

# CONGRESS PROGRAMME



## PROGRAMME AT A GLANCE

Monday 20 January 2020	Tuesday 21 January 2020	Wednesday 22 January 2020	Thursday 23 January 2020 <i>EB Community ticket holders may only attend this day</i>
DAY 1	DAY 2	DAY 3	DAY 4
Welcome by Jouni Uitto	Welcome by Jemima Mellerio	Current dilemmas – <i>Funding challenges</i>	A global approach to EB
Classification	Cell manipulation & therapies	Update on clinical management strategies	Updates in EB research
Current dilemmas – <i>The complexity of EB</i>	Gene manipulation & therapies	Clinical & management case histories	Current dilemmas – <i>Patients in clinical trials</i>
The challenges of EB skin	Clinical trials & research programmes	Quality of life (pain & itch)	Living with EB
Inflammation, fibrosis, & therapeutics	Current dilemmas – <i>Measurement &amp; research</i>	Support models	Updates in EB clinical care
Cancer & cancer therapeutics		Strategies for care – status update	EB Community open forum
Therapeutics			
Natural history – implications for clinical trial design			

# CONGRESS PROGRAMME



**MONDAY, 20 JANUARY 2020**

TIME	PLENARY SESSIONS – DAY 1	SPEAKER
08:30	<b>WELCOME</b>	<b>Chairs:</b> Jemima Mellerio & Jouni Uitto
	Challenges and unmet needs – the breadth of preclinical work ongoing	Jouni Uitto
08:40	<b>CLASSIFICATION</b>	<b>Chair:</b> Jo-David Fine
	EB classification and updates from the 2019 classification meeting	Cristina Has
08:55	<b>CURRENT DILEMMAS – A CONGRESS DEBATE</b> <i>The complexity of EB</i>	<b>Chair:</b> Jo-David Fine
	EB as a systemic disease – implications for research	Johann Bauer
	Modelling EB in the research environment	Leena Bruckner-Tuderman
	Curative vs disease modifying strategies	Jakub Tolar
	<i>Open debate – how to focus research in EB; avoid being spread too thinly because of complexity; where and how to direct the research; is prioritisation possible or feasible</i>	
09:30	<b>COFFEE BREAK</b>	
10:00	<b>THE CHALLENGES OF EB SKIN</b>	<b>Chair:</b> Peter Marinkovich
	Wound healing mechanisms – macrophages	Sabine Eming
	Immune function and bacterial challenge	Josefine Hirschfeld
	Clinical research	Maria Jose Escamez
	Learnings from other diseases (cell inflammation)	David Abraham
	<i>Panel discussion</i>	
11:05	<b>INFLAMMATION, FIBROSIS, &amp; THERAPEUTICS</b>	<b>Chair:</b> Leena Bruckner-Tuderman
	Fibrosis in EB – mechanisms and anti-fibrotic strategies	Alexander Nyström
	Losartan for RDEB trial – results and international perspectives	Dimitra Kiritsi
	miRNA in EB	Giovanna Zambruno
	<i>Panel discussion</i>	
	Oral presentations	
	<b>112</b> Novel players in the establishment and progression of fibrosis in recessive dystrophic epidermolysis bullosa	Esteban Chacon-Solano

