

**Visit by Rare Diseases Policy Team, 19<sup>th</sup> January 2015**

**Chestnut Suite, Hinxton Hall  
From 13.30 Meadow Room, EBI South Building**

**AGENDA**

**Sanger Institute Participants:**

Julia Wilson, Associate Director External Relations  
Matthew Hurles, Group Leader Human Genetics Human Genetics Programme  
Caroline Wright, Senior Scientific Manager, Human Genetics Programme  
Helen Firth, Honorary Faculty Member, Human Genetics Programme/ Consultant Clinical Geneticist,  
Anna Middleton, Senior Staff Scientist, DDD Human Genetics Programme  
Jeff Barrett, Group Leader, Human Genetics Programme

**EBI Participants:**

Janet Thornton, Director  
Rolf Apweiler, Joint Associate Director and Senior Scientist  
Ewan Birney, Joint Associate Director and Senior Scientist  
Lindsey Crosswell, Head of External Relations  
Steven Newhouse, Head Technical Services  
John Overington, ChEMBL, Group Leader - ChEMBL  
John Marioni, Research Group Leader  
Alvis Brazma, Senior Team Leader, Functional Genomics

**DoH Participants:**

Mark Bale	Sinead Lane
Sylvia Pratt	Julie Pettman
Ray Smith	Catherine Byers
Francesca Orpen	Mel Pepper
Alice-Nancy Terry	Ben Halliday
Robin Herbert	Ian McKay

10:00	Sign in at Security Reception and walk to Hinxton Hall	Met by Francesca Gale
10:30	Welcome, coffee and Introductions Overview and purpose of visit	Julia Wilson and Mark Bale
11:00	Deciphering Developmental Disorders <ul style="list-style-type: none"> <li>- Research Programme</li> <li>- Ethics and Society considerations</li> <li>- Wider application to other rare diseases</li> </ul>	Matt Hurles Anna Middleton Helen Firth / Caroline Wright
12:15	Wrap up discussions	
12:30	Joint lunch with Sanger Institute and EBI	Chestnut Suite
13:30	<b>Meadow Room, EMBL-EBI South building</b>  Overview of EBI (Janet Thornton)  Tea and Coffee	Janet Thornton  Rolf Apweiler  Ewan Birney  Lindsey Crosswell
14:30	Current Health Research at EMBL-EBI (Ewan Birney) <ul style="list-style-type: none"> <li>- 100k genomes</li> <li>- Addenbrookes collaborations</li> <li>- Research</li> </ul>	As above, joined by :  John Overington  John Marioni  Alvis Brazma  Steven Newhouse
15.30	Wrap up meeting	Janet Thornton  Ewan Birney  Rolf Apweiler  Lindsey Crosswell
16.00	Depart	