and 10-year experience as a qualified Life Line lay counsellor, she provides practical assistance and emotional support to patients near and far. Recently appointed as the President of the newly established DEBRA International Youth Council, Toni continues her mission to uplift and upskill the next generation of leaders. With being half Swiss, her efforts extend beyond borders as she travels both locally and internationally, championing for EB awareness, advocating for improved guidelines, and amplifying the voices of patients. With unwavering commitment and compassion, Toni strives to make a tangible difference in the lives of those impacted by EB, embodying the spirit of empathy and empowerment.

Vlasta Zmazek: DEBRA Croatia

My son Matija, 1983-2018, was my inspiration and force for battle to achieve better quality of life for persons with EB and their families. I was founder of DEBRA Croatia in 1996, collaborating and learning from DEBRA UK. As DEBRA Croatia, we have organised numbers of Seminars and Congresses for the entire region. For Croatian families we have achieved all items for care, free of charge as well as treatments needed. As EBWB regional ambassador, I supported development of DEBRAs in the region: Serbia, Bosnia, Slovenia, and Poland. As President of Rare Diseases Croatia in parallel I was member of the Board of Directors at EURORDIS for 10 years. I am one of the founders of DEBRA International and representative on the GlobalSkin. Last year,I was invited to the UN as an example of holistic patient-centred care organised through our Rare Resource Centre.

Zlatko Kopecki: DEBRA Australia / UniSA

Zlatko is a Senior Research Fellow based at University of South Australia in Adelaide, South Australia. His doctoral studies centered on developing novel therapies for patients with skin blistering diseases and he has been working in the field since 2007. His latest work is focussed on combating wound infection in blistered wounds. Zlatko has been a DEBRA Australia director since 2013 and joined DEBRA International in 2016. He is passionate about the EB Without Borders programme and has led the development of DEBRA Cuba. Additionally, Zlatko's clinical studies led to development of the in-home nursing program for EB sufferers in Australia. As Treasurer of DEBRA International he hopes to work with the Executive Committee in developing long-term sustainability and support for national DEBRAs around the world.

Zlatko Orucevic: DEBRA Norway

Zlatko has been a part of the DEBRA Norway family since his younger sister was born with recessive dystrophic epidermolysis bullosa (RDEB) when he was three years old. He is the deputy leader of DEBRA Norway and has been a board member since 2018, his membership in the organization stretching back to 1993, reflecting a longstanding dedication to advocacy work. Outside of his advocacy work, Zlatko holds a master's degree with a specialization in industrial economics and technology management, and he works as a project manager in a consulting firm. Beyond his career, he enjoys skiing and swimming, symbolizing his love for an active lifestyle. Driven by a desire to raise awareness about EB and support the work of DEBRA Norway, Zlatko is dedicated to making a difference for "butterfly children." His contributions reflect a combination of personal passion and professional skill, aimed at contributing to a better future for those affected by EB.

National Speakers

Adel Botros Zaghloul

Consultant dermatology and Venereology, Cairo Skin VD hospital (Al Haud Al Marsoud) from 1981 - 2022. Consultant Sexually Transmitted Infections at WHO (EMRO and Genéve 2005- 2013). Life member American Academy of Dermatology 1987- present.

Adel Hussein Amr

Professor of plastic and reconstructive surgery sub-specializing in hand surgery. He graduated from Ain Shams University in Cairo, Egypt, in 1998 and has since been working there, ascending from residency to assume his current position as professor. After completing basic training in plastic surgery, he specialized in reconstructive hand surgery, obtaining his masters and doctorate degrees in microneural surgery. He is a fellow of UNESP university in São Paulo, Brazil. Dr Adel is also a consultant of hand surgery in several governmental and private hospitals in Egypt, where his work mainly focuses on trauma patients and congenital hand deformities. He has been operating on EB patients' hands for almost a decade, creating a multidisciplinary team for management of this condition.

Ahmed Mohsen Sadek

PhD, MSc, DBA, MB, BCh - Dermato-Venereology & Laser Consultant Manager of Cairo Hospital for Dermatology & Venereology (Al-Haud Al-Marsoud), Cairo, Egypt . Owner & Manager of Dr. Sadek Clinics. Vice Manager of Cairo Health Affairs Directorate for the Egyptian Fellowship Affairs. Cairo Health Affairs Directorate Clinical Research Coordinator . Trainer at The Egyptian Dermatology Fellowship Program , Secretary General of the Egyptian Society of Dermatology & Venereology. Secretary General of the European Society of LASER & Energy Based Devices. Executive Board Member of the International Dermoscopy Society (IDS), Chair of Imaging in Medical Aesthetics Task Force. IMCAS Scientific Board Member. Course Director of Egyptian Ministry of Health & Population Clinical Dermatology Mega Training Program. International Fellow of the American Academy of Dermatology. International Fellow of the European Academy of Dermatology & Venereology. Deputy Editor of the Egyptian Journal of Dermatology & Venereology. Dermato-Oncology Section Editor of Dermatology Practical & Conceptual Journal.

Emadeldin Abdelmoneim Elgamal

Professor Emeritus of Dermatology,Venereology and Andrology Damietta Faculty of Medicine, Al-Azhar University Membership of National Societies: Egyptian Society of Dermatology, Venereology and Andrology: Past board member. Alexandria Society of Dermatology, Venereology and Andrology: Board member and Secretary General. Egyptian Society for Psoriasis, established and registered officially 2016:President. Membership of International Societies: American Academy of Dermatology AAD European Academy of Dermatology and Venereology EADV Skin Inflammation and Psoriasis International Network SPIN:Representative of Egypt and member of Scientific Committee. Former Egyptian coordinator of the “Genodermatoses and Mediterranean “ project. Supervisor and mentor of many MD and Master theses. Reviewer of National and International Journals. Publications on: Hair diseases and surgery, psoriasis, psoriatic arthritis, cryosurgery, lichen planus, leprosy, acne, urticaria, melasma, scars, male and female sexual dysfunction.

Eman Rabei

Eman Rabie is a post-doc researcher at the Medical Molecular Genetics department of the Institute of Human Genetics and Genome Research (IHHGR), National Research Centre (NRC), Egypt. In 2012, Eman joined the Genodermatoses research group as a research assistant aiming for diagnosis and treatment of this class of orphan diseases. In 2016, Eman earned her M.Sc. in Biotechnology from the American University in Cairo (AUC) following her research on molecular diagnosis of Xeroderma Pigmentosum (XP) in Egypt. Her PhD in Biotechnology last year focused on molecular diagnosis of autosomal recessive Ectodermal Dysplasia (ED) using next generation sequencing (NGS). For both disorders, Eman has developed and applied targeted cost-effective mutational testing serving inpatients from clinical genetics clinics and referred outpatients across Egypt. Besides her experience in NGS and data analysis, Eman has worked on novel therapeutic approaches, particularly, gene editing of induced pluripotent stem cells using CRISPR-Cas9.

Ghada El-Kamah

MBBCh, MSc, PhD. Professor Clinical Genetics, founder of The Hereditary Blood Disorders (HBD) and Genodermatoses clinics and research team, Human Genetics and Genome Research Institute (HGGRI), National Research Centre (NRC). Focus rare disorders, dysmorphology, community genetics, science management and genetic education. Executive Committee member of; National Strategic Programs for Biotechnology and Genetic Engineering, the African Society of Human Genetics, Arab Association for Genetic Research, the Egyptian Committee for Pathology Training (ECPT)-Genetics, editorial board of the Journal of Genetic Engineering and Biotechnology, associate editor Journal of Human Genomics, ethical coordinator HGGRI and NRC-IRB, head of HGGRI scientific and cultural relations committee. Representing Egypt at Global Globin Network, Human Variome Project. Guest lecturer at YARSI University, Indonesia and expert member of the Genodermatoses Network Scientific Committee (2000-2018). PI and participant in 30 national and international research grants concerned with clinical, diagnostics, educational, and ethical aspects of human genetics.

Hagar El Sayed

Lecturer of dermatology, Kasr Alainy School of Medicine, Cairo University. Spent her residency and post-doctoral training in Kasr Alainy school of medicine following up patients suffering epidermolysis bullosa, different genodermatoses, bullous diseases, psoriasis among others. Mentored by Professor Mohammad El Darouti, she was following up hundreds of epidermolysis bullosa patients. She contributed to studies and research reports on EB, psoriasis, vitiligo, laser and energy-based devices. She's a member of the psoriasis and LASER units in Kasr Alainy School of Medicine. She is responsible for the assessment of residents' training in the psoriasis and LASER units as well as a member of the residents' training team. She is currently conducting several research grants, mentoring dermatology fellows in different topics of dermatology and is the organizer of the weekly scientific meeting of the dermatology clinic planned by Prof. ElDarouti. She contributed her research to several national dermatology conferences and workshops.

Hanaa El Sadat

Mother of Yasmin El Samra, who was born with RDEB (1997-2012), wife to a professor of ophthalmology, and mum to 3 gentlemen free of EB and a grandmother of a boy and a girl free of EB. She has been the founder of Yasmin EL Samra Charity Foundation since 2014. Chairperson of the foundation after being officially established in 2018. The foundation became one of Debra International groups in 2021. She is a member of the Board of Committee at Debra International from 2022 until 2024. The Yasmin ElSamra Foundation, DEBRA egypt, is hosting the EB congress 2024 in EGYPT.

Heba Mostafa Gamal El Din

PhD holder in Oral Medicine, Periodontology, Oral Diagnosis and Radiology, Al Azhar University, Dental School. Researcher at the Oro-dental Genetics Department, Human Genetics and Genome Research, National Research Center, Cairo, Egypt. Lecturer at Oral Medicine, Periodontology, Oral Diagnosis and Radiology Department, Must University from 2020-2023. Member in a STDF project titled “A regenerative approach for treating bony defects in conditions associated with premature loss of teeth”. Collective B member in the European Society of Human Genetics.

Hoda Yousry

Associate Professor of Medical Genetics, Director of Molecular Biology Unit, Quality Manager Center of Excellence in Molecular and Cellular Medicine, Faculty of Medicine - Suez Canal University. Holder of a degree in medical education and bioinformatics. Alumnus of the international Leadership and Management Program (UNILEAD 2021) - Oldenburg University, Germany and of Interdisciplinary Research Excellence Program. She has a special interest in technology-based entrepreneurship where she founded her startup “Med Predict”. Dr Hoda was the PI and Co-PI of multiple international and national funded projects for cancer, dermatological diseases, autoimmune diseases, COVID19, research infrastructure development. Organizer of several international and national events in her field and mentor of more than 40 MSc and PhD students. Her main research interest is related to cancer and autoimmune diseases.

Inas Sayed

an Associate professor of Oro-dental Genetics in Institute of Human Genetics and Genome Research, National Research Centre, Egypt. She got her Masters and PhD degrees from Cairo University. Her duties in Oro-dental Genetics Department include oro-dental examination of patients attending Orodental Genetics clinic, Neurogentics clinic as well as Limb malformation and skeletal dysplasia clinic which aids in their diagnoses. She is the PI of number of research projects involving the diagnosis of Ectodermal Dysplasias.

Khalda Sayed Amr

Professor and former head of Medical Molecular Genetics Department, Human Genetics and Genome Research Division of National Research Centre (NRC), Cairo, Egypt. She received her PhD in “Human Genetics” Cairo University (Egypt). Awarded; the Academy of Scientific Research and Technology award for encouragement of ongoing research, Misr El Kheer Charity for the highest indexed publication (Misr El Kheer charity), and the National Research Centre award for Scientific Advancement in Medical Sciences. Prof. Khalda was selected at Marquis Who’s Who World International Scientists in Medicine and Healthcare. She mentored 15 Master degrees and 27 PhD students, participated in more than 32 international, national projects as a principle investigator or consultant. She authored more than 64 publications on different genetic disorders in high indexed international Journals, cited 2061. h-index: 23 ORCID NO: 0000-0001-8472-5911 an genetics.

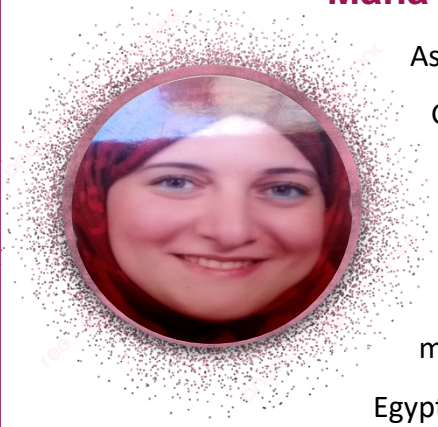
Khaled Helmi El-Wakeel,

MBBCh, Faculty of Medicine, Ain-Shams University, M.Sc, Medical Genetics, Ain-Shams University, Ph.D. in Medical Genetics, Alexandria University, Post graduate two years Diploma in Statistics, Institute of Statistical Studies, Cairo University, Assistant professor, Biological Anthropology Department, Medical Research Division, National Research Centre. Beside his main specialty in Anthropology-Genetics, he has experience in Information theory, we developed a function (infoFunction) which measure amount of information in a system of different hypothesis such as a list of differential diagnosis. It resembles Shannon's Entropy function with some differences, tatistical analysis, as well as developing modules for statistical analysis, and development of Software systems including Tr@cer, a software for measurements (Applied in Orthodontics, Endodontic, Genetics, anthropometry, etc), NeuroNet, an artificial intelligence engine (neural network), ZageL, a communication software (SMS & email gateway), Other database including medical database.

