BADGEM Clinical Meeting – BAD annual meeting Manchester

Date: Thursday 9th July
Time: 08:30-12:00
Room: Exchange Hall
Chair: Professor Birgit Lane
Professor John McGrath

badgem | British Association of Dermatologists Dermatology and Genetic Medicine

Thursday 9th July

PROGRAMME		
08:30 – 08:35		Welcome BADGEM Clinical Meeting – **Professor Irene Leigh**
08:35 – 08:45	BG01	**Xeroderma pigmentosum: increasing awareness of the variation in presenting clinical features in order to avoid diagnostic delay and improve prognosis** M. Sethi, A. Lehmann, D. McGibbon, R. Sarkany and H. Fassihi
08:45 – 08:55	BG02	**Clinical heterogeneity in autosomal recessive congenital ichthyosis due to *NIPAL4*/ichthyin mutations** S.T. Ngu, H. Cordey, A. Affleck, A. Terron-Kwiatowski, D. Baty, D. Goudie, S.J. Brown, C.J. Jury and M. Zamiri
08:55 – 09:15		Invited Talk **Using rare disease genetics to understand common skin disorders** Professor Maurice Van Steensel
09:15 – 09:25	BG03	**Co-occurrence of *NRAS* and *BRAF* activating mutations in congenital melanocytic naevi** S. Polubothu, J. Chalker and V. Kinsler
09:25 – 09:45		Invited Talk **A clinical update on filaggrin** Professor Alan Irvine
09:45 – 09:55	BG04	**A novel peeling skin syndrome** S. Chinthapalli, D. Buckley, Z. Lin, D. Nitoiu, C. Scott, F. Smith, Y. Yang, D. Kelsell and E. O'Toole
09:55 – 10:20		Invited Talk **Congenital erythroderma: the many shades of red** Professor Eli Sprecher
10:20 – 10:45	Break	Exhibition Hall
10:45 – 11:10		Invited Talk **Delivering the National Rare Disease Register for England** Dr Jem Rashbass
11:10 – 11:25		**BADGEM Initiatives** Professor Edel O'Toole & Dr Neil Rajan
11:25 – 11:35	BG05	**Mutations in desmoglein 1 causing clinically heterogeneous inherited palmoplantar keratoderma** M.-L. Lovgren, N.J. Wilson, M.E. Schwartz, F.J.D. Smith and M. Zamiri
11:35 – 12:00		**Genetics and Immunology of Alopecia Areata** Professor Angela Christiano

POSTERS		
	BG06	**Prospective Epidermolysis Bullosa Longitudinal Evaluation Study (PEBLES): development of an electronic data capture tool in recessive dystrophic epidermolysis bullosa** E. Pillay, S. Robertson, A. Martinez and J. Mellerio
	BG07	**Characterization and selection of a patient cohort for a phase I/II study of *ex vivo* gene therapy for recessive dystrophic epidermolysis bullosa (GENEGRAFT)** A. Abdul-Wahab, M. Titeux, J. Mellerio, J. McGrath and A. Hovnanian
	BG08	**Farber disease: an unusual case in a dermatology clinic** P. Gupta and P. Beattie
	BG09	**Two cases of patients with Ehlers–Danlos syndrome type VIII and hoarseness** S. George, A. Vandersteen, E. Nigar, D. Ferguson, E. Topham and F.M. Pope
	BG10	**Chromosome 3 partial triplication in association with pigmentary mosaicism** R. Waas, M. Splitt and S. Leech
	BG11	**Two cases of rare variants of dystrophic epidermolysis bullosa with late onset of symptoms** W. Szczecinska and A. Heagerty
	BG12	**A second family with a telomerase reverse transcriptase (*TERT*) promoter gene mutation associated with melanomas** D. Tang, S. O'Shea, D. Espinoza, N. Rajan, A. Carmichael and J. Newton-Bishop
	BG13	**Full-thickness skin grafting for pseudoainhum in loricrin palmoplantar keratoderma** P. Broadbent, S. Turner and M. Zamiri
	BG14	**Neurofibromatosis type I or Legius syndrome? An emerging diagnostic challenge** W.Y. Haw, V. Harrison, M. Oakford and K. Godfrey
	BG15	**Deafness and a recurrent erythrokeratoderma-like rash with marked perineal involvement, due to a heterozygous mutation in *GJB2* (connexin 26, F142L)** M.-L. Lovgren, J. Robinson, A. Terron-Kwiatowski, D. Baty, M. Zamiri and A. Waters
	BG16	**New missense mutation in type VII collagen in an adult with autosomal recessive pretibial epidermolysis bullosa** L. Cunningham, L. Liu, S. Menzies, J.A. McGrath and A. Lally
	BG17	**Linear skin defects and microphthalmia: a case report** A. Durack, N. Burrows and S. Mehta
	BG18	**Cystic fibrosis carrier status presenting with aquagenic palmar wrinkling** A. Durack, A. Sterling and P. Todd

The poster viewing session will take place on Thursday lunchtime