

InSiGHT/Human Variome Project Meeting All day May 10th 2010
UNESCO Building Paris

Draft Times allocated

Proposed agenda: Welcome

1 **9:00** Governance Committee presentation (*Presenting in 15 mins: F Macrae and J den Dunnen*)

1.1 Alignment of LOVD data policy and InSiGHT terms of reference for governance (J den Dunnen*, T Smith, H Howard*, R Sijmons*, P Peltomaki)

1.2 Agreement on new document for Council Endorsement

1.3 Continuing governance processes: teleconferencing

1.4 The International MMR Consortium

1.5 MTAs, MOUs (*F Macrae*, A Devereau*): (*including recommendations for template agreement*)

1.6 Indemnity update

1.6.1 InSiGHT incorporation (*F Macrae**)

1.6.2 Negotiation with DMuDB (*F Macrae*, A Devereau*)

1.5 Incorporating CGen into the LOVD/ InSiGHT database? (*Presenting in 25 mins including discussion and recommendation: P Moller*, J den Dunnen**)

Participants: M Woods*, P Peltomaki, H Vasen, J den Dunnen*, A Devereau, T Smith, H Howard*, R Sijmons*, A Brookes, P Moller* but all welcome)

2 **9:40** Curation: (*Presenting: M Woods*: 10 mins*)

2.1 Processes and scale of curation, resources needed (M Woods*, R Sijmons*, P Peltomaki)

2.2 Funding Updates: (*Presenting: F Macrae*, R Cotton* T Weber*: 10 mins*)

Adopt a gene (Deloittes, Levi Strauss, Wrangler) R Cotton

NCI Market Requisition (F Macrae, S Schully)

R01 Application to NCI (*M Jenkins, F Macrae*)

Philanthropy (F Macrae, R Cotton)

Colon Cancer Challenge Foundation support for InSiGHT /HVP / Curation (*T Weber**)

1.Update on 2010 Colon Cancer Challenge, March 28th New York City

2. 2010 New York City Marathon Charity Runner Program: An additional opportunity for HVP / InSiGHT fundraising.

Other ideas: *anyone- please let Finlay know*

3 **10:00** Morning tea

4 **10:30** Interpretation Committee

4.1 Face to face introductions (*Chair: M Genuardi*: 5 mins*)

4.2 "Peak Picker" presentation: (*B Bapat*: (7 mins)*)

4.3 cDNA before functional assays? *E Holinski Feder* (7 mins)*

4.4 Agreement and Implementation of interpretation algorithm (Presenting: *S Tavgian* and M Greenblatt*, S Lipkin**, for *S Tavgian*, D Goldgar, M Greenblatt*, F Macrae*, S Lipkin*, A Spurdle*, T Weber** others) *48mins*

4.5 Endorsement of pathogenicity assignment of a panel of definitely pathogenic and definite polymorphisms: variants as presented by R

Hofstra. (Chair: M Genuardi*, assisted by R Hofstra*, M Woods*: 30 mins)

- 4.6 Assignment of pathogenicity to a group of variants as yet unclassified using the IARC/InSiGHT algorithm (as a test of the Interpretation committee processes: (M Greenblatt/S Tavtigian*: variants to be presented by M Woods*: 30 mins)

Participants in Interpretation Committee: M Genuardi* (Chair), J Utsunomiya, R Ramesar*, J Burn*, M Greenblatt*, P Peltomaki, R Hofstra*, R Sijmons*, R Scott, M Corish*, D Goldgar, M Woods*, B Bapat, S Tavtigian*, A Spurdle*, S Lipkin*, M Dunlop, I Frayling*, Elke Holinski Feder, D Goldgar, A Lindblom*, T Weber*, F Macrae*

5 12:30 Lunch

- 6 1:30 Functional assay Committee: (Presenting: R Hofstra*, R Sijmons*, L Rasmussen*, T Frebourg*, N de Winds*, E Holinski Feder*, M Nystrom: 90mins)
6:1 Consideration of variants selected by Interpretation Committee for use in calibrating/assessing functional assays.
6.2 Other items

7 3:00 Afternoon tea

- 8 3:30 Submission, disclosure and interconnectivity processes:
8.1 Number of Submissions to date; hits per month (Presenting: J den Dunnen*: 5 mins)
8.2 Progress with national and individual lab submissions (unclassified variants; all variants) (Presenting: Annika Lindblom*: 15 mins including discussion: Anyone - please let Finlay and Annika know)
8.3 Novel Strategies for the identification of MMR Mutation families in USA (T Weber*: 15 minutes)
4.4 Interconnectivity to and from LOVD
4.4.1 Updates: NCBI (Presenting 10mins altogether: Donna Maglott*), EBI (Presenting: Ilkka Lappalainen*, Paul Flicek)
4.4.2 Models of National nodes for the HVP: the Australian experience (Presenting: Alan Lo*: 15 mins)

5. 4:30 Phenotype issues

- 5.1 Processes of phenotype submission from clinics – beta testing volunteers (Presenting: F Macrae for the phenotype committee: 15mins: , F Macrae*, (Chair) , R Scott, S Clarke, C Burke, T Weber*, P Watson, A Lindblom*, P Rozen, G Moeslein*, I Bernstein*, A Spigelman*, H Vasen*)
5.2 Alterations to database? Eg ICD coding... (Any comments)
Phenotype Committee

7. 4:45 Virtual Histology

- 7.1 Progress (Presenting: Hans Morreau* 15mins, and others as interested – contact Hans)

8. 5:00 APC and MYH mutation databases report

Presenting 20 mins: Stefan Aretz and Carli Tops**

9. 5:15 Cost benefit analysis of the HVP – the InSiGHT pilot – (*Presenting: D
Quin*: 15 mins*) .

10: 5:30 Close

Other attendees: Stephen Haas,

Apologies: Paul Wise (Vanderbilt University), Paul Flicek (EBI) , Minna Nystrom (Helsinki), Sue Clark (St Marks) , Carol Burke (CCF), Randy Burt HCI, Utah), Eli Brazowski (TASMC, Israel), J Wijnen (Leiden), Paul Rozen (Israel), J P Mecklin (Helsinki), Rodney Scott (Australia)

Attendance uncertain: Paivi Peltomaki (Helsinki) Malcolm Dunlop (MRC HGU, Edinburgh), David Goldgar (HSC, Utah) , James Church (CCF),S Schully (NCI), John Baron (Dartmouth), G Suthers (Australia) .