BADGEM Clinical Meeting

Date: Thursday 7th July
Time: 13:15-17:30

badgem | British Association of Dermatologists Dermatology and Genetic Medicine

Chair: **Professor Irene Leigh & Professor Sara Brown**

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PROGRAMME		
13:15 – 13:20		Welcome BADGEM Clinical Meeting – **Professor Irene Leigh**
13:20 – 13:45		Invited Talk **Lessons from EB-Clinet – A clinical network for a rare disease** PROFESSOR JOHANN BAUER (Salzburg, Austria)
13:45 – 13:55	BG01	**Lessons learnt from the design and development of a multicenter rare genetic skin disease research study** S.Tso, J. Simpson, M. Martinez-Queipo, E.Glass and J. McGrath
13:55 – 14:05	BG02	**Vulvovaginal symptoms in women with epidermolysis bullosa** E. Orrin, N. Alband, A Abdul-Wahab, E. Wedgeworth and J. Mellerio
14:05 – 14:30		Invited Talk **Cutis laxa: the journey to a clinical nosology** PROFESSOR BERT CALLEWAERT (Ghent, Belgium)
14:30 – 15:15 15:15 – 15:30 15:30 – 16:00	Break	Rook Oration Focus Sessions
16:00 – 16:25		Invited Talk **Hereditary Cancer as seen from the outside** SIR JOHN BURN (Newcastle)
16:25 – 16:35	BG03	**Genetic and phenotypic heterogeneity in Ferguson-Smith Disease: a case series** Z. Shukur, D. Goudie, N. Chung, S. Whittaker, A. Shaw and K. Lacy
16:35 – 16:45	BG04	**Cutaneous hyperpigmentation and familial gastrointestinal stromal tumour associated with c-kit mutation** G. Wali, D. Halliday, J. Dua, E. Ieremia, T. McPherson and R. Matin
16:45 – 17:10		Invited Talk **Dysmorphology in the sequencing era; clinicians still have a role** PROFESSOR DION DONNAI (Manchester)
17:10 – 17:20	BG05	**Novel mutations in SLURP1 causing Mal de Meleda palmoplantar keratoderma with varying phenotypes** S. Ziaj, M. Pigors, C. Scott, F. Lewis, I. Ali, N. Roberts, D.P. Kelsell and E. A. O'Toole
17:20 – 17:30		**BADGEM Initiatives** Neil Rajan (Newcastle) & Dr Simon Tso (London)
POSTERS		
	BG06	**Mild arthrogryposis, renal dysfunction and cholestasis (ARC) syndrome caused by a novel splice site mutation in *VPS33B*** A. Ahmed, L. McGinty, D. Blaydon, D. Kelsell, E. O'Toole and B. De Silva
	BG07	**Tracking tumour kinetics in patients with germline CYLD mutations** S. Brown, S. Worthy, S. Barnard, J. Langtry and N. Rajan
	BG08	***FOXN1* duplication and congenital hypertrichosis: a case report** E. Gilhooley, C. Feighery and S. Collins
	BG09	**Will a genetic diagnosis of poikiloderma with neutropenia influence management?** S. El-Heis, M. Ardern-Jones and K. Godfrey

	BG10	**A dramatically different case of Dowling-Degos disease** S. Sherif, N. Spierings, R. Betz, H. Chong and M. Singh
	BG11	**Muckle-Wells Syndrome: a novel presentation** J. Clowry, E. Nic Dhonncha, J. Aróstegui, P. Brogan, D Rowczenio, C. Sheey, P. Hawkins and S. Field
	BG12	**Terminal Osseous Dysplasia with Pigmentary Defects** S. H. Foo, R. Goodwin, A. Rawlinson, V. Jones, A. Fry and C. Moss
	BG13	**Multiple Well-differentiated SCC's: A case of Ferguson-Smith Disease and a review of the literature for use of Acitretin in this condition** A. Lowe, W.A Woo, L. Atkinson and E. Topham
	BG14	**Dermatologists play a key role in diagnosing LEOPARD syndrome.** S. Mirhadi, H. Fassihi, L. Islam and R. Ramesh
	BG15	**Brooke-Spiegler Syndrome: An unusual presentation with a novel underlying mutation** S. Whittaker and J. Hughes
	BG16	**An interesting case of LEOPARD syndrome** S. Krishna, J. Natkunarajah and A. Fogo