Khaled R Gaber

Professor of Prenatal Diagnosis, founder and former Head of the first integrated prenatal diagnosis and fetal medicine department in NRC and Egypt (2003-2021). In addition, he established Outpatient clinics, for Maternal-Fetal care and recurrent pregnancy loss.The researches performed in the department include clinical, ultrasonographic and cytogenetic, biochemical and molecular studies. This achievement was in collaboration with Professor Mona K Farag and Professor Sanaa Helmy. Prof. Gaber had his basic education at the Peres Jesuites School and graduated from Faculty of Medicine Cairo University (1981). He obtained his MSc Degree from Cairo University in Obstetrics and Gynecology and his Ph.D. Degree in Human Genetics from Alexandria University. He got his experience in the field of prenatal diagnosis during his stay in Germany (Ulm University). He has over 60 peer-reviewed publications in national and international journals, mentored more than 50 M.Sc. and Ph.D. students in his field. In addition to his contribution in over 25 projects either as a principle or participant investigator.

Maha Abdelfattah Ibrahim Abdelfattah



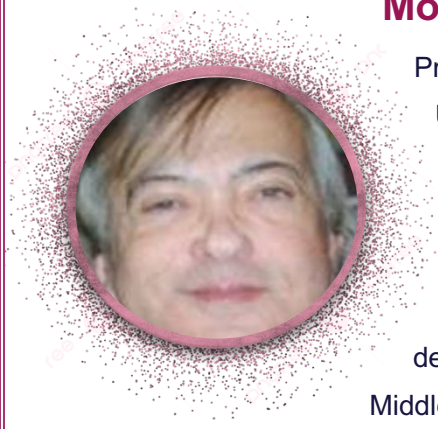
Assistant professor of Oro-dental Genetics department, Human Genetics and Genome Research institute, National Research Centre, Giza, Egypt. *Bachelor* of Oral and Dental Medicine, Faculty of Oral and Dental Medicine, Cairo University, Cairo – Egypt. Master Degree in Oral medicine and Periodontology, Faculty of Oral and Dental Medicine, Cairo University, Egypt. Doctoral Degree in Oral medicine and Periodontology, Faculty of Dental Medicine, Suez Canal University, Egypt. Master and Doctoral thesis were done in Beijing Institute of Dental Research, under supervision of Prof. Dr. Songlin Wang Vice President Capital Medical University (Beijing, China), Scientific Director of Beijing Institute of Dental Research, Professor and Chief Salivary Gland Disease Center & Molecular Lab for Gene Therapy and Tooth Regeneration.

Dr. Maha Nasr

A Professor and Researcher of Pharmaceutics and Industrial Pharmacy, and the Executive director of the academic cooperation and international relations sector at the Faculty of Pharmacy, Ain Shams University, Egypt (h index 40). She has published more than 140 international papers in the field of drug delivery and nanotechnology. Dr. Maha was awarded several international awards, among which is the African Kwame Nkrumah regional award in the field of Life Sciences 2019, and Khalifa award for Education in 2023 (Distinguished University Professor at the level of the Arab world in the field of Higher Education). She is currently a member of the Expert committee for reviewing monographs in the Egyptian Pharmacopoeia in the Egyptian Drug Authority EDA, a member of the Drug Research Council by a Ministerial decision, and a member of the National Drug committee.

Mohamad Nagy

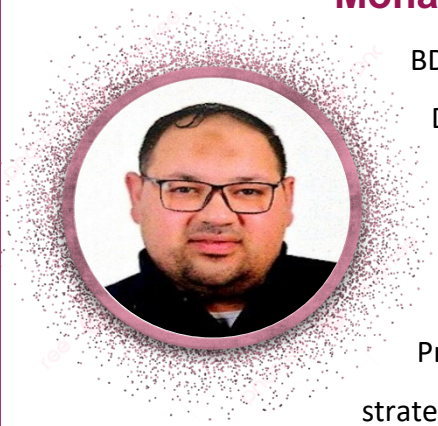
Pharmacy Director and the founder of the Personalized Medication Management Unit at the Children's Cancer Hospital 57357. He is a clinical instructor at Colorado university school of pharmacy. Mohamed is considered one of the Experts in clinical pharmacogenomics and Nutrigenomics in Middle East and Africa. He dedicated his genetic knowledge not only to patients but also to allow healthy individuals to find the best sports and nutrition routine tailored to their unique individual genetic makeup in order to achieve the best health outcomes. Mohamed is an instructor of Personalized Medicine and Nutrigenomics at Tanta University and Alexandria University. Mohamed is a founder in Pharmacogenomics Access & Reimbursement Coalition (PARC) and he is also a founder of the Standardizing Laboratory Practices in Pharmacogenomics (STRIPE). Currently, Mohamed is leading the Pharmacogenomics Research Network Developing Countries Committee for the area of Middle East and Africa. He is a member in the Clinical Pharmacogenetics Implementation Consortium (CPIC) and he participates in developing the international guidelines for clinical guidelines.



Mohammad Ali El Darouti

Prof. of Dermatology & Dermatopathology Kasr Alainy School of Medicine, Cairo University (1991 till present). Instructor of Dermatopathology in Lybia, Oman and Kuwait, examiner at Dermatopathology board in Kuwait and Teaching Clinical Cases in Bahrain. Developed stem cell therapy for Epidermolysis Bullosa, described Muckle Wells syndrome, Familial gigantic Melanocytosis and the new distinctive skin lesion of Necrolytic Acral Erythema in Hepatitis-C patients. He established: the first dermatopathology unit and the first unit for treatment of critical skin disorders in the Middle East, the first dermatology unit and the first unit for management of life threatening skin diseases in Egypt. He founded Dermatopathology as a subspeciality in Medical Doctorate of Dermatology, Kasr Alainy. Authored six books in dermatology and dermatopathology. Mentored several fellows, residents and medical students and holds a weekly teaching clinical round in Kasralainy outpatient dermatology clinic. Awarded Omar bin Abdul Aziz Al- Sheikh Prize for scientific research and community service 2014, Honored several times by the faculty of medicine and by Egyptian Medical Syndicate 2023.

Mohamed Abdel-Kader



BDS Oral and dental medicine ,MDS of Dental Prosthodontics , Cairo University & DDS. Dental Prosthodontics Al Azhar University Associate Prof, Orodental Genetics Department, Oral & Dental Research Division, National Research Center. CAIRO, EYGPT. Founder of ECTODERMAL ORAL REHABILITATION CLINIC , Orodental Genetics Department, National Research Center. CAIRO, EYGPT. Principal investigator of National research centre internal project entitled “A new strategy for Prosthodontic Rehabilitation of patients suffering from Ectodermal dysplasia syndrome” Associate Prof, Removable Prosthodontics Dept., Misr International University (MIU) – Egypt., & AL Ahram Canadian University (ACU) – Egypt. Consultant of Prosthodontics and Dental Implantology at Ftama Dental Clinic & Dental district dental care center (Dr. Ihab Ibrahim) Cairo Eygypt. AFSHG (African Society of Human Genetics) young investigator prize: 1st prize for poster presentation. November, 2007.

Nesrine S. Gomaa

MSc, MD. Lecturer of dermatology, Faculty of Medicine, Tanta University. Co-founder of pediatric dermatlogy unit- Tanta University Hospitals. Spent her residency and master degree at Tanta university and was genuinely interested in pediatric dermatology especially genodermatoses. Inspired and mentored by Professor John McGrath, head of St John's Institute of Dermatology, King’s College, London.UK . She jointly has conducted her MD/PhD degree studies at St John's Institute under Prof. McGrath supervision. She has participated in many researches and also gained clinical experience at Guy's and St Thomas' hospitals (GMC registered since 2016 till present). Back home, at Tanta University hospitals with the unique location at the centre of Delta and its dense population, serving hundreds of patients, where Dr Nesrin gets the chance to continue helping her community.

Rasha El Barbary

Professor of dermatology and venereology at Al-Azhar University. With years of experience in this field, she is known for her expertise in diagnosing and treating various skin diseases and sexually transmitted diseases. Dr. Al-Barbari is committed to educating the next generation of dermatologists through her teaching and research activities. She also actively participates in community outreach programs to raise awareness about skin health and disease prevention. Dr. El-Barbari is one of the founders of the Egyptian Society of Dermatopathology and a member of the Egyptian society of Psoriasis and the Egyptian Society of Dermatology and Venereology. She is a former associate editor at jweds magazine. She has conducted many researches into the genetic factors related to skin diseases. She is know the vice dean of education and student affairs in the Faculty of Nursing Al-Azhar University.

Reham William Naguib Doss

Assistant professor of Dermatology, Faculty of Medicine, Beni-Suef University-Egypt. M.B.B.CH. Faculty of Medicine, Cairo University, Beni-Suef branch. M Sc Dermatology and Venereology from Cairo University, Egypt. Diploma in Medical Laser Applications, National Institute of Laser Enhanced Sciences, Cairo University. MD Dermatology, Cairo University, Egypt. Member of the Royal Colleges of physicians (MRCP) in UK 2016. Academic Appointments: Served as resident at the Dermatology department, Beni-Suef University Hospitals, and Cairo University Hospitals 2004 - 2007. Assistant Lecturer (2007-2011), Lecturer (2011-2018) of Dermatology, Faculty of Medicine, Beni-Suef University.

Sherif Kamal

Sherif Kamal is one of a second generation Egyptian clinical pharmacists, with more than 20 years of experience in this field, involved in the planning, designing and implementation of pharmaceutical care in oncology centers and hospitals as senior Clinical Pharmacy consultant. Sherif Kamal is the Advisor of the Chairman of EHA board for medication management and pharmacy affairs. He earned his BS in Pharmacy 1998, an MSc in Clinical Pharmacy and is currently a PhD candidate at the Cairo University School of Pharmacy. Sherif has also completed a visiting fellowship at St. Jude's Children Hospital in the United States in 2008. He is a certified Professional trainer by the AUC and made a healthcare management diploma in AUC with the thesis title :Pharmacy Digitalization - Drug Information and Pharmacy Resource Services . For more than 20 years, Sherif has led a team that implemented clinical pharmacy services at various hospitals throughout Egypt and the region . He is the recipient the Egyptian Hospital Pharmacist Association Award and founder of the Egyptian Association of Clinical Pharmacists in 2010 . He is a member of several international organizations including the International Society for Oncology Pharmacists, the European Society of Clinical Pharmacy, the American Society of Health-System Pharmacist, and the American College of Pharmacy. He is the Vize President of the European Society of Oncology Pharmacy Global for all Non-European countries. He is a member of the Board for International Accreditation of Hospital Pharmacy in ASHP. He has conducted a wide variety of research studies involving the pharmacotherapeutics management of pediatric oncology diseases and presented his work at international meetings. Sherif is working with university of Colorado , American Society of Health system pharmacist ,American college of clinical Pharmacy ,American council for Pharmacy Education and European society if oncology Pharmacy to develop the pharmacy academy program for thehealth sciences Academy. We started Pharm D program, Residency program and many short courses. The program also introduced successfully the future of pharmacy which Is pharmacogenomics and personalized medication management. Sherif was the leading consultant working to implement clinical pharmacy in Egypt(4 Ministry of health Hoapitals;2 Military Hospitals and 3 Police Hospitals) . Sherif is also leading an African initiative to improve healthcare via clinical pharmacy and medication management in Africa ,including Sudan,Ethiopia,Malawi , Uganda and Botswana. He was the first International Residency program director in the region and the first Pharm D program director in collaboration with Colrado run by a Hospital. His primary research interest focuses on pharmacy practice, patient safety, pharmacogentics, Pharmacoeconomics, pharmacovigilance , Green Pharmacy and the pharmaco-epidemiology of pediatric cancer. Sherif is also a professionally and practically qualified project manager in fundraising and business development, with more than 20 years experience in this field involved in the fundraising activity of the National Cancer Institute and Children Cancer Hospital Foundation 57357. He is a Certified Lean six sigma green belt.

Oral Abstracts for the EB Congress

Day 1 – Genodermatoses

Session I: The situation

Mohamed El-Darouti

New systematic therapeutic options for the treatment of dystrophic EB

EBD is the most mutilating form of EB. Here we present our journey in reaching the best regimen for treating EB with detailed explanation of the mechanism of action of the given drugs and interesting pictures before and after treatment.

Ghada El-Kamah

Genodermatoses challenge

Genodermatoses are a large group of heritable disorders where dermatological manifestations are the clue to diagnosing a wide array of multisystem affection disorders with clinical symptoms and signs differing in severity from mild to lethal. This special nature requires multidisciplinary approaches for those disorders considered as orphan diseases contrary to their prevalence within consanguineous populations such as MENA region and some African countries. As most genetic disorders, the absence of curative treatment, imposes tremendous psychological burden on the patients and their families as well as financial burden on the governmental level maintaining lifelong symptomatic management. In confrontation to those challenges, Genodermatoses as an entity was introduced to the National Research Centre (NRC) as the first clinic and research team in the region/possibly world-wide at the year 2000. This collaborative continuously growing team provides genetic assessment to confirm diagnosis, aid management decision as well as prevention strategies where genetic counselling, carrier detection and premarital/prenatal and preimplantation services among others were ultimately offered. Tremendous progress has been made in understanding the genetic basis of different forms of genodermatoses: Type, number, as well as the spectrum of mutations and subsequent quantitative or qualitative disruption of protein expression. In the genomic era, this information provides the bases for several novel therapeutic approaches paving paths for cure rather than current prevention and symptomatic management.

