Amba Hotel, Charing Cross, London
11-12 March 2016

AGENDA

Friday 11 March

15.00 – 19.00	Registration Open (Outside the Ballroom, 1st Floor)
19.00 – 19.45	Drinks Reception (The Regency, 1st Floor)
20.00 – 22.30	Welcome Dinner (The Ballroom, 1st Floor)

Saturday 12 March

TIME	TITLE	SPEAKER	SUMMARY
08:30 – 08:45	Welcome	**Caroline Harding** CEO Genetic Disorders UK	Caroline Harding will introduce the work of Genetic Disorders UK and provide an overview of the day's programme.
08:45 – 09:15 Incl. Q&A	Gene editing for genetic disorders – cut, correct and cure?	**Dr Patrick Harrison** University of Cork	Dr Patrick Harrison will explain what gene editing is (and isn't), show how it is being used as a research tool for a range of genetic disorders, and discuss the challenges in bringing this technique to the clinic.
09:15 - 09:25	Georgia's journey: The story of the first child diagnosed by the 100,000 genomes project.	**Amanda Walburn-Green** Mother	Amanda Walburn-Green will talk about her search for a diagnosis for her daughter, Georgia, and how it felt to finally get a name for the genetic disorder that affects her special little girl.
09:25 – 09:45	The evolving landscape for the management of genetic disorders within the NHS	**Andrew Wilkinson** Deputy Director Specialised Healthcare Alliance	Andrew Wilkinson will provide an overview of how genetic disorders are managed at a national level as part of specialised commissioning and the changes that may affect patients and clinicians in the months ahead.
09:45 – 10:05	How to make a short film about your genetic disorder?	**Gretchen Shoring** Managing Director Citizen Films	Gretchen Shoring will talk about the key components to making an engaging film about a genetic disorder and how patient groups might approach making a film regardless of budget.
10:05 – 10:15	Collaboration Works! The Haemochromatosis Society and Boots Pharmacies	**David Head** Chief Executive The Haemochromatosis Society	David Head will explain how The Haemochromatosis Society was able to secure the support and co-operation of Boots Pharmacies to create CPD level training material for their pharmacies and pharmacy technicians.
10:15 – 10:45	BREAK		
10:45 – 11:15 Incl. Q&A	The teenage years: The evolving nature of dependence and interdependence.	**Professor Adrian Sutton** Consultant in Child & Family Psychiatry and Psychotherapy	Professor Adrian Sutton will give an overview of the relational challenges for teenagers, parents and practitioners when families are affected by a genetic disorder.
11:15 - 11:30	Life as a teenager with a genetic disorder	**Holly van Geffen**	'Always have hope' - Holly van Geffen will talk about how a can-do attitude motivated her to achieve her aspirations throughout her teenage years despite the challenges of a life-limiting genetic disorder.
11:30 – 11:40		**Annabelle Davis**	'Always looking up' - Annabelle Davis will speak about how she has taken life's emotional and physical challenges in her stride with determination and optimism.
11:40 – 12:05 Incl. Q&A	Providing genetic counselling to teenagers in families affected by a genetic disorder.	**Dr Christine Patch** Consultant Genetic Counsellor Guy's Hospital	Dr Christine Patch will discuss the provision of genetic counselling to both carrier and affected teenagers, and provide a practical overview of the approach to this evolving area of counselling.
12:05 – 12:20	Building relationships with young people through digital channels	**Jacqueline Ali** Head of Information and Support & **Holly-Rae Smith** Youth Empowerment Officer Cystic Fibrosis Trust	Jacqueline Ali and Holly-Rae Smith will discuss how the Cystic Fibrosis Trust used a short film to kick start a programme of online engagement with young people affected by cystic fibrosis.

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12:20 – 12:40	How to make a website fit for 2016 and beyond?	**Robert Jones** Managing Director, Noovo Creative	Robert Jones will explain some of the easy ways to improve and manage a website in just a few minutes a week as well as explaining how to give visitors a better user experience.
12:40 – 13:50	LUNCH		
13:50 – 14:20	Transition: Supporting young people move into adulthood.	**Julia Hodgson** Practice Development Manager Together for Short Lives	Julia Hodgson will discuss the problems faced by many young adults with complex health conditions as they transfer from children's to adult services, and will outline the work that the Transition Taskforce is doing to address them.
14:20- 14:40	Courage is Compulsory	**Laura, Tori and Sam**	Laura, Tori and Sam will discuss the book they have written together about disability and the lessons they have for young people and their families who are facing similar challenges.
14:40 – 14:55	Creating Roald Dahl Nurse Specialists	**Sophie Dziwinski** Roald Dahl's Marvellous Children's Charity	Sophie Dziwinski will talk about the work of Roald Dahl's Marvellous Children's Charity in supporting children and young people with rare diseases, genetic disorders and undiagnosed conditions by establishing new Roald Dahl nurse posts.
14.55 – 15.15	RD Connect: big data for rare disease	**Dr Pauline McCormack** Policy, Ethics & Life Sciences Research Centre Newcastle University	Pauline will talk about RD-CONNECT – an integrated platform connecting databases, registries, biobanks and clinical bioinformatics for international rare disease research.
15:20 – 15:40	BREAK		
15:40 – 16:00	Monitoring rare disease in the child population	**Richard Lynn** Scientific Coordinator British Paediatric Surveillance Unit	Richard Lynn will talk about the work of the British Paediatric Surveillance Unit and how the sharing of data is evolving to identify children affected by rare genetic disorders.
16:00 – 16:20	Creating and implementing a cost effective disorder-specific registry	**Daniel Lewi** Co-founder and Director Cure & Action for Tay Sachs Foundation **Mel McIntyre** Managing Director OpenApp	Daniel Lewi from CATS and Mel McIntyre from OpenApp will talk about how small charities supporting rare diseases can create and implement a cost effective disease specific registry.
16:20 – 16:30	Collaborating with industry on drug development	**Dr Julie Vallortigara** Research Officer Ataxia UK	Dr Julie Vallortigara will explain how Ataxia UK have collaborated with Pfizer on a drug discovery programme for Friedreich's ataxia.
16:30 – 17:00	The changing face of research into Batten disease	**Dr Sara Mole** Genetics and Genomics Medicine Programme UCL Institute of Child Health	Dr Sara Mole will discuss how her research into Batten disease has evolved over the last twenty years and reflect on where she hopes her current work on new drugs and therapies will take her and the European-wide consortium she now leads.
17:00 – 17:15	Wrap-up	**Caroline Harding** CEO Genetic Disorders UK	

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