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	<b>25</b> Skin microbiome characteristics of dystrophic epidermolysis bullosa patient	Liat Samuelov
<b>12:25</b>	<b>LUNCH</b>	
<b>13:25</b>	<b>CANCER &amp; CANCER THERAPEUTICS</b>	<b>Chair:</b> Johann Bauer
	<b>KEYNOTE: Precision medicine for SCC</b>	<b>Kevin Harrington</b>
	Genetic overview/current RDEB knowledge	Andy South
	Interdisciplinary management and therapies for EB associated cancers (skin, mucosal, internal)	Leena Bruckner-Tuderman
	RDEB-SCC protocols (rigosertib, pembrolizumab, cetuximab & nivolumab, SCC cream)	Jemima Mellerio
	<i>Panel discussion</i>	
	Oral presentations	
	<b>73</b> Morphological and morphometrical analysis of cutaneous squamous cell carcinoma in patients with recessive dystrophic epidermolysis bullosa: a prospective study	Angela Filoni
	<b>128</b> Heterogeneous addiction to TGFβ signalling in life threatening cutaneous squamous cell carcinomas arising in recessive dystrophic epidermolysis bullosa	Jasbani Dayal
<b>15:10</b>	<b>TEA BREAK</b>	
<b>15:40</b>	<b>THERAPEUTICS</b>	<b>Chair:</b> Jouni Uitto
	EBS target and treatment options/targeting EB as an inflammatory disease	Thomas Magin
	Read-through therapeutics: drug re-purposing for EB patients	David Woodley
	Topical treatments – an update	Dedee Murrell
<b>16:20</b>	<b>NATURAL HISTORY – IMPLICATIONS FOR CLINICAL TRIAL DESIGN</b>	<b>Chair:</b> Jouni Uitto
	PEBLES	Jemima Mellerio
	The EB clinical characterisation and outcomes database	Anna Bruckner
	<i>Discussion</i>	
<b>16:55</b>	<b>CLOSE</b>	

# CONGRESS PROGRAMME



**TUESDAY, 21 JANUARY 2020**

TIME	PLENARY SESSIONS – DAY 2	SPEAKER
08:30	<b>WELCOME</b>	<b>Chairs:</b> Jemima Mellerio & Jouni Uitto
	From theory to practice – translational medicine into clinical trials	Jemima Mellerio
08:40	<b>CELL MANIPULATION &amp; THERAPIES</b>	<b>Chair:</b> John McGrath
	Combining approaches – BMT & systemic treatment	Jakub Tolar
	Fibroblast gene therapy	Su Lwin
	HMGB1 peptide	Katsuto Tamai
	State of the art iPSCs	Dennis Roop
	<i>Panel discussion</i>	
	Oral presentations	
	<b>115</b> Efficient genome editing for correction of recessive dystrophic epidermolysis bullosa in iPS cells using CRISPR/Cas9 RiboNucleoProtein complexes	Joanna Jackow
	<b>7</b> Self-assembled skin substitutes and retroviral gene therapy for the permanent treatment of recessive dystrophic epidermolysis bullosa	Martin Barbier
10:25	<b>COFFEE BREAK</b>	
10:55	<b>GENE MANIPULATIONS &amp; THERAPIES</b>	<b>Chair:</b> Jakub Tolar
	State of the art gene editing	Fernando Larcher
	EBGRAFT and advances in skin grafting using improved vectors	Alain Hovnanian
	Hologene projects	Laura de Rosa
	Next generation sequencing applications for mutation detection in EB	Jouni Uitto
	Gene therapy for RDEB (ex-vivo vs in-vivo)	Peter Marinkovich
	Exon skipping for RDEB	Peter van den Akker
	COL17A1 editing using CRISPR/Cas9	Ulrich Koller
	Non-viral gene therapy	Lara Cutlar
	<i>Panel discussion</i>	
	Oral presentations	