More importantly, the team is continuously working on raising awareness about the disorders among health care professionals, Policy makers as well as the public through education, networking and advocacy together with continuing trials to grasp the knowhow of latest therapeutic researches worldwide.

Session II A: Genodermatoses - from orphan to priority

Khaled Gaber

Prenatal diagnoses experience in genodermatoses

Gaber K.
Prof. Prenatal Diagnosis - National Research Centre – Egypt

Genodermatoses are a broad group of disorders with specific and no-specific skin-based phenotypes, most of which are monogenic disorders. The genetic and clinical heterogeneity bring great challenges for diagnosis. The discovery of pathogenic mutations in inherited skin diseases represents one of the major landmarks of late 20th century molecular genetics. One of the most significant benefits of translational research has been the development of prenatal diagnosis for the couples at risk of recurrence of severe inherited skin diseases. Initial tests were based on skin biopsy sampling, but these have largely been superseded by DNA based analyses. A team approach and collaboration between Dermatologists, Geneticists, and Obstetric Geneticist enabled us in the National Research Centre – Egypt in optimization the patient management in terms of diagnosis, improve genetic counseling and earlier and safer prenatal diagnostic service.

Khalda Amr

Molecular profiling of some genodermatoses

Diagnosing genodermatoses remains a challenging task due to their rarity and the diversity of their heterogeneous phenotypes. Approximately one third of hereditary disorders show characteristic cutaneous findings. Genodermatosis are considered among orphan disorders although they constitute serious health problem in some North African countries. There is no exact estimate of genodermatoses prevalence in Egypt, however, a set of features affect markedly the prevalence and distribution of genetic diseases in our region; founder effect, genetic drift, selective pressure, and consanguinity favouring heritable conditions. Genetic studies have led to the recognition of novel disorders. Next generation sequencing (NGS) has entered routine clinical practice guiding precise correlation of clinical and molecular findings, unveiling definitive diagnosis and determining causality of novel variants among genetic skin disease patients with complex phenotypes. Among serious genodermatoses frequently seen in genodermatosis clinic at National Research Centre, Egypt (NRC) are Epidermolysis Bullosa, autosomal recessive congenital ichthyosis (ARCI), Neurofibromatosis (NF), Ectodermal Dysplasia (ED), and Xeroderma Pigmentosum (XP). Other ultra-rare skin syndromes were also referred to benefit from accurate clinical and molecular investigation with either targeted sequencing and/ or NGS analysis. Thorough application of different genetic testing led to identification of numerous novel causative variants confined to Egyptian patients and/or shared with other North African cohorts due to founder effect and migration. Genetic analysis has impacted our understanding of the molecular pathology of genodermatoses, unveiled the population-specific mutational landscape of these heterogenous phenotypes, and ultimately provided proper genetic counselling, preventive approaches and future therapeutic strategies.

Rasha El Barbary
Al-Azhar University, Egypt

Genetics in Vitiligo

Khalid Al Aboud

Genodermatoses; a view from Saudi arabia

Ahmad Al Aboud[1] , Khalid Al Aboud[2] , and members of dermatology dept[2] .
[1] Dermatology Department , King Abdullah Medical City, Makkah, Saudi Arabia.
[2] Dermatology Department, King Faisal Hospital, Makkah, Saudi Arabia

Abstract

Background: Genodermatoses remains a major public health problem in eastern hemisphere and developing countries. This is because of different factors like consanguinity. Aims and Objectives: To present families affected with different genodermatoses , from Saudi Arabia. Materials and Methods: Families affected by Kindler syndrome , hereditary hypotrichosis simplex , and familial multiple trichoepitheliomas well be discussed. Results, limitations and conclusions: National steps to decrease genetic disorders (including genodermatoses) , such as pre-marital medical screening will be presented. Active measures can decrease the burden of genodermatoses in the community.

Keywords: Dermatology, consanguinity, Genodermatoses, Saudi Arabia, Skin disorders

Nesrine Gomaa

Study of Molecular Genetic and Clinical Spectrum of Some Egyptian Cases with Premature Grey Hair

Abstract
Objective: This work aimed at studying genetic and molecular spectrum of some cases who presented with early grey hair. Patients and methods: This study included 30 patients having premature grey hair. Complete general and cutaneous examination was done. Genetic analysis was done to confirm diagnosis and study the genetic background of each syndrome. Results: Most cases were diagnosed as Chediak Higashi syndrome (13 patients, 43.33%). Griscelli type 2 (5 patients, 16.67%), Griscelli type 1(3 patients, 10%), primary haemophagocytic-lymphohistiocytosis (2 patients, 6.67%) and Niemann Pick syndrome (one patient, 3.33%). 6 patients (20%) remained undiagnosed. Conclusion:The results of this study expand the spectrum of premature grey hair syndrome clinically and genotypically, improving the overall diagnosis and prognosis of these conditions.

Heba Ahmad, Eman Rabie, and Inas Sayed

ED experience - clinical, molecular, and dental care

Ectodermal dysplasia is a heterogeneous group of disorders characterized by primary defect of two or more structures of ectodermal origin including the skin, teeth and appendageal structures such as hair, nail, exocrine and sebaceous glands. To date, more than 160 ED phenotypes have been described, and the underlying

molecular pathogenesis of more than 50% remains unknown.When identified, ED-causing genetic variations expectedly affect genes encoding members of either major developmental pathways or complex structural molecules that are vital for the structural and functional development of the skin and its ectodermal tissue derivatives. Our talk tackles different aspects of ED based on clinical diagnosis and management experience from Genodermatosis and Oro-dental clinics together with molecular diagnosis experience from Medical Molecular Genetics department of NRC. The most common form of ED is HypohidroticEctodermal Dysplasia. The clinical spectrum varies between mild to severe life-threatening ectodermal affection due to hyperthermia as a consequence of anhydrosis/hypohidrosis and recurrent infections. Tooth agenesis, which is particularly severe in ED, malformed teeth, hyperthermia and recurrent infections of ED patients place major psychological and financial burdens on the patients and their families. Prosthetic rehabilitation is important to restore function and esthetics which are severely impaired in these patients. Other syndromic ED identified in our clinics include Ellis–van Creveld syndrome, and Ectodermal dysplasia-syndactyly syndrome. For molecular diagnosis, we present results of utilizing targeted NGS panel and whole exome sequencing for diagnosis of 75 ED cases highlighting the impact of molecular diagnosis in patient management, genetic counselling, and eligibility for the emerging therapeutic options.

Heba Gamal El Din

Gingival manifestations in oral genodermatoses

Heba Mostafa Gamal El Din [1]

[1] Oro-dental Genetics Department, Human Genetics and Genome Research, National Research Center, Cairo, Egypt.

Aim: Genodermatoses are a group of rare hereditary dermatological disorders with single gene mutation. Oral Genodermatoses (OG) are inherited dermatological disorders with oral manifestations where the oral mucosa, tongue, gingiva, palate, dentition and salivary glands maybe also affected. In this poster we are reviewing the different gingival lesions in oral genodermatosis and emphasizing on the importance of oro-dental examination in patients with genodermatosis as sometimes the gingival lesions maybe the first clue to the diagnosis of these disorders or might help with the differential diagnosis for such diseases as in Papillon Lefevre syndrome, Infantile systemic hyalinosis, Ehlers danlos syndrome, Chediak Higashi syndrome, Kindler syndrome and many others. Materials and methods: The present poster provides an overview of all thedifferent gingival lesions associated with genodermatosis. Results: Some genodermatosis present with distinct gingival manifestations that may be pathognomic in diagnosis of such diseases, especially if the dermatological lesions were mild or not yet recognized. Gingival manifestations vary from severe periodontitis as in Papillon Lefevre syndrome, lack of zone of attached gingiva in Periodontal-type Ehler danlos syndrome, fibrotic gingival enlargement in Infantile Systemic Hyalinosis to unilateral gingival hemangiomas in Sturge Weber syndrome. Conclusion: It is very important for dermatologists and dentists to recognize that not only some genodermatoses show concomitant gingival lesions but also manifestations of some of these diseases may be preceded by the gingival lesions. Therefore, we are shedding light on the significance of oro-dental examination which can aid in the early diagnosis of such diseases.

Session II B: Genodermatoses, Precision Medicine Era

Khaled Helmi El-Wakeel

Photo-Anthropometry in Clinical Genetics

Anthropometric measurements are an important part of the diagnosis of dysmorphic children. Direct measurements are the standard way to obtain these measurements. It is well known for every clinical geneticist that it is time-consuming and not easy, especially in non-cooperative patients. Photographic records are another important part of clinical dysmorphology as "A picture is worth a thousand words". Traditional measurements from photos lack absolute measurements due to the magnification error, which is why the ratios were used in photo measurements. There exist many 3D systems for medical imaging but they lack the practicalities in daily clinical practice as patients need to be in fixed positions for a longer time, and they lack advanced measurement capabilities. We have applied Tr@cer as a 2D measurement system for clinical genetics for performing simple measurements such as nose length, and palpebral fissure length, up to sophisticated measurements such as interpupillary distance (measure from calculated pupils' centers), ear areas, the level difference in bilateral structures such as the vertical asymmetry of the outer canthi on standardized patients' photos with n complexity level, i.e. It is very easy to measure corneal diameter within seconds with very high accuracy. The system is equipped with 17 different types of geometric objects, which enable designing datasets with different complexity levels. For example, you could define a line parallel to Frankfort's line, and calculate projections from other landmarks to that constructed line. Photos calibration is a core functionality in Tr@cer performance as the system calibrates photos in 2D, i.e. there is a vertical scale as well as a horizontal scale. Although the system facilitates anthropometric measurements, there are still some obstacles such as the proper positioning of patients when they resist stability (non-cooperative patients) but it shortens the needed cooperation time which is very beneficial. The new system is sophisticated and simple to use and enables getting anthropometric measurements in an easier way, in less time, and accurately. It opens doors to define new measurements which were not practically possible before. It can help us to change many qualitative traits into corresponding quantitative ones. It facilitates creating patient profiles (such as hand profiles in z scores). The quantifying of traits is a new opportunity to enforce artificial intelligent diagnostic systems such as neural networks to achieve better results. Tr@cer can empower existing medical database systems through integration, completing the lack of these functionalities.

Sherif Kamal

EHA board for medication management and pharmacy affairs

Medication Use Evaluation: TDM vs MDT

Hoda Yousry

Biomarkers in Psoriasis

Abdallah H.[1&2]
[1]Medical Genetics Unit, Histology and cell Biology Department, Faculty of Medicine, Suez Canal University, Ismailia, Egypt
[2]Center of excellence in Molecular and Cellular Medicine, Faculty of Medicine, Suez Canal University, Ismailia, Egypt

Psoriasis is a complex polygenic disease in which exposure of hereditary predisposed persons to specific environmental stresses could provoke disease development. In the precision medicine era, advances on omics technologies unraveled novel biomarkers from diverse biological signatures associated with psoriasis pathogenesis. In this talk, I highlighted the current status of psoriasis omics-driven biomarkers, emphasizing the role of the transcriptomic, epigenomic, proteomic, and metabolomic biomarkers, in addition, I gave special consideration to the current status of psoriasis molecular signatures in Egypt. Furthermore, I reflected my insights considering the limitations and future directions of the current psoriasis biomarker discovery strategies.

Mohamad Nagy
Children's Cancer Hospital, Egypt

Clinical pharmacogenetics implementation hopes or hype.

Session III: Challenging cases and management

Adel Botros Zaghloul
Genodermatoses among Burundian

Dr. Adel Botros Zaghloul
Consultant Dermatology, Al Haud Al Marsoud Hospital, Cairo, Egypt

Background; As a volunteer in dermatology, I visited, many African countries, among them Burundi, considered to be one of the poorest country in the world and only two physicians practicing dermatology, to serve approximately eight millions habitant. Observations: Clinical examination took place in “Regent Charles hospital” and “Prince Louis Rwagasore Clinique” in Bujumbura the capital of Burundi. Sixty to eighty patients were examined on daily basis, coming from all over the country. I recognized the commonest genodermatoses, which include, in order of frequency patients with; neurofibromatosis, ichthyosis, epidermodysplasia verruciform, oculocutaneous albinism, xeroderma pigmentosa, epidermal nevi along the lines of Blaschko, giant congenital melanocytic nevi, spinal dysraphism, palmoplantar keratoderma, Klippel Trenaunay syndrome . I am going to demonstrate this collection of cases and more. Key messages: Dermatologist are rare in poor African countries. Most patients presenting with major genodermatoses “Orphan diseases”, may get frustrated after consultation and may not seek any medical and social advice any more. Without early diagnosis and treatment, skin diseases can negatively impact individuals, families and communities by causing long-term disability,stigmatization and mental health conditions. Teledermatology, should be more implemented in many African countries and to be supported by most dermatology centers in Africa and elsewhere. Volunteerism and patient support groups, generic list of dermatologic agents provided by the WHO and the Regional Dermatology Training Center (RDTC) in

Moshi, Tanzania. In addition orphan products should be included in the strategY of treatment whenever applicable.

