

BADGEM Clinical Meeting Glasgow 1st July 2014 9am – 1pm, Dochart 2 room

09.00 – 09.05 Welcome to BADGEM Clinical Meeting, Professor Irene Leigh

Chair: Professor Irene Leigh

09.05 – 09.30 **Invited Talk 1** “What’s new in Epidermolysis bullosa?” Professor John McGrath, King’s College London

09.30 – 09.40 **Case report 1** “Ear complications in inversa type recessive dystrophic epidermolysis bullosa (RDEB-I); a case series of 6 patients” Presenter – Dr. Susan Robertson, Guy’s and St Thomas’ NHS Foundation Trust/Great Ormond Street Hospital, London

09.40 – 09.50 **Case report 2** “Epidermolysis Bullosa Pruriginosa: Two novel COL7A1 mutations” Presenter – Dr. Arpita Debroy Kidambi, Royal Hallamshire Hospital

09.50 – 10.15 **Invited Talk 2** “Ichthyoses: what’s new?” Dr. Vinzenz Oji, University Hospital Münster

10.15 – 10.25 **Case report 3** “Progressive Osseous Heteroplasia” Presenter – Dr. Prativa Jayasekera, Royal Liverpool and Broadgreen Hospitals NHS Trust

10.25 – 10.35 **Case report 4** “Acral Peeling Syndrome secondary to CSTA gene mutation” Presenter – Dr. Kayria Muttardi, Watford General Hospital

10.35 – 10.55 **Coffee Break**

Chair: Professor John McGrath

10.55 – 11.20 **Invited Talk 3** “What’s new in keratodermas?” Professor Edel O’Toole, Barts and London School of Medicine and Dentistry

11.20 – 11.30 **Case report 5** “Vohwinkel syndrome with a novel Loricrin microdeletion” Presenter – Amjad Khan, Great Ormond Street Hospital, London

11.30 - 11.40 **Case report 6** “Bullous Ichthyosis of Siemens” Presenter – Dr. Anna Dubois, Royal Victoria Infirmary, Newcastle Upon Tyne

11.40 – 11.50 **Case report 7** “A novel RASA1 mutation presenting with multiple cutaneous capillary malformations” Presenter – Dr. Sairan Whitaker, Royal Victoria Infirmary, Newcastle Upon Tyne

11.50 – 12.15 **Invited Talk 4** “New ways of diagnosing genetic skin disease” Professor David Kelsell, Barts and London School of Medicine and Dentistry

12.15 – 12.25 **Case report 8** “Focal dermal hypoplasia (Goltz syndrome)” Presenter – Dr. Roland Chu, Royal Infirmary Edinburgh

12.25 – 12.35 **Case report 9** “Annular erythema of infancy? A common disease pathway with atypical HUS” Presenter – Dr. Ting Seng Tang, Nottingham University Hospitals NHS Trust

12.35 – 13.00 **Invited Talk 5** “XP and DNA repair. What can a national service offer?” Dr. Robert Sarkany, Guy’s and St Thomas’ NHS Foundation Trust, London