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	<p><b>81</b> CRISPR/Cas9-based gene editing strategies for clinically-relevant ex vivo correction of Recessive Dystrophic Epidermolysis Bullosa</p> <p><b>86</b> Possible application of broad and unidirectional genome editing using the novel CRISPR-Cas3 system for autosomal dominant Epidermolysis Bullosa</p>	<p>Jose Bonafont Arago</p> <p>Hiroyuki Morisaka</p>
<b>13:25</b>	<b>LUNCH</b>	
<b>14:25</b>	<b>CLINICAL TRIALS &amp; RESEARCH PROGRAMMES</b>	<b>Chair: Johann Bauer</b>
	Introduction – clinical trial design	Johann Bauer
	Large wounds: an update on natural history data and EB101	Jean Tang
	Recombinant collagen 7: a systemic approach to RDEB therapy	Theresa Podrebarac
	A tale of two therapeutic approaches - an update on clinical studies in diacerein and FCX-007	Mary Spellman
	B-VEC off the shelf topical gene therapy for DEB	Suma Krishnan
	ABCB5+ mesenchymal stem cells for the treatment of recessive dystrophic epidermolysis bullosa – from bench to bedside	Kathrin Dieter
	<i>Questions</i>	
<b>16:10</b>	<b>TEA BREAK</b>	
<b>16:40</b>	Partnerships – charities, regulators, and industry	Brett Kopelan
<b>16:50</b>	Partnership Approach to research funding	Sharmila Collins
<b>17:00</b>	<b>CURRENT DILEMMAS – A CONGRESS DEBATE</b> <i>Measurement &amp; research</i>	<b>Chair: Jouni Uitto</b>
	EB relevant endpoints	Dedee Murrell
	Quality of life measures	Elena Pope
	Regulatory perspectives and challenges	Brett Kopelan
	Planning for clinical translation	Catriona Crombie
	Innovative therapeutic development	Mauro Peretti
	<i>Open debate – how to direct and support the development of meaningful outcome measures to ensure a positive outcome; working together – charities, regulators, and industry – where are the opportunities and gaps</i>	
<b>17:45</b>	<b>CLOSE</b>	

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**WEDNESDAY, 22 JANUARY 2020**

TIME	PLENARY SESSIONS – DAY 3	SPEAKER
08:30	<b>WELCOME</b>	<b>Chairs:</b> Jemima Mellerio & Jouni Uitto
	Update – EB-CLINET	Sophie Kitzmüller
	Clinical challenges of EB skin – inside and out	Anna Martinez
	Advances in diagnostics/phenotyping (CPG)	Cristina Has
	<i>Questions</i>	
08:55	<b>CURRENT DILEMMAS – A CONGRESS DEBATE</b> <i>Funding challenges</i>	<b>Chair:</b> Al Lane
	Individualised therapies	Leena Bruckner-Tuderman
	Allocation of research funds	Johann Bauer
	Funding for EB research	Al Lane
	Costs and accessibility	Peter Marinkovich
	<i>Open debate – how to increase research funding for EB; models of funding research; return on investment, ensuring we can develop management strategies for the wider group of patients/all types of EB</i>	
09:35	<b>UPDATE ON CLINICAL MANAGEMENT STRATEGIES</b>	<b>Chairs:</b> Anna Martinez & Ignacia Fuentes
	Superficial EBS/peeling skin syndromes	Eli Sprecher
	Wound care (JEB)	Katie Plevy
	Oesophageal management	Anna Bruckner
	Gastrostomy	Joe Curry
	ENT	David Albert
	Oral health	Susanne Kramer
	<b>COFFEE BREAK</b>	
	Eye	Michael Bartschi
	Growth patterns	Antonia Reimer
	Anaemia	Irene Lara-Corrales
	Hand surgery – management strategies	Gill Smith
	Surgery for SCC	Catina Bernardis
	Podiatry	Tariq Khan
	<i>Questions</i>	

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12:25	<b>LUNCH</b>	
13:25	<b>CLINICAL &amp; MANAGEMENT CASE HISTORIES</b>	<b>Chairs:</b> Eli Sprecher & Hana Buckova
	CASE REPORT: SCC management and response	Anna Bruckner
	CASE REPORT: SCC management and response	Pavel Rotschein
13:55	<b>QUALITY OF LIFE (PAIN &amp; ITCH)</b>	<b>Chair:</b> Marieke Bolling
	Cannabinoids – pain and itch	Nic Schrader
	Small fibre neuropathy	Margarita Calvo
	Prevention and treatment of pain	Boris Zernikow
	Pruritus in EB	Hagen Ott
	<i>Questions</i>	
14:50	<b>TEA BREAK</b>	
15:20	<b>SUPPORT MODELS</b>	<b>Chairs:</b> Anna Martinez & Anja Diem
	Multidisciplinary team best practice (GSTT) Rare Disease Centre	Jemima Mellerio
	Managing EB in a resource limited setting	Ravi Hiremagalore
	Telemedicine	Danielle Greenblatt
	EB-associated SCC: The Jefferson Adult EB and Complex Skin Cancer Clinic	Bahar Dasgeb
	Educational programmes	Christine Bodemer
	International EB patient registry	Godfrey Fletcher
	Innovation in patient data platforms	Michael Hund
	<i>Panel discussion</i>	
16:45	<b>STRATEGIES FOR CARE – STATUS UPDATE</b>	<b>Chairs:</b> Anna Martinez & Anja Diem
	A potential new therapeutic approach targeting wound infection – disrupting bacterial communication	Erik Gerner
	Development of two topical approaches to wound healing in EB. An update on progress with Oleogel-S10 and AP 103	Mark Sumeray
	The impact of antimicrobial resistance on topical treatment selection	Gilles Brackman
	The role of Hydrofiber dressings in EB wound management	Rachel Torkington-Stokes
	<i>Questions</i>	
18:15	<b>CLOSE</b>	