Ahmed Sadek
Al-Haud Al-Marsoud Hospital, Egypt

Challenging cases in genodermatosis I

Mohamed El-Darouti
Cairo University, Egypt

Chalenging cases in genodermatoses II

Adel Hussein Amr
Ain Shams University, Egypt

The use of non-coverage cost effective technique for management of epidrrmolysis bullosa bands in Egypt: a series of 16 cases.

Mohamed Abdel Kader
National Research Centre, Egypt

Fifteen years of oral rehabilitation of ectodermal dysplasia

Session IV: Global perspective

Hagar El Sayed
Pigmentary Lesion in EB patients

Epidermolysis bullosa (EB) represents a heterogeneous group of mechano-bullous disorders genetically rooted in an intrinsic defect of the structural components of the basement membrane zone. The development of lesions with atypical clinical features in patients with different forms of EB has occasionally been pointed out. In this lecture we are pointing out to different pigmentary lesions that may be encountered in EB patients, their mechanisms and implications.

Mona Mostafa Korany
A better life for EB patients

Epidermolysis bullosa is a very mutilating genetic disease. Hundreds of researches have been done in the field of improving the treatment strategies, yet improving quality of life for those patients is as important as medical treatment. Evidence- based research according to DEBRA recommendations states that psychosocial support with physiotherapy, nutritional care and wound care is important to enhance quality of life and preventing complications. wound care clinic in cairo university exerts a huge effort to give the proper wound care and dressings for EB patients.

Maha Abdelfattah

Effect of different concentrations of human platelet rich plasma contrasted with fetal bovine serum on periodontal ligament stem cells

Maha I Abdelfattah [1,2] and Heba M. Gamal El Din [1]

[1] Department of Oro-Dental Genetics, Medical Research Center of Excellency, National Research Centre, Cairo, Egypt.
[2] Stem Cell Laboratory, Center of Excellence for Advanced Sciences, National Research Centre, Cairo, Egypt.

Abstract

Periodontitis is a complex immune-inflammatory disease, characterized by the destruction of the periodontium. Periodontal regeneration implicates the use of mesenchymal stem cell populations for cell-based therapies. Periodontal ligament stem cells (PDLSCs) isolated from the PDL tissue of human teeth, are the most extensively studied and applied for regeneration of the periodontal tissues. Platelet rich plasma (PRP) is widely used for various tissue engineering applications, because of its several advantages over fetal bovine serum. Aim: To study the effect of different PRP concentrations contrasted with fetal bovine serum, on proliferation and osteogenic differentiation capability of PDLSCs. Methods: we isolated PLSC from four extracted sound wisdom teeth. Venous blood was drawn from healthy male and activated PRP was prepared. We had four experimental groups; 10% FBS, 5% PRP, 10% hPRP and 15% hPRP and we evaluated the proliferation and osteogenic differentiation capability for these groups. Results: Statistical analysis was done using a One-way analysis of variance (ANOVA). Conclusion: Our study showed that PRP can effectively induce hPDLC's proliferation as well as stimulate osteogenic differentiation of hPDLC's. The 10%hPRP had the highest proliferation rate than 5% hPRP, 15% hPRP as well as 10% FBS with statistical significance at 3 and 5 days' groups. The RT-PCR showed that osteopontin gene in 10% FBS expressed higher than all PRP groups with no statistical significance and the osteopontin gene expression in the 10% hPRP group was higher than the 5% PRP and 15% PRP with no statistical significance.

Liliana Virginia Ziadé

Global science, genetic diseases and rare diseases in social networks. Perspective from the ethics of responsibility. Leading case 2023-2024

Fundamentals

Today we live in a time of scientific and social development, says Manolo Morente, in which on the one hand the great scientific-technological advances and, on the other, we witness the phenomena of society characterized by g and diverse communication. Faced with this reality, we run the risk of moving forward without taking into accou possibilities offered by relating the most powerful axes in global

development, creating a bridge between bio-techno-scientific development and communication in new virtual spaces. Hence the importance of ethical reflection thinking about what type of science we want to promote. Science is understood in this paper as a process formed by three pillars that are the production of knowledge, its circulation and appropriation, represented in uses and products, which does not occur outside of society or global sociocultural processes. Thus, in this research the unspoken question arises of thinking about how the population participates in the science-health-patients, families, community process. In this case, in the field of social networks in light of the ethics of responsibility. Artificial intelligence, in this case, is used as a tool in social networks, applied through big data in listening to digital citizens on issues related to genetic diseases and rare diseases. This tool applied to social networks constitutes a necessary technological advance for science as an act and power, a possibility, if we imagine the opportunity to learn from the current opinion of digital citizenship that guides us in

implementing communication policies both atthe institutional level. as in health teams and the States themselves. Aim of the Study This article analyzes and reflects on the impact on social networks of the significance of genetic diseases and rare diseases from listening to opinions and perceptions of digital citizens who participate in X ex Twitter, and other networks that range from their concerns, expectations, emotions and feelings. The purpose is to access the opinions of the authors and identify their perspectives, how they conceive the topic of study, what actors and general problems they are associated with, and what future demands they express. Methods: Regarding the methodology applied in this research, we work with a quantitative-qualitative method that allows us to see the impact of the topic on social networks based on keywords, comparing the data obtained, such as main influencers, content with greater engagement, concerns and emotions. Results: The results of this work through the application of artificial intelligence and big data in social networks show the lack of information aimed at the population that promotes science, based on its impact on public opinion, which indicates the limited use of these technological tools in communication by scientific work teams. Conclusions: The application of these technologies, artificial intelligence and big data, in the new communication spaces adopted by society, have great potential for their development. In this case, it is observed that social networks can be very useful to promote information and education in society that generates an impact on public opinion such as generating greater awareness, participation not only of patients, families, and communities but also society in general.

Reham Doss

Genomic management of a polygenetic disorder

Doss R. W. [1]

[1] Dermatology Department, Faculty of Medicine, Beni-Suef University, Egypt.

Psoriasis is a chronic immunological skin disease. Genetics play important role in psoriasis pathogenesis. Nowadays, several biological and small molecule drugs are being developed for management of psoriasis. These therapies vary in efficacy from one person to another. Response rates to systemic treatments for moderate-to-severe psoriasis range from 35 to 80%. Pharmacogenetics is the study of variations in DNA sequence related to drug response. Numerous genetic variants (such as ABC transporter, DNMT3b, MTHFR, ANKLE1, IL-12B, IL-23R, MALT1, CDKAL1, IL17RA, IL1B, LY96, TLR2, etc.) were found to be associated with treatment response for methotrexate, cyclosporin, acitretin, anti-TNF, anti-IL-12/23, anti-IL-17, anti-PDE4 agent There is rising demands to identify biomarkers that could help predict treatment outcomes and individualize treatment for patients with psoriasis. Pharmacogenetics are being used to search for biomarkers that can predict response to systemic treatments. These biomarkers could improve patient quality of life and reduce health costs and potential side effects.

Keywords: Psoriasis, Pharmacogenetics, Pharmacogenomics

Pertunia Mutheiwana

Adapting the blended learning distributed classroom model for training multiple learner groups in genomic medicine

Mutheiwana P.[1], Fadlelmola F.[2], Nembwaware V.[3], Radouani F.[4], Mulder N1 on behalf of the african genomic medicine training initiative members[5]

1 Computational Biology Division, Department of Integrative Biomedical Sciences, Faculty of Health Sciences, University of Cape Town, Cape Town, South Africa
2 Kush Centre for Genomics and Biomedical Informatics, Biotechnology Perspectives Organization, Khartoum, Sudan
3 Division of Human Genetics, Department of Pathology, Faculty of Health Sciences, University of Cape Town, Cape Town, South Africa
4 Chlamydiae and Mycoplasmas Laboratory, Research Department, Institut Pasteur du Maroc, Casablanca, Morocco

5 List of members in the acknowledgement

The African Genomic Medicine Training Initiative (AGMT) was launched in May 2016, in Senegal, by a group of stakeholders from the Human Heredity and Health in Africa (H3Africa) Consortium and the African Society of Human Genetics (AfSHG). This initiative started as a response to the lack of Genetics and Genomics training for Health Care Professionals in Africa. The first two iterations of the AGMT's basic Genomic Medicine course ran in 2017 and 2019 with nurses as the targeted participants. Considering that Genomic Medicine, like most clinical practices, depends on many facets, including effective collaborative patient management workflows that promote quality patient care, the course was expanded in 2022 and 2023 to include additional groups of Health Care Professionals to strengthen collaboration towards patient management. This expansion was also due to the demand for Genomic Medicine training from various Health Care Professional groups as well as considering the potential impact that these groups are likely to have on the implementation of Genomic Medicine. This presentation showcases the blended learning distributed classroom model used to deliver Genomic Medicine training to Health Care Professionals in Africa and how this was adapted to train multiple learner groups in a single iteration. The presentation also provides lessons learnt during this process to inform and improve future implementation activities and highlights the major accomplishments of the training iterations. Training in Genomic Medicine is scarce and even when implemented, it rarely reflects the collaborative nature of patient management processes. The presented model showcases how training strategies could reflect the collaborative patient management processes currently implemented in the clinics. We conclude by outlining future work required to continue this initiative.

Keywords: Health care professionals, Introductory training, Genomic medicine, Collaboration, Africa

Day 2 - Friday 26th April – Advocacy

Session I: Patients (and communities) as partners in research

Panel session: The patient researcher: partnering with patients at various stages of therapeutic development and access

Sophie Strobl

BUR-EB - the importance of understanding the socio-economic burden of the disease

STROBL S.[1] on behalf of the BUR-EB study group

[1] Faculty of Psychology, Sigmund Freud University Vienna, Freudplatz 1, 1020 Vienna, Austria

Cost-of-illness (COI) studies play a crucial role in evaluating the socio-economic impact of diseases, guiding policy decisions and resource allocation. Chronic rare diseases like epidermolysis bullosa (EB), characterized by ongoing wound care and substantial medical costs, impose a significant financial burden on affected families and society. The "Social Previous cost-of-illness European study revealed a mean annual cost of €31,390 per EB patient across 8 EU countries, highlighting the financial strain experienced by affected families. However, there remains a lack of comprehensive longitudinal data on the economic and cost-related burden of EB. The BUR-EB study, conducted in 7 EU countries in close collaboration with local patient organizations (DEBRA), aims to fill this gap by shedding light on the socio-economic impact of EB on patients, caregivers, and society as a whole. Utilizing a societal perspective, the study will estimate resource use and cost of illness, encompassing direct healthcare and non- healthcare expenses, as well as labor productivity losses of patients and their caregivers and out-of-pocket expenses of the families. Through comparative analysis with data from a decade ago, the study seeks to identify shifts in the socio-economic burden of EB over time, uncovering trends and potential driving factors behind these changes. Ultimately, the BUR- EB study seeks to offer actionable insights at three distinct levels: the individual patient level, the national level, and the broader international/EU level, contributing to informed decision-making and improved support for EB patients and their families.

Keywords: Cost-of-illness, Socio-economic burden, Epidermolysis bullosa

Dimitra Kiritsi
Aristotle University of Thessaloniki, Greece

The COSEB initiative - developing a core outcome set for EB

Dedee Murrell
University of New South Wales, Australia

Incorporating patient-reported outcome measures (PROMs) as proof of treatment efficacy

Emeline Baillargeault
URGO Medical, France

Patient driven evidence generation for advocacy - the Brazil project

Session II: The patient advocate - a vital player in health care

Panel session: Collaborating with patients: building on lived experiences for improved health outcomes

Ritu Jain
DEBRA International, Singapore

The role of patient advocates in health care

Andreas Miller
DEBRA Germany, Germany

The voice of EB patients - experts in their own condition

Mehar Singh
DEBRA International, India

The voice of EB patients - experts in their own condition

Ignacia Fuentes
DEBRA Chile, Chile

Involving the patient voice in research - a research perspective

Kalsoom Begum
University Hospitals Birmingham, UK)

Involving the patient voice in health care - a clinical perspective

Session III: Increasing the reach of DEBRA and awareness of EB worldwide.

Panel session: Best practices and challenges of patient advocacy organisations (PAOs) - what can we learn from each other?

Zlatko Kopecki
DEBRA Australia, Australia

Priscila Keiko Matsumoto Martin
DEBRA Brazil, Brazil

Ryan Hultman
DEBRA Canada, Canada

Zlatko Orucevic
DEBRA Norge, Norway

Panel session: Forming a patient advocacy group - how to go about it and what makes one successful?