# CONGRESS PROGRAMME



**THURSDAY, 23 JANUARY 2020**

TIME	PLENARY SESSIONS – DAY 4	SPEAKER
09:00	<b>WELCOME</b>	<b>Chairs:</b> Jemima Mellerio & Jouni Uitto
09:05	<b>A GLOBAL APPROACH TO EB</b>	<b>Chair:</b> Brett Kopelan
	DEBRAs around the world – a 40 year history	Evanina Morcillo Makow
	DEBRA International – co-ordination and collaboration	Brett Kopelan/Jimmy Fearon
	<i>Questions</i>	
09:40	<b>UPDATES IN EB RESEARCH</b>	<b>Chair:</b> Jouni Uitto
	Research update (cell)	Su Lwin
	Research update (gene and protein)	Peter Marinkovich
	Cancer & cancer therapeutics	Jemima Mellerio
	Advances in clinical management strategies	Anna Martinez
	Biotech commercial development	Brett Kopelan
	<i>Questions</i>	
10:55	<b>COFFEE BREAK</b>	
11:35	<b>CURRENT DILEMMAS – A CONGRESS DEBATE</b> <i>Patients in clinical trials</i>	<b>Chair:</b> Jouni Uitto
	Clinical trials explained	Gabriela Petrof
	Patient and parent perspectives	TBA
	Challenges of clinical trial design	Christine Prodingler
	Using registries and big data	Godfrey Fletcher
	<i>Open debate – clinical trial challenges and dilemmas from all points of view</i>	
12:15	<b>LIVING WITH EB</b>	<b>Chairs:</b> Anna Martinez & Anja Diem
	Quality of life – clinician and patient working together	Natasha Harper
	Family and community How can patients & parents become active in the community, family dynamics and living with EB across the ages	TBA
	Education and family support/care & social services	Sam Geuens
	<i>Discussion</i>	
	<i>Chair invites people to submit written questions during lunch and tea breaks for the open forum at the end of the day</i>	



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13:15	LUNCH	
14:30	UPDATES IN EB CLINICAL CARE	<b>Chairs:</b> Jemima Mellerio & Agnes Schwieger-Briel
	Clinical practice guidelines, patient versions and EB infographics updates	Kattya Mayre-Chilton
	The importance of exercise in EB	Amy Price
	Occupational therapy in EB	Jennifer Chan
	Psychosocial guidelines	Petra de Graaf
	EB and pregnancy	Danielle Greenblatt
	Nutrition	Lynne Hubbard
	Oral health	Susanne Kramer
	<i>Questions</i>	
15:55	TEA BREAK	
16:40	EB COMMUNITY OPEN FORM <i>Ask the panel</i>	<b>Facilitator:</b> Jemima Mellerio
	<p>Questions invited on:</p> <ul style="list-style-type: none"> <li>• Itch and pain management</li> <li>• Bandaging</li> <li>• Post hand surgery</li> <li>• Approaches to different treatments for wounds on the face, neck, and ears</li> <li>• Psychosocial support for patients, carers and family</li> <li>• Gynaecology</li> <li>• Pregnancy and childbirth</li> <li>• Eye care</li> <li>• Treating wounds in tricky areas</li> <li>• Nutrition</li> <li>• Oral health</li> <li>• Exercise</li> </ul>	<b>Panel members:</b> Jane Clapham, Lynne Hubbard, Mark Popenhagen, Rachel Box, Susanne Kramer, Susana Morley
17:25	CLOSE	

# CONGRESS PROGRAMME



## SIDE FORA

### EB COMMUNITY – OPEN FORUM

*(FOR COMMUNITY MEMBERS TO DISCUSS INDIVIDUAL ISSUES WITH HEALTHCARE PROFESSIONALS)*

DAY	TIME	TOPIC
23 January	13:00 - 15:00	Allied healthcare professionals in-depth discussions

### CLINICAL PRACTICE GUIDELINE (CPG) MEETINGS – BY INVITATION ONLY

DAY	TIME	TOPIC
19 January	09:00 - 17:00	Transition in EB
20 January	09:00 - 17:00	Pregnancy, childbirth, and aftercare in EB
21 January	09:00 - 17:00	Palliative care in EB
22 January	09:00 - 17:00	Enteral and parenteral nutrition in EB
23 January	09:00 - 17:00	Neonatal care in EB

### DEBRA GROUPS – BY INVITATION ONLY

DAY	TIME	TOPIC
20 January	10:00 - 12:00	Marketing and Communications
20 January	13:30 - 15:30	Community/Family support
21 January	11:00 – 13:00	Fundraising
21 January	14:30 - 16:30	Advocacy/Lobbying
22 January	18:00 - 21:00	DEBRA International and DEBRA groups

### ALLIED HEALTHCARE PROFESSIONALS FORUM – SIGN-UP ONLY

*([WWW.EBWORLDCONGRESS.ORG/ALLIED-HEALTHCARE-PROFESSIONALS-FORUM](http://WWW.EBWORLDCONGRESS.ORG/ALLIED-HEALTHCARE-PROFESSIONALS-FORUM))*

DAY	TIME	TOPIC
21 January	14:30 - 17:30	Allied Healthcare Professionals

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## OTHER MEETINGS – BY INVITATION ONLY

DAY	TIME	TOPIC
19 January	08:30 - 12:00	DEBRA International
19 January	13:00 - 17:00	EB Without Borders
22 January	08:00 - 17:00	DEBRA International

## ALLIED HEALTHCARE PROFESSIONALS COURSE – SIGN-UP ONLY ([WWW.DEBRA.ORG.UK/PODIATRY](http://WWW.DEBRA.ORG.UK/PODIATRY); BURSARIES AVAILABLE)

DAY	TIME	TOPIC
21 January	09:00 - 13:00	EB Skills in Podiatry Course

## ALL CONGRESS DELEGATES – SIGN-UP ONLY ([WWW.EBWORLDCONGRESS.ORG/RDC-TOUR](http://WWW.EBWORLDCONGRESS.ORG/RDC-TOUR))

DAY	TIME	TOPIC
21 January	18:15 - 19:45	Rare Diseases Centre Tour

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## ORAL POSTER STATION PRESENTATIONS