Faiza Ambereen
DEBRA Pakistan, Pakistan

Navigating Cultural Terrain: Establishing Our Group in a Male-Dominated Society

Vlasta Zmazek
DEBRA Croatia, Croatia

as the hub for expertise in the region

Hanaa El-Sadat
DEBRA Egypt, Egypt

as the hub for expertise in the region

Toni Roberts
DEBRA South Africa

"From patient to advocate"

Toni Roberts and Ira Jain
DEBRA South Africa, and DEBRA Singapore

Shaping the patient leaders of tomorrow - DEBRA International Youth Council

Megan Foster Flaherty
DEBRA International, UK

EB Awareness Week 2024 - an international campaign

Session IV: Creating contacts for life

Day 3 - Saturday 27th April - Research and Healthcare in EB

Session I: Updates in EB research I

Mohamed El-Darouti
New topical therapeutic options for the treatment of dystrophic epidermolysis bullosa

Systemic treatment should be combined with topical treatment to reach the best results in EB patients in this lecture we will discuss different modalities of topical treatment with a focus on the new lines of topical treatment

Ignacia Fuentes
Debra Chile, Chile

Research update on genetics of EB; cell, gene, and protein therapies

Zbigniew Ruszczal
Sheikh Khalifa Medical Centre, UAE

Epidermolysis Bullosa: The newest news

Dimitra Kirits
Aristotle University of Thessaloniki, Greece

Research update on symptom prevention and relief

Dedee Murell
University of New South Wales, Australia

Research update on the EB development pipeline

Session II: Updates in EB research II

Ignacia Fuentes
Evaluating ELK-003 Eye Drops for Ocular Manifestations in Dystrophic and Junctional Epidermolysis Bullosa Patients in Chile: A Pilot Study

Ignacia Fuentes, Arturo Kantor, Felipe Mellado, Belkis Noya, Victoria Prieto, Armen Karamanian, Francis Palisson

Fundación DEBRA Chile, Departamento de Biología Celular y Molecular, Facultad de Ciencias Biológicas, Pon(ficia Universidad, Católica de Chile, Centro de Gené(ca y Genómica, Facultad de Medicina Clínica Alemana, Universidad del Desarrollo

Aim of the Study: Dystrophic Epidermolysis Bullosa (DEB) and Junctional Epidermolysis Bullosa (JEB) commonly result in severe ocular complications, including frequent corneal abrasions that significantly increase the risk of childhood blindness. With current treatments being limited to supportive care, such as lubricants and artificial tears, there is a critical need for effective therapeutic options. This study aims to evaluate the safety and efficacy of ELK-003 eye drops, a standardized acellular amniotic fluid formulation, in alleviating ocular manifestations in DEB and JEB patients. Methods: This study employs an open-label, self-controlled design, allowing each participant to act as their own control. The study is structured into two distinct phases: an Observational Phase and a Treatment Phase. In the Observational Phase, we will establish each patient's baseline disease status, which will then be compared against outcomes observed during the Treatment Phase. Both phases are intended to span approximately 6 months and we aim to recruit around 30 participants. Evaluations will include assessments of ocular-related quality of life questionnaires, symptoms associated with corneal abrasions, the frequency and duration of corneal abrasions, and other ocular manifestations commonly encountered in this patient population. Results: The study, sponsored by DEBRA Chile, has received the approval of the Ethics Committee of the Universidad del Desarrollo, Clinical Alemana (Chile) and the clearance from the Instituto Publico de la Salud (Chile). The study is scheduled to commence in March/April 2024. Conclusions: This pilot study represents an important step towards addressing the unmet medical needs of DEB and JEB patients suffering from severe ocular manifestations. By evaluating the safety and efficacy of ELK-003 eye drops we aim to offer a potential therapeutic option beyond the mere supportive care. As the trial progresses, ongoing data analysis will provide a better understanding of the disease’s natural course and ELK-003’s potential therapeutic effects. Future larger-scale trials will be warranted to confirm findings and establish the broader applicability of this innovative treatment approach for EB patients.

Albert Mellick

An RNA marker of cancer in recessive dystrophic epidermolysis bullosa (RDEB)

Wang C. [1,2] , Chen K. [1,3] , Zauner R. [4] , Wally V. [4] , Hofbauer J.-P. [4] , Shah M.M. [1] , Zhou J. [1,2] , Zwicklhuber J. [4] , Ramos J.-S. [1,2] , Mann T. [1,3] , Sajinovic M. [1,5] , De souza P. [1,5] , &
Mellick A.S. [1]

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Aim: If they survive into young adulthood, patients with Recessive Dystrophic Epidermolysis Bullosa (RDEB) may develop an aggressive and often lethal form of nonmelanoma skin cancer. Early and effective detection, as well as timely intervention is the key to effective treatment. However, lack of well-defined biomarkers and the trauma associated with biopsy excision can make compliance to a regular screening regime, challenging. The aim of our work has been to identify biomarkers of malignancy in RDEB and apply them to the development of a minimally invasive diagnostic that will improve quality of life for patients. Method: To do this we have applied methods in multivariate in situ hybridisation (ISH) and filtration-based circulating tumour cell (CTC) isolation to identify novel biomarkers of aggressive squamous cell carcinoma (SCC), from RDEB, as well as non-RDEB patients. Results/Discussion: We have identified a novel set of molecular markers, small noncoding

microRNAs/miRNAs, which are linked to malignancy in SCCs, including miR-10b (Wimmer et al. Cell Comm. Signal, 2020). Notably, we have also demonstrated that miR-10b can be detected in CTCs isolated from the blood of patients, including those with RDEB. Conclusion: Given the previously demonstrated clinical significance of miR-10b (Plummer et al. Cancer Res. 2013; Gasch et al. Sci. Reports, 2015), and the fact that there are few validated biomarkers of malignancy in SCCs, this finding is significant; and is the basis of ongoing work to the develop a liquid biopsy-based diagnostic for early detection of cancer spread in 'at risk' patients

Keywords: Circulating tumour cells, Squamous Cell Carcinoma, microRNAs, In situ hybridization, Biomarkers

Julia Carnaz Benincasa

Gene therapy for restoring collagen VII expression in Recessive Dystrophic Epidermolysis Bullosa

Benincasa, J.C. , Rossetti, L.B. , Munhoz, A. , Gachet, C. , Borsoi, J , Torres, D.C., Martin, P.K.M. , Weinlich, R. 1

Albert Einstein Iraelite Hospital - Education and Research Institute, 05652-000 SãoPaulo, Brazil

Recessive Dystrophic Epidermolysis Bullosa (RDEB), a severe subtype of EB, stems from COL7A1 gene mutations, causing reduced or absent collagen VII (C7) expression vital for skin integrity. Brazilian RDEB patients often harbor exon 54 mutations, notably the c.5047C>T variant. Our project aims to develop gene-editing strategies to restore functional C7 expression in primary fibroblasts and keratinocytes, evaluating efficacy in human skin equivalents (HSEs). We explore exon 54 skipping and mutational correction techniques. Guide RNAs and base editors were cloned, amplified, and introduced into HEK293T cells via nucleofection, achieving a 10-22% transfection rate evidenced by GFP expression within 72 hours. Concurrently, HSE models from healthy and RDEB donors were established. Histological analysis of HSEs stained with hematoxylin and eosin revealed resemblance to human skin, featuring all epidermal sublayers. Next steps involve extending editing techniques to EB patients-derived fibroblasts and keratinocytes, alongside confirming histological findings and characterizing HSEs using immunofluorescence. Successful project completion promises a novel therapy prototype for RDEB, potentially enhancing wound healing and patients' quality of life.

Keywords: RDEB, Base Editing, Exon Skipping, c.5047C>T, 3D skin model

Hadeer Ibrahim

Do EB cells react differently from healthy keratinocytes to biofilm presence in EB wounds?

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The rupture of the blisters/bullae that characterize epidermolysis bullosa (EB) leads to open wounds, exposed to the microbial world, and which may be acute or become chronic in nature. Chronic wounds impact significantly on patients and healthcare systems. Biofilms, comprise microbial aggregates embedded in a self-produced matrix and play a crucial role in wound chronicity, adding diagnostic and management challenges to different medicalconditions such as diabetic ulcers, where biofilms are reported in 60% of cases. Our earlier work demonstrated, for the first time, that chronic EB wounds also exhibit biofilm presence. In this study we report an in-vitro model to investigate whether biofilms play a role in EB wound chronicity and whether EB cells react differently to biofilm presence, in comparison with healthy keratinocytes. Biofilm-conditioned medium (BCM) and planktonic bacteria- conditioned medium (PCM) were prepared using a co-culture of biofilm-forming bacteria commonly found in EB chronic wounds, namely, Staphylococcus aureus SA SH1000 and Pseudomonas aeruginosa PA01. Their effect on a primary immortalized EB cell line (KEB-7) and healthy keratinocytes (NEB-1) was investigated. Preliminary findings obtained by microscopy demonstrated that EB cells show remarkable changes in response to the presence of BCM, including cell detachment, cell death, rounding up, and membrane irregularities compared to healthy keratinocytes. In this study, we compare different inflammatory cytokines produced by EB and healthy keratinocyte cell lines in responses to BCM and PCM, using ELISAs. This study offers novel insights pertaining to the pathophysiology of EB wound chronicity despite meticulous medical care and may lead to changes to EB wound management focused upon biofilm disruption.

Keywords:
Epidermolysis bullosa, chronic wounds, biofilm

Zlatko Kopecki

Development of bacteria activated stimuli response dressing for management of infection in Epidermolysis Bullosa

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Aim: Large areas of open skin provide an inadequate barrier to protect against infection for patients with Epidermolysis Bullosa (EB). The success of current treatment is limited, and high cytotoxicity is a major side effect with currently available silver dressing. The aim of this project was to develop a and validate a stimuli-responsive silver nanoparticle (AgNP) dressing offering the on-demand release of silver ions triggered by changes in wound microenvironment. Method: Optimization and characterization of the hydrogel delivery system was achieved using cross-linking of N-isopropylacrylamide with acrylic acid and loading with ultrasmall AgNPs. Material characterization, biocompatibility and release studies were undertaken to demonstrate temperature and pH responsive properties and in-vitro efficacy against common wound pathogens from EB wounds. Demonstration of in-vivo antimicrobial safety and efficacy was achieved using a preclinical murine wound infection. Results / Discussion: We demonstrate that the dual-responsive hydrogel is highly sensitive to a typical pH and temperature changes during wound infection development, with restricted release of silver ions at acidic pH (<pH 5.5) and significant release in alkaline conditions (>pH 7.4) (>90% release). The pH dependent release and antimicrobial effect resulted in elimination of 95% of pathogens in-vitro at alkaline pH which was confirmed by clearing of S. aureus infection and significant improvement in healing using preclinical models including faster reepithelization and improved early collagen deposition. Conclusion: This multifunctional hydrogel presents a promising bacteria responsive delivery platform that serves as an on-demand carrier to not only reduce side effects but also boost the antibacterial efficiency based on physiological needs. It offers great potential to improve the way we manage wound infections in EB, providing a single platform for a long-lasting application in wound management.

Keywords: Wound Infection, Epidermolysis Bullosa, pH-responsive, Synergistic Therapy, Localized Delivery

Session III: Health care in EB

Sarah Elshakankiry

Keynote presentation: Supporting EB patients psychologically transitioning from pediatric to adult care

Malachite Institute for Behavioral Health, Egypt

Epidermolysis Bullosa (EB) presents unique challenges for patients, particularly during the transition from pediatric to adult care. This presentation explores the essential requirements for facilitating a smooth transition process for EB patients, ensuring continuity of care and improved quality of life. The transition from pediatric to adult care is a critical phase for EB patients, marked by numerous physical, emotional, and social changes. This presentation highlights the specific needs of EB patients during this transition period, focusing on medical, psychosocial, and practical considerations. Medical requirements include comprehensive healthcare planning, specialized multidisciplinary care teams, and continuity of treatment plans. Psychosocial support is vital, addressing emotional challenges, mental health needs, and empowering patients to self-manage their condition effectively. Practical aspects such as education, vocational training, and financial assistance also play a significant role in ensuring a successful transition. Drawing on current research, clinical guidelines, and patient experiences, this presentation provides practical recommendations and strategies for healthcare professionals, caregivers, and EB patients themselves to navigate the transition process effectively. By addressing the unique requirements of EB patients during this critical period, we can improve outcomes, enhance patient satisfaction, and promote long-

Keywords: Epidermolysis Bullosa, Psychological Care

Kalsoom Begum
University Hospitals Birmingham, UK

Keynote presentation: Clinical and social management - the role of the EB CNS

Sophie Kitzmüller

EB Clinet - an international network to support care and retain expertise

Toni Roberts

Consensus-based guidelines for the provision of palliative and end-of-life care for people living with epidermolysis bullosa

Popenhagen M. [1,2]* Genovese P. [3] Blishen M. [4] Rajapakse D. [5] Diem A [6] King A [7] Chan J [8] PelliceR Arasa E [9] Baird S [10,11] Ferreira DA Rocha A [12,13] Stitt G [14] BadgeR K [3] , Zmazek V [15,16] Ambreen F [17,18] Mackenzie C [19] Price H [3] Roberts T [20,21] Moore Z [22] Patton D [22] Murphy P [22] & mayre-chilton K [23,24]

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Aim of the Study
Inherited epidermolysis bullosa (EB) is a cluster of rare, genetic skin and mucosal fragility disorders with multi-system and secondary effects, in which blistering and erosions occur in response to friction/mechanical trauma. Considering the incurable and potentially life-limiting nature of the condition and the challenges posed by its symptoms, a palliative approach to EB-related care is necessary. However, knowledge and experience related to the provision of EB palliative care is minimal. Evidence-based, best care guidelines are needed to establish a base of knowledge for practitioners to prevent or ease suffering while improving comfort at all stages of the illness, not just the end of life. Methods This consensus guideline (CG) was begun at the request of DEBRA International, an international organization dedicated to improvement of care, research, and dissemination of knowledge for EB patients, and represents the work of an international panel of medical experts in palliative care and EB, people living with EB, and people who provide care for individuals living with EB. Following a rigorous, evidence-based guideline development process, the author panel identified six clinical outcomes based on the results of a survey of people living with EB, carers, and medical experts in the field, as well as an exhaustive andsystematic evaluation of literature. Recommendations for the best clinical provision of palliative care for people living with EB for each of the outcomes were reached through panel consensus of the available literature.

Keywords: Epidermolysis bullosa, Palliative care, End-of-life, Clinical practice guidelines, Consensus guidelines

Niraj Parajuli

Managing cases of Epidermolysis bullosa in a resource-limited setting: An experience from Nepal

Parajuli N.
Department of Dermatology & Venereology, National Academy of Medical Sciences, Bir Hospital, Kathmandu, Nepal
Article Info Abstract

Aim: To provide an insight on the management of EB from a resource-constraints setting. Method: A case series of five different cases of Epidermolysis bullosa will be presented. The difficulties in management will be presented. Case 1:EB case from a very remote village in the northern part of the country where it took the parents two days of hike and another 22 hour bus ride just to reach our center. Case 2: A case of EB simplex in a prisoner who was being treated as Bullous Pemphigoid for several years. Case 3: A Muslim parents with 5 kids and youngest one presented with EB, where the parents were totally confused on the disease and its course. Case 4: A case of Dystrophic EB who developed malignancy and a team of global experts united to provide their expertise through an online platform, however the ending was not what we expected. Case 5: A probable case of EB

diagnosed through telemedicine. Results / Discussion: A elaborate discussion on the way forward for taking care of EB patients in resource-limited setting will be presented. The role and use of EB diagnostic matrix will be highlighted. Conclusion: EB is a complex disease and has varied presentation. A lesson learnt from managing cases in Nepal is have a good clinical etiquette, make use of the available resources, and provide empathy to patients and family.

Keywords:
low-resource setting, epidermolysis bullosa, telemedicine

Elena Belonogova

Nutritional deficiency in patients with congenital epidermolysis bullosa

Relevance:
Congenital epidermolysis bullosa (CEB) is a group of genetically and clinically heterogeneous disorders characterized by a tendency to develop blisters and/or erosions on the skin and mucous membranes with minimal trauma. Clinical manifestations of CEB can vary in severity from mild to extremely severe, depending on the form of the disease. Patients with CEB often suffer from nutritional deficiencies caused by multiple factors. Aim of the study: to assess the nutritional status of children with simplex, junctional and dystrophic CEB, to calculate the correlation of the Birmingham Epidermolysis Bullosa Severity Score (BEBS) and diagnostic Z-score values of anthropometric indicators. Materials and methods: The study included 101 children aged 3 to 17 years with simplex (n=25), junctional (n=10) and dystrophic (n=66) forms of CEB. Assessment of physical development parameters was carried out using Z-score indices: weight-for-age (WAZ), height-for-age (HAZ), and BMI-for-age (BAZ). The BEBS scale was used to evaluate the severity of the disease. Results: Mild malnutrition was detected in 20% (n=5) of patients with simplex CEB, 40% (n=4) of patients with junctional CEB, and 22.7% (n=15) of patients with dystrophic CEB. Moderate malnutrition was observed in 12% (n=3) of patients with simplex CEB, 40% (n=4) with junctional CEB, and 18.8% (n=12) with dystrophic CEB. Severe malnutrition was detected in 30.3% (n=20) of patients with dystrophic CEB. Nutritional deficiency showed a significant reverse correlation with the BEBS score (p<0.005). Conclusions: The analysis shows obvious malnutrition in children with CEB, which are inversely correlated with the severity of the disease. Malnutrition can occur in different forms CEB.

Session IV: Health care in rare diseases

Nabeelah Peerbhai

The importance of genetic counselling in rare diseases and cultural challenges faced

Peerbhai N [1]
[1] University of Cape Town, Cape Town, South Africa

Genetic counsellors are healthcare professionals trained in medical genetics, counselling and communication skills. Genetic counselling is an evolving profession that offers support to individuals who have or are at risk of having a genetic condition. It plays an important role in the management and understanding of rare diseases, where comprehensive knowledge of genetic variation is essential for diagnosis, treatment and family planning. As rare diseases often have a genetic bases, tailored genetic counselling provides patients and their families with accurate information regarding inheritance patterns, prognosis and available resources. By facilitating informed decision-making, genetic counselling empowers individuals to navigate the uncertainties associated with rare diseases, fostering proactive healthcare management and enhancing patient outcomes. Genetic counsellors undergo additional training in cultural competency, acknowledging the impact of language barriers,

diverse beliefs, values, and cultural traditions on patient experiences. They recognize how cultural beliefs and practices may influence perceptions of genetic risks, treatment decisions and utilization of healthcare resources. Genetic counsellors are adept at employing culturally sensitive communication strategies to convey medical information effectively, aligning with patients' values and preferences to ensure they feel respected and understood. Addressing these cultural nuances are imperative to ensure effective communication and understanding between counsellors and patients from diverse backgrounds. Drawing from real-world examples, we discuss strategies for navigating cultural challenges and delivering culturally competent care. This includes respectfully acknowledging diverse perspectives, building trust and rapport in the counselling relationship and highlighting the need for collaborative efforts among genetic counsellors, healthcare providers, and community leaders to bridge cultural gaps and ensure fair access to genetic services.

Keywords: Genetic counselling, Rare diseases, Cultural competency, Healthcare management, Multidisciplinary care

Anica Ježić
Government of Croatia, Croatia

Long term care and holistic approaches to care for rare disease and EB patients

Day 4 - Sunday 28th April – Panel sessions

Panel session I: How various initiatives can be consolidated towards positive healthcare outcomes for those with rare disorders

Alexandra Heumber
RDI, Switzerland

Connecting the MENA region with global rare disease initiatives

Ethical engagement with pharmaceutical organisations

Shirlene Badger
Illumina, UK

Connecting diagnosis to care

Sophie Kitzmüller
EB Haus, Austria

The importance of knowledge retention, education, and connection in the healthcare professionals' community

Panel session II: Overcoming challenges to development and access to medicines and devices in the MENA region and internationally

Sherif Kamal
EHA board for medication management and pharmacy affairs

Value based Healthcare Integrated Model for Rare diseases

Nourhan Tahoun
Egyptian Drug Authority, Egypt

EDA Role to Ensure the Ease of Access and Availability of Medicines for Rare Diseases

Maha Nasr
Drug Research Council, Ain Shams University, Egypt

The National Consultancy Centre: A proposed initiative by the Drug Research Council to overcome challenges of pharmaceutical industry in Egypt

Drug is a strategic commodity related to the country's national security due to its direct impact on the health of the citizens and its important role in shaping up the economy that may affect the country's international decisions. Therefore, the development of the pharmaceutical sector in Egypt in order to provide a high-quality medication to citizens and to increase the competitiveness of the Egyptian pharmaceutical products to reach an advanced world ranking is an essential goal. As an initiative of the national drug research council in Egypt, we have developed three roadmaps till current date, the most recent one aimed at developing a complete vision to raise the quality of Egyptian medicine, improve accessibility of patients to medications, and overcome the challenges of pharmaceutical industry in Egypt through the establishment of an internationally accredited national center for pharmaceutical consultancy. Therefore, the presentation sheds the light on the stages of this initiative, as well as its outcome and future perspective.

Shaheer Bardissi
Minapharm pharmaceutical

Development and access of advanced biologics in emerging countries: a real case study

Panel session III: The patient journey and care approaches that meet the needs of patients and physicians

Carol Hlela
Red Cross Children's Hospital, South Africa

Managing EB with limited resources, funding, and care teams - a South African experience

Shaden Abdel Hadi
Sheikh Khalifa Medical City, Abu Dhabi, UAE

Establishing Epidermolysis Bullosa House in UAE: Experience and Aspirations

Rahul Mahajan
Postgraduate Institute of Medical Education and Research, India

Mutational analysis and management strategies in epidermolysis bullosa - an Indian experience

Khadija Sellami

Hedi Chaker Hospital, Tunisia

EB management updates - a MENA region experience

Session IV: Poster prizes and closing remarks

Poster Abstracts for the EB Congress

Clarissa Alves Gomes Bittencourt

Use of tlc-ag healing matrix in post-operative correction of pseudosyndactyly in a patient with recessive dystrophic epidermolysis bullosa - case report

Aim of the study: Describe the TLC-Ag healing matrix action in the postoperative of Pseudosyndactyly correction in a patient with Recessive Dystrophic Epidermolysis Bullosa (EB). One of the aggravations of the disease is pseudosyndactyly with the progressive loss of skin elasticity, the appearance of flexion contractures of the fingers or even friction between them which leads to ulcerations and adhesions. The TLC-Ag Healing Matrix is indicated for the local treatment of wounds that show signs or risk of infection. The dressing is composed of a non-occlusive polyester mesh impregnated with the TLC-Ag healing matrix.Methods: This is a clinical, descriptive, retrospective and interventional case report. The use of antimicrobial dressings on the postoperative wound after correction of pseudosyndactyly was observed. Monitoring was carried out during dressing changes in the surgical center every 7 days by the orthopedic hand surgery team at a Hospital in the Metropolitan Region of Campinas fromOctober 25, 2022 to January 10, 2023. Results: In this case, it was observed that it was easier to change the dressing due to non-adhesion, infection control and rapid healing. In addition to the surgical technique, it is extremely important to use adequate primary coverage. Conclusions: This study contributed to showing the safety and effectiveness of the TLC Ag Matrix when used in surgical lesions with a high risk of complications, as it optimized healing, promoted comfort and avoided an infectious process, reducing the costs of prolonged hospitalization resulting in quality of life for the patient. patient.

Dalia hasan ahmed abuauf

Beyond traditional cases of epidermolysis bullosa simplex A Rare case reported regarding interplay between epidermolysis bullosa simplex and aplasia cutis (Bart syndrome)

Inherited epidermolysis bullosa (EB) encompasses a number of disorders characterized by recurrent blister formation as the result of structural fragility within the skin and selected other tissues. All types and subtypes of EB are rare , one of them reported in our article due to plakophilin deficiency in interplay with aplasia cutis in a rare syndrome called Bart syndrome . Bart's syndrome is an uncommon inherited congenital disorder associating congenital cutaneous aplasia of the extremities (lack of skin) and inherited epidermolysis bullosa with nail anomalies. Bilateral and symmetrical involvement of the limbs is exceptionally described on black skin. In most cases, the diagnosis is clinical; however, the management remains very difficult, and the extended forms are a real therapeutic challenge.2 cases of Bart's syndrome observed in a sub-Saharan African country (Senegal, Dakar). Clinical manifestations range widely, from localized blistering of the hands and feet to generalized blistering of the skin and oral cavity, and injury to many internal organs. Each EB subtype is known to arise from mutations within the genes encoding for several different proteins, each of which is intimately involved in the maintenance of keratinocyte structural stability or adhesion of the keratinocyte to the underlying dermis. EB is best diagnosed, and sub classified by the collective findings obtained via detailed personal and family history, in concert with the results of immunofluorescence antigenic mapping, transmission electron microscopy, and in some cases, by DNA analysis. Optimal patient management requires a multidisciplinary approach and revolves around the protection of susceptible tissues against trauma, use of sophisticated wound care dressings, aggressive nutritional support, and early medical or surgical interventions to correct whenever possible the extra cutaneous complications. Prognosis varies considerably and is based on both EB subtype and the overall health of the patient.

Keywords: Bart's syndrome, congenital absence of skin, epidermolysis bullosa.

Darya Drozdovskaya

Our experience of cutaneous squamous cell carcinoma treatment in patients with the dystrophic form of epidermolysis bullosa.

Drozdovskaya D.A., Zinoviev G.V., Ebert M.A. Anokhina E.M., Pleshkov A.S., Kornev V.I., Sabitova A.A.

Introduction:Cutaneous squamous cell carcinoma (cSCC) is the leading cause of death in patients with autosomal recessive dystrophic epidermolysis bullosa (RDEB). Objective: to demonstrate our experience in treatment of SCSC in patients with RDEB Materials and methods: from November 2019 to February 2024 a total of 11 patients (6 male and 5 female) with previously diagnosed RDEB developed cSCC. The tumor was localized at the extremities in 73%, and in the torso in 27% of cases. The cSCC was multifocal in 73% and presented by single focus in 27% of cases. All cases were confirmed by morphology. Radical surgery was performed in 70% and palliative in 10% of cases, accordingly. One patient (10%) refused surgical treatment and two patients (20%) had inoperable tumour due to locally advanced disease. Three patients (30%) underwent systemic immunotherapy (EGFR inhibitors, PD1 inhibitors). Results: two-year overall and event-free survival rates were 75% and 31%, respectively Conclusions: 1. The gold standard for treatment of cSCC in patients with RDEB is radical surgery 2. Chronic skin inflammation in patients with RDEB is the cause of metachronous cSCC 3. Experience with the use of systemic therapy for cSCC in patients with RDEB is limited and requires further study

Elena Belonogova

Epidermolysis bullosa and osteogenesis imperfecta

Elena G. Belonogova[1,2], Sofia A. Malakhova[1,2]

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Epidermolysis bullosa (EB) and osteogenesis imperfecta (OI) are clearly rare pathologies characterized by physical diseases, high financial costs and restrictions in the daily life of patients. The EB group is a genetic disease characterized by increased sensitivity to injury to the skin and mucous membranes, which leads to the formation of blisters and ulcers. OI, in turn, is a rare inherited disease that causes brittle bones and an increased susceptibility to fractures. At first glance, the word “frailty” can “combine” only two random diseases; in EB this means skin fragility, and in OI it means bone fragility. Patients and methods. Two cases: a 5-year-old boy and a 13-year-old girl, patients with EB and registered with the Butterfly Children Foundation, Russia. It was established that skin lesions were detected at birth in both children. The boy was first diagnosed with EB at the age of 3, and the girl was diagnosed at birth. In both cases, bone fractures and joint contractures were noted in the first year of life. FKBP10 variants were examined using a next-generation sequencing (NGS)-based gene panel test. Results. In both patients, pathogenic variants were identified in 2 genes: in exon 3 of the KRTI4 gene (rs60725382) in a homozygous state, previously described in patients with the simple type of EB. And the pathogenic variant of the FKBP10 gene c.321_353del (p.Met107_Leu117del) in a homozygous state, which leads to the development of OI and is described in combination with EB simplex. Conclusions. We would like to show that two rare diseases can occur in one child. Important to see comorbidities and early initiation of therapy to prevent fractures, reduce long bone deformities, and improve quality of life by reducing pain.