20 JANUARY

STATION	TIME	PRESENTER	POSTER
ONE	12:30 - 12:40	Christina Guttman-Gruber	<p>34</p> <p>The impact of low-dose calcipotriol ointment on wound healing, pruritus and pain in patients with dystrophic epidermolysis bullosa</p> <p><i>Christina Guttman-Gruber, Josefina Piñón Hofbauer, Birgit Tockner, Victoria Reichl, Peter Hofbauer, Martin Wolkersdorfer, John E. Common, Anja Diem, Katharina Ude-Schoder, Wolfgang Hitzl, Florian Lagler, Julia Reichelt, Johann W. Bauer, Roland Lang and Martin Laimer</i></p>
ONE	12:45 - 12:55	Hannah Mumber	<p>137</p> <p>Biallelic JUP Mutation in Families with Arrhythmogenic Right Ventricular Cardiomyopathy and Skin Fragility in the Form of Epidermolysis Bullosa Simplex: Naxos Disease</p> <p><i>Hannah Mumber, Anjali Rajan, Hasssan Vahidnezhad, PhD, Leila Yousefian, MSc, M. Faghankhani, MD, Nikoo Mozafari, MD, Amir Hossein Saeidian, MSc, Fatemeh Niaziarimi, MSc, Fahimeh. Abdollahimajid, MD, Soheila Sotoudeh, MD, F. Rajabi, MD, Liaosadat Mirsaefaei, MD, Alizadeh-Sani, Lu Liu, PhD, Alyson Guy, MSc, Sirous Zeinali, PhD, Ariana Kariminejad, MD, John A. McGrath, MD, FRCP, FMedSci, and Jouni Uitto, MD, PhD. # Both authors contributed equally to this work</i></p>
TWO	12:30 - 12:40	Vicki Chen	<p>107</p> <p>Understanding ocular disease in the DEB mouse model: challenges of asymmetry and survival</p> <p><i>Vicki M. Chen, MD, Rajani Shelke, MS, Noorjahan Panjwani, PhD, Zhiyi Cao, PhD, Michael Esmail, VMD, M. Peter Marinkovich, MD</i></p>
TWO	12:45 - 12:55	Christina Prodingler	<p>10</p> <p>A case series of six paediatric cases with laryngo-onycho-cutaneous syndrome (LOC)</p> <p><i>Christine Prodingler, Subhanithaya Chottianchaiwat, Jemma Mellerio, John McGrath, Lu Liu, Martin Laimer, Gabriela Petrof, Anna Martinez</i></p>

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21 JANUARY

STATION	TIME	PRESENTER	POSTER
ONE	13:35 - 13:45	Karen Snelson	6  Guidelines for the care of adults with EB undergoing clinical and surgical procedures.  <i>Karen Snelson</i>
ONE	13:50 - 14:00	Cristina Has	2  Molecular and mutational signatures of squamous cell carcinomas in epidermolysis bullosa  <i>Sisi Lu, Maria E. Hess, Antonia Reimer, Daniele Castiglia, Yinghong He, Dagmar von Bubnoff, Giovanna Zambruno, Andreas Volz, Hauke Schumann, David Rafei-Shamsabadi, Frank Meiss, Melanie Boerries, Cristina Has</i>
TWO	13:35 - 13:45	Ashjan Alheggi	39  Treatment of multifactorial anaemia in adults with severe epidermolysis bullosa using intravenous ferric carboxymaltose: a single institution, observational, retrospective study  <i>A. Alheggi, J.A. McGrath, L. Hubbard and D.T. Greenblatt. J.E.Mellerio</i>
TWO	13:50 - 14:00	Vicki Chen	120  Anterior segment spectral domain optical coherence tomography in epidermolysis bullosa  <i>Nihaal Mehta BA, Calvin Robbins BA, Elizabeth Noh BS, Nadia K. Waheed MD MPH, Vicki M. Chen MD</i>

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22 JANUARY

STATION	TIME	PRESENTER	POSTER
ONE	12:40 - 12:50	Catherine Miller	42  Hand Contracture Development in Children with Recessive Dystrophic Epidermolysis Bullosa  <i>Ms Nicky Jessop &amp; Ms Catherine Miller</i>
ONE	12:55 - 13:05	Mark O'Sullivan	93  The effect of rocker bottom footwear on foot biomechanics and the development of plantar blisters in patients with epidermolysis bullosa simplex; a pilot study  <i>Mark O'Sullivan, Lisa James, Adrian Heagerty, Natasha Harper</i>
TWO	12:40 - 12:50	Suma Krishnan	52  Results from a Phase I/II study of a topical gene therapy (bercolagene telserpavec, B-VEC) in patients with recessive dystrophic epidermolysis bullosa (RDEB)  <i>M P. Marinkovich, Schuyler Vinzant, Pooja Agarwal, Suma Krishnan</i>
TWO	12:55 - 13:05	Tobias Zahn	47  Losartan for EB, or 'It takes a village to raise a child'  <i>Dimitra Kiritsi, Tobias Zahn, Frank Hoffmann, Sigrid Saaler- Reinhardt, Leena Bruckner-Tuderman</i>