Elena Belonogova

Clinical case: epidermolysis bullosa simplex and autism spectrum disorders

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Introduction and Purpose
Epidermolysis bullosa (EB) can coexist with other conditions and developmental disorders. It often associated with significant morbidity in infancy and early childhood. EB is often combined with other serious diseases, both somatic and mental. The cost of psychiatric care for children with various forms of EB is high. EB may be associated with attention deficit hyperactivity disorder (ADHD) and autism spectrum disorder (ASD). Because the autistic phenotype is very heterogeneous, and children with impairments along these three dimensions may exhibit very different behaviors along the continuum, diagnosis is sometimes problematic.Patient and methods: A 6-year-old girl was diagnosed with a simple type of EB. The patient does not speak. For a long time, doctors believed that the lack of speech was associated with blisters on the mucous membrane of the mouth, and the inability to walk was associated with the development of blisters on the feet and pain when standing on them. Psychiatric assessment was performed using the Autism Diagnostic Observation Schedule (ADOS). Whole exome sequencing was performed. Results: The patient has the following clinical symptoms: impaired speech and language, impaired communication and lack of social skills, stereotypic movements, little interest in toys, does not respond to names, selectivity in food. An audiological study was carried out - no hearing impairments were detected. A consultation with a psychiatrist was carried out, and a diagnosis of ASD was established according to DSM-5 criteria. Whole exome sequencing data: detected in exon 1 of the KRTI4 gene (rs28928893) in a heterozygous state, previously described in patients with the simple type of EB. A pathogenic variant of CACNA1S and KCNQ1 has been discovered that leads to channelopathies. These mutations have been described in the literature in children with autism spectrum disorders. Conclusions: It has been found that EB may have comorbidities such as a developmental disorder. Until now, there is little awareness about the disease EB, and accordingly, concomitant pathology is hidden as a manifestation of the underlying disease, that is, as EB.

Eman AbdelAlim Rabie

Xeroderma Pigmentosum: Molecular diagnosis impacts the rare genodermatoses landscape

Background: Xeroderma Pigmentosum (XP) is a rare autosomal recessive genodermatoses caused by loss of function variants in the genes encoding the proteins governing the DNA's nucleotide excision repair. XP manifests as dry atrophic freckle-like pigmentation of the skin with photosensitivity and photophobia. UV-induced basal and squamous cell carcinomas reach 10,000-fold increase in XP patients, in addition to higher risk for ocular, oral and internal cancers. Death eventually occurs from complications of different carcinomas or from neurodegeneration (affects 25% of cases). In 1964, 50 Xeroderma Pigmentosum cases were reported in Egypt, the highest reported number of cases in one country at that time. Twenty years later, Egypt harbored 11% of XP cases identified worldwide. In the largest recorded Egyptian study of 660,280 children, the frequency of genodermatoses reached 2% of genetic disorders, of which 16% were XP. XP was molecularly diagnosed via complementation tests on patient' skin fibroblasts, however, no genetic causative variants were identified for Egyptian families. Aim: Our Genodermatoses clinic launched the largest study to identify the mutation and clinical spectra of Egyptian XP patients. Methods: Thorough clinical investigation of 55 XP patients was employed followed by mutation detection via Sanger sequencing and whole exome sequencing. Results: We identified four XPA, eleven XPC, and one XPG disease-causing variants in 54 patients of variable clinical presentation. Conclusions: We present clinical and molecular findings of recurrent and novel disease-causing

variants consistent with North African countries with emphasis on population structure, ancestral origins and migration flows. Molecular diagnosis evidently guided subsequent clinical management and genetic counselling.

Hagar El Sayed

Assessment of growth hormone and insulin-like growth factor 1 in children with epidermolysis bullosa dystrophica

Introduction and background: Epidermolysis bullosa dystrophica (EBD) is characterized by muco-cutaneous fragility with blistering, scarring and severe growth retardation attributed to many factors. Materials and methods: This cross-sectional study included 51 patients aged 1-12 years with EBD. Weight and height were measured, with the calculation of weight standard deviation score (SDS), height SDS, and body mass index (BMI), followed by plotting them on Egyptian growth curves. Serum levels of basal growth hormone (GH), insulin like growth factor 1 (IGF-1), hemoglobin (HB) level, erythrocyte sedimentation rate (ESR), and thyroid functions (TSH and T4) were measured. Growth hormone stimulation test was performed in 10 patients. Results: Weight SDS and height SDS were significantly lower than normal measurements (P< 0.05*). Growth hormone, growth hormone stimulation, and IGF-1 were significantly lower than the normal range (P< 0.05*). HB levels were significantly lower than normal, while ESR levels were significantly elevated (P <0.001*). A negative correlation was found between ESR and basal GH, and a positive correlation between ESR and IGF1. Conclusion and recommendation: Children with generalized DEB have poor growth and low circulating GH and IGF-1 levels, likely due to malnutrition, anemia, and inflammation that suppresses GH/IGF-1 axis. Future treatments targeting the correction of GH and IGF1 levels and anti-inflammatory treatment should be considered.

Hoda Abdallah

Whole exome sequencing of epidermolysis bullosa in egyptian pedigrees; identification of forty- one novel mutations

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Epidermolysis bullosa is one of approximately 16 different genetic conditions of blistering disorders identified by skin fragility and mechanically induced blistering on the skin and mucous membranes. Blistering diseases are a rare group of disorders that can often be debilitating and fatal. EB can be classified into 4 main types and more than 30 subtypes based on the level of skin cleavage, as well as clinical and molecular features. There are at least 21 genes associated with EB causing different forms of the disease. The aim of our study is to assess the clinical and molecular profiles of EB in Egypt, a highly consanguineous population from North Africa. 100 EB patients from 140 unrelated consanguineous pedigrees were clinically diagnosed based on detailed dermatological and histopathological finding. Whole-exome sequencing was performed for denitrification of pathogenic variants in EB using genomic DNA from each case of EB. Potentially pathogenic mutations were subsequently confirmed by Sanger sequencing. Pathogenicity of variants was assessed ac cording to American College of Medical Genetics (ACMG) guidelines as well as bioinformatic tools (e.g., Polyphen, SIFT, and Alamut). WES was successful in finding the causative mutations in all patients with all mutations occurring in known EB genes (COL7A1,LAMA3, LAMB3,COL17A1,KRT5,DST,DSP,PLEC, FERMT1 and EXPH5). We identified 60 pathogenic mutations: 41 mutations were novel and 19 have been previously reported. COL7A1 mutations were found in 72 patients being the most common cause of EB in Egypt. Conclusion: This is the first study to establish the EB genetic profile in Egypt using whole exome sequencing. COL7A1 is the most common gene contributing to 72% of the identified Egyptian EB genetic spectrum. Our results significantly expand the mutation database in this disease and points to a heterogeneous and different genetic makeup of Egyptian patients

Keywords: Epidermolysis bullosa Genetic Whole-exome sequencing

Inas S. M. Sayed
Clinical overlap between NECTIN1 and NECTIN4 related Ectodermal dysplasia

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Pathogenic variants in NECTIN1 and NECTIN4 genes cause cleft lip/palate-ectodermal dysplasia syndrome (CLPED1) (OMIM# 225060) and Ectodermal dysplasia-syndactyly syndrome-1 (EDSS1) (OMIM # 613573) respectively. Both syndromes are recessively inherited Ectodermal dysplasia and share common clinical features including hypotrichosis, tooth agenesis and soft tissue syndactyly; On the other hand, CLPED1 is characterized by the presence of cleft lip/palate. Aim of the study: Here, we report an Egyptian female patient from a consanguineous marriage presenting with dental anomalies, hypohidrosis, sparse hair, syndactyly (affecting middle and ring fingers in the left hand and lateral four toes bilaterally) and exhibiting unilateral groove between the philtrum and left lateral part of the upper lip resembling repaired cleft lip. The latter feature may represent mild form of cleft lip. Methods: Complete clinical examination and Whole exome sequencing (WES) were done Results: WES revealed a novel homozygous pathogenic variant in NECTIN4 (NM_030916.3:c.880C>T:R294*) and not NECTIN1. Other clinical features were dry skin, low set cupped ears and bilateral hand clinodactyly, and fetal pads. Orodental examination revealed hypodontia, microdontia, spacing and tapered upper permanent central incisors. Conclusion: The defective lip fusion and the presence of ear anomalies are typical features of CLPED1 but not EDSS1. This expands the clinical spectrum of NECTIN4 related disorder and display the overlap between NECTIN1 and NECTIN4 related phenotypes.

MennatAllah Ismail Mehrez
Oro-dental features of hereditary epidermolysis bullosa spectrum

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Aim of the study: Hereditary epidermolysis bullosa (HEB) is a disorder of genetic heterogeneity caused by over 15 genes. These genes are responsible for mucosal and skin integrity, particularly the dermal-epidermal junctional integrity. It is classified into four clinical types; junctional, dystrophic, simplex and Kindler syndrome. The disorder causes skin fragility that result in bloody blisters on minor trauma that heal by ulceration and scarring. It also affects the gastrointestinal tract starting by the oral mucosa. In 2021, it was placed in the consensus report of the WHO for oral cancer as an oral potentially malignant disorder. This study reports the oro-dental features of HEB spectrum over a period of one year. Materials and Methods Patients were recruited from the genodermatoses and oro-dental clinics of the National Research Centre, Egypt over a period of one year. Parental consents to take part in the study were obtained according to the declaration of Helsinki. Demographic data were collected and pedigrees were charted and analyzed. A comprehensive oro-dental examination was then performed. Results The study included 15 cases. Eleven males and 4 females. Their ages ranged from 1 month to 23 years. Consanguinity was present in 11 cases. One case showed dominant inheritance with the mother and child affected. Three cases were Kindler syndrome two of which were siblings. Oro-dental features of the 12 HEB cases included; long philtrum (8/12), thin upper lip (6/12), enamel affection (6/12). Bad oral hygiene was present in 9/12 patients with rampant decay of their teeth. Lesions were present in 9/12 cases.

They were more common on the tongue, followed by the palate and buccal mucosa. Most of the cases over 1year of age had tightness of the lips, tongue restrictions with one case completely adhering to the floor of the mouth (ankyloglossia). The 3 cases with no oral lesions were epidermolysis bullosa simplex. As for the Kindler syndrome cases 3/3 showed gingivitis which is the early sign of desquamative gingivitis. common in these cases. They were normal dentally. One of the cases had tongue hemangioma. Conclusion Hereditary epidermolysis bullosa causes restriction of the oro-dental function as patients progress in age which impacts their quality of life as well as provision of dental medical care. Their inability to maintain optimum oral hygiene leads to teeth decay in addition to the structural defect in the enamel shown in the study at hand. Finally, long philtrum and thin upper lip might be facial features of the disorder.

Nehal F. Hassib

De novo TP63 variants in four unrelated Egyptian patients with failure in the repaired palatal clefts

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Tp63 gene is a regulatory gene for the proliferation, differentiation, and apoptosis of progenitor epidermal cells. The mutant gene generates a wide spectrum of multiple abnormalities conjugated with seven syndromes with variable phenotypes such as Ankyloblepharon-ectodermal defects-cleft lip/palate syndrome (AEC), Ectrodactyly-ectodermal dysplasia-cleft lip/palate syndrome 3 EEC3), Limb-mammary syndrome (LMS), Acro-dermo-ungual-lacrimal-tooth syndrome (ADULT), Rapp–Hodgkin syndrome (RHS), Split-hand/foot malformation 4 (SHFM4), and Orofacial cleft 8. The mutational variant in TP63 affects features which include ectoderm, limb, and oral clefts. We present four unrelated patients suffered of full phenotypic symptoms having palatal clefts, hypodontia, and ectodermal features. Accordingly, a questionable failure of repaired palatal cleft is of high interest. The WES was performed for the affected patients to detect the causative variants and Sanger segregation of the parents. Heterozygous TP63 mutations were identified in the four patients. Three different missense variants were identified c.1027C>T (p.Arg34Trp), c.1727T>C (p.Iso576Thr) and c.605A>C (Tyr202Ser). Of them, the c.1027C>T Het. (p.Arg34Trp) was recurrent in two patients. All variants were confirmed to be “de novo” in patients. We observed incompletely repaired hard palate in our four affected children denoting a serious complication correlated with the defective gene. Correspondingly, understanding the pathophysiology of the TP63 gene may help in future genomics repair to achieve a better outcome after surgical intervention.

Keywords: Tp63, EEC, AEC, Rapp-Hodgkin, palatal cleft

Nesrin Sabry Gomaa

EB lessons from our clinic- Tanta University

This is a retrospective study of cases presented with epidermolysis bullosa to our pediatric dermatology clinic in Tanta University. Patients were evaluated phenotypically and categorized in the spectrum of the disease. Complications and treatment outcomes were reported. A structured registry was designed to include all relevant items for the purpose of documentation of enrolled patients’ data as well as future patients. The study is essential

to outline the real size of the disease, improve the physician skills, gain more experience and better outcomes for the patients.

Nesrin Sabry Gomaa

Ectodermal dysplasia-skin fragility syndrome: molecular and clinical study

Back ground:Plakophilin-1 (PKP1) is a specific component of desmosomes which are essential intracellular structures for cell adhesion and skin integrity. PKP1 mutations result in ectodermal dysplasia-skin fragility (EDSF) syndrome . Results: screening of PKP1 identified new homozygous frameshift mutations in two patients: c.409_410insAC (p.Thr137Thrfs*61) and c.1213delA (p.Arg411Glufs*22). Comprehensive analyses were performed for the 18 cases with confirmed bi-allelic PKP1 gene mutations. Skin fragility and nail affection were present in all affected individuals (18/18), palmoplantar keratoderma (16/18), alopecia/hypotrichosis (16/18) and perioralfissuring/cheilitis (12/15). Conclusion: These results expanded both the molecular basis and clinical manifestations of EDSF as one of the desmosomal disorders.

Olga Orlova

Bone mineral density as a sign of nutritional deficiency in patients with epidermolysis bullosa

Due to complications in severe forms of CEB, nutritional insufficiency of multifactorial origin develops, which leads to growth retardation and full development, as well as to the development of osteopenia and osteoporosis. aim of the study: to evaluate the relationship between the presence and degree of nutritional deficiency and bone mineral density (BMD) in different forms of congenital epidermolysis bullosa (CEB). methods: The study involved 55 patients with simple (n=19), junctional (n=5), and dystrophic (n=31) CEB. The mean age of the patients was 11 ± 3.53 years. Physical growth was assessed using the WHO Anthro Plus program. BMD was determined by X-ray densitometry (DXA) with Z-criterion assessment. results: According to the results of osteodensitometry, all patients with CEB simplex had normal BMD values. Two patients (40%) had chronic nutritional deficiencies and three patients (60%) had mild nutritional deficiencies while
assessing anthropometric parameters in patients with junctional CEB. 3 patients (60%) was revealed to have decreased BMD. In patients with dystrophic CEB 11 patients (35,4%) had severe nutritional insufficiency, 5 patients (16,1%) had moderate nutritional insufficiency, and 5 patients (16,1%) had mild nutritional insufficiency. According to osteodensitometry data, low BMD values were revealed in patients with dystrophic CEB. conclusions: Nutritional status disorders are more common in dystrophic and junctional CEB. Protein and energy deficiencies and nutrient deficiencies are among the causes of bone resorption in CEB. To reduce the risk of osteopenia and
osteoporosis, control of anthropometric parameters, administration of nutritional mixes and vitamins for preventive and therapeutic purposes are necessary.

Priscila Keiko Matsumoto Martin

Genetic profile of EB in Brazil: EXOMA 400 Program

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Epidermolysis bullosa (EB) comprises a group of rare genetic disorders affecting the skin and mucous membranes due to mutations in at least 20 genes. These mutations disrupt skin structure and function, resulting in blister formation. Diagnosis involves genetic testing of 25 genes, crucial for distinguishing EB from similar conditions, yet hindered by high costs and limited accessibility. Aim of the study: In a groundbreaking effort from 2021 to 2023, Debra Brazil partnered with Illumina and Mendelics to sequence DNA from 400 EB patients nationwide. Methods and results: Buccal swabs provided the samples, analyzed using Illumina's platform for Exoma, revealing a significant discordance (47%) between clinical and molecular diagnoses across EB

subtypes (Junctional, Dominant or Recessive Dystrophic, Simplex or Kindler), showcasing disease phenotypic variability. Recessive Dystrophic EB proved most prevalent (55%), followed by Dominant Simplex EB (15%), Dominant Dystrophic EB (12%), and Junctional EB (8%). Notably, mutations in KRT5 and KRT14 genes predominated in SEB cases (72%), while COL17A1 and COL7A1 genes exhibited significant variants, including c.6527_6528insC and c.5047C>T, notably prevalent in DEB. Two cases of KEB were identified, while 11 cases remained undetermined due to variant ambiguity or lack of EB-related variants. The families received assistance in interpreting the report and received genetic counseling when requested, leading to improved clinical management following this study. Conclusion: This study underscores the critical role of genetic diagnosis in EB within Brazil, essential for optimal clinical management and
future therapeutic advancements.

Rana A. Youness

A Non-coding RNA Signature as Novel Diagnostic & Prognostic Biomarker in Young Egyptian Breast Cancer Patients

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Background: Breast cancer continues to be a highly complex disease despite extensive research efforts. It is the most commonly diagnosed cancer among young women and a leading cause of cancer-related deaths worldwide. Unfortunately, young women with breast cancer are often diagnosed at later stages and have more aggressive types of the disease. Non-coding RNAs (ncRNAs) have emerged as a significant area of interest in cancer research. These ncRNAs play a role in regulating gene expression and hold potential as diagnostic and prognostic markers for cancer. Moreover, non-coding RNAs offer advantages over traditional biomarkers as they provide greater accuracy and are less invasive in nature. Aim: The aim was to investigate the expression levels of the ncRNAs (miR-744, miR-4317, miR-96 and MIAT and HOTAIR), and their significant correlation with the parameters (tumor grade, lymphnode metastasis, molecular subtypes, tumor size, Ki-67, and age) to determine their potential role in breast cancer and their reliability as diagnostic and prognostic biological markers. Methods: Female BC patients (n=30) were recruited. Total RNA was extracted using Qiazol, reverse transcribed into cDNA, and expression levels of miR-744, miR-4317, and MIAT were quantified using qRT-PCR. Results: MiR-744, miR-96, HOTAIR and MIAT were found to be overexpressed in BC tissues compared to their normal counterparts. While miR-4317 was found less expressed in BC tissues compared to their normal counterparts. Upon patients' stratification, a significant high expression for HOTAIR in patients expressing high levels of Ki-67. Nonetheless, miR-744 and miR-96 expression levels were found to be associated with a high tumor grade, positive lymphnode metastasis, and younger age, and miR-4317 expression levels were found to be associated with TNBC subtype. Conclusion: This study sheds light on the significant involvement of miR-744, miR-96, miR-4317, HOTAIR and MIAT in BC. Moreover, it casts these ncRNAs as potential diagnostic and prognostic biomarkers for Egyptian Young BC patients.

Serge Bohbot

Using a novel contact layer for the management of epidermolysis bullosa skin lesions

MD, BOHBOT S

Aim: Epidermolysis bullosa (EB) is a heterogeneous group of rare, inherited skin diseases
characterized by recurrent painful skin lesions, often precipitated by minor trauma resulting in dermal-epidermal separation or split. Treatment for this family of 23 genetic skin disorders is mainly supportive as there is as yet no radical and effective therapy for them. The objective of this study was to evaluate the acceptability, tolerance

and efficacy of a novel contact layer dressing* in the management of epidermolysis bullosa (EB) skin lesions. Methods: This was an open-label clinical trial involving 20 patients (11 adults and nine children) suffering from EB simplex or dystrophic EB. Patients were selected from the register of EB patients at the investigating center and included subjects who presented with at least one skin lesion. Lesions were treated with the study dressing for a maximum of four weeks. All dressing changes, wound parameters, pain and effect on quality of life were recorded. Results: All the patients completed the trial. Nineteen out of 20 wounds healed within 8.7 +/- 8.5 days. Overall, 11 patients (55%) considered that their quality of life had improved following use of the dressing, which was also reported to 'very easy' or 'easy' to remove at most dressing changes. Nineteen out of 20 patients stated that they would use the study dressing to manage their lesions in the future. Conclusion: This study confirmed the very good acceptability and efficacy of UrgoTul contact layer dressing in the treatment of skin lesions in patients with EB.

Serge Bohbot

Lipido-colloid dressing associated with absorbent and protective pad: a therapeutic alternative in the management of epidermolysis bullosa lesions

Epidermolysis Bullosa (EB) is a heterogeneous and congenital disease characterised by recurring painful skin lesions, often precipitated by minor trauma, resulting in dermato-epidermal separation or split. A clinical study published in 2002 by Dr Blanchet-Bardon concerning 20 children and more than 200 dressing changes demonstrated the value of lipido-colloid interface (TLC) in the treatment of these lesions. The healing time was 8.7 days on average, but the most marked benefit was the pain reduction. 95% of the children (or parents) would like to treat their future lesions with this dressing. Methods: Here we report on a clinical trial with lipido-colloid interface with a protective pad in the management of these lesions located on flat zones. For lesions located in folds or on joint areas, the lipido-colloid contact layer is more flexible and therefore suitable for these kinds of wounds. Results: The results obtained for this lipido-colloid dressing with non-woven pad
were comparable to those for lipido-colloid dressing in terms of time healing and the quality of life improvement of these children and their families. The additional advantage offered by the contact layer associated with pad is easier and more rapid dressing changes by the family: it is no longer necessary to cover the dressing with a secondary dressing; the dressing is simply fixed in place with a bandage. Conclusion: The lipido-colloid interface combined with a protective pad can therefore be included in the extensive range of dressings used to cover EB lesions on flat areas.

Serge Bohbot

Management of epidermolysis bullosa with a non-adherent contact layer with technology lipido-colloid (tlc)

Aim: Epidermolysis bullosa (EB) is a heterogeneous group of rare, inherited skin diseases characterized by recurrent painful skin lesions, often induced by minor trauma resulting in dermal-epidermal separation or split. Approximately 1000 patients suffer from EB in Australia and this pathology has important implications on the psychological, physical and social well being of the child and the family. The local management of these lesions requires a non adhesive and non-adherent dressing, to prevent pain at removal and to help improve the quality of life of these patients. Methods: This case study presents the case of two young Australian patients suffering from non Herlitz junctional epidermolysis bullosa, and the treatment with a non-adherent contact layer with Lipido-Colloid Technology (TLC). Results: These cases confirm the efficacy of this non-adherent contact layer with TLC in the treatment of EB. Conclusions: The non-adherent contact layer with TLC brings management in line with best practice in the treatment of EB, providing further choice for wound management.

Serge Bohbot
Value of a new interface in the local treatment of hereditary epidermolysis bullosa lesions

Bohbot S.

Aim: Hereditary Epidermolysis Bullosa (HEB) is a group of genetic diseases caused by a defect in cohesion between epidermis and dermis. It results in skin separation and blistering, occurring spontaneously or following mild trauma. In 2000, in a clinical case study conducted on 20 patients with HEB lesions, a lipido-colloid interface combining carboxymethylcellulose and petrolatum on a polyester mesh demonstrated excellent acceptability (painless removal), along with a satisfactory healing rate with an improvement in patients' quality of life. However, the location of certain lesions does not always enable optimum application of this interface. Since April 2009, a new, more conformable lipido-colloid interface has been available. The flexibility of this new mesh is designed to improve the dressing's conformability to the wound, especially in HEB patients, whose wounds may be located in awkward anatomical areas. Methods: The authors report the results of their experiences with this new interface, through clinical case studies on children with dystrophic HEB. The lesions treated were followed up by the physician until they had healed (clinical, planimetric and photographic assessment). Results: In addition to complete re-epithelialisation of the lesions, dressing changes proved to be painless, thereby improving patients' quality of life. Conclusions: These clinical case studies demonstrate the good tolerance and excellent conformability of this new interface, along with its efficacy, justifying its availability to health professionals for use in this indication. The painless character of daily dressing changes with this new interface improves patients' quality of life and makes nursing procedures easier.

Solwan I. El-Samanoudy
IL-10 rs1 800871 genotype may increase the susceptibility to increased body mass index in patients with psoriasis vulgaris: A case control study

Background: IL10 is an anti-inflammatory cytokine, the deficiency of which was reported in patients with psoriasis. Aim of this study: is to evaluate a possible association between 3 single -nucleotide polymorphisms (SNPs) of the IL10 gene cluster: -1082 G/A (rs1 800896), -819 C/T (rs1 800871), and -592 C/A (rs1 800872) and the risk of developing psoriasis. A possible association between the above-mentioned SNPs and psoriasis area and severity index (PASI), extent of body surface area involved (BSA) and body mass index (BMI) among Egyptian patients with psoriasis vulgaris will be evaluated. Methods: Blood samples of 200 patients with psoriasis vulgaris and 100 age and sex matched healthy controls were screened for the three SNPs PCR (Real Time Polymerase Chain Reaction) method. PASI, BSA and BMI were evaluated. Results: There was a significant association between the mutant homozygous genotype of -819 C/T (rs1 800871) and psoriasis (p value <0.001, OR=3.8, 95% CI=1.4-10.2). Other genotypes showed no significant association with psoriasis. -819 C/T was also significantly associated with increased BMI (p value = 0.008). Conclusion: This study suggests a possible association between IL10 -819C/T polymorphism and increased body mass index in patients with psoriasis vulgaris.

Keywords: Psoriasis, IL10, Polymorphism, BMI

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