Meeting Minutes
BM1105 WORKING GROUP LEADERS MEETING:
June 23-24, 2013
Institute Cochin, Paris

1. **Nelly provided an overview of the COST meeting in Latvia including the following highlights and « lessons learned » from successful actions :**
    a. Key metrics are: website hits, young investigators trained, publications & consensus statements, grant funding (i.e. FPs, Marie Curie, etc)
    b. A Wikipedia page on the action can help draw attention to the site
    c. Linking the Action website to individual team sites will make Google searching more successful and will draw traffic (**see action items**)
    d. Clinical consensus papers i.e. what is delayed puberty, evaluation of CHH, and treatment approaches to CHH should be goals for the clinical working group (WG1)
    e. An e-learning platform may be useful for education patients and clinicians alike on this rare disease
    f. A quarterly newsletter highlighting work of the Action and deliverables (web based)
    g. Patient advocacy groups should lobby their national funding agencies for increased attention to this condition
    h. Need to document inter-group publications across 2 or more countries → notify Ginny & Andrew
    i. Nicolas, Leo, & Mohammed to investigate beginning a ESPE working group on puberty (**see action items**)
    j. Discussed 1 page overview and 3-page summary article on CHH that could be translated (**see action items**)
    k. Lobbying for funding led to a discussion on names for a call and it was decided “Puberty disorders: Mechanisms and lifelong health consequences”
    l. Discussion of Marie Curie joint PhD training platform → Uli & Manuel to discuss and initiate (**see action items**)
    m. Connections with other COST Actions
        i. NGS
        ii. Zebrafish Core
        iii. DSD (Olaf Hiort (invite to Berlin)
        iv. Imprinting (Prof. Thomas EGGERMANN chair of BM1208)

2. **Website: Virginia Hughes provided an tour of the website and the following points were discussed and decided upon:**
    a. Separate Centers of Excellence: one for clinical, one for Molecular
    b. Separate Research Centers: one for clinical and one for genetics
    c. For Research page: map with centers, keywords for profiles
    d. Larger discussion of the utility of a format similar to Growth Consortium Site
    e. Decision to graphically display (i.e. venn diagram) molecular basis of nIHH/KS/syndromic CHH
    f. For gene curation page → add simple text on lay definitions for: mutation/variant, architecture/oligogenicity, penetrance, phenotype

    g. Patient information was reviewed and addition of female schematic was proposed

3. **Minimal Dataset: there was a robust discussion regarding the MDS its structure and utility. This led to a move for several minor adjustments and the following steps:**
    a. Examine rules for using database sent by J. Leger (**see action items**)
    b. Investigators to submit the MDS to the national level to determine sharing of anonymous patient data
    c. Revisit protections with Genohm for online Slims database
    d. Underscoring that individual investigators are individually responsible for ensuring adequate informed consent and adhering to national guidelines for data sharing.

4. **Discussion of Berlin Meeting March 5-8, 2014**
    a. Plenary and symposia were discussed and chairs will be responsible for securing speakers and presentations → to contact in July-August (**see action items**)
    b. Poster session will be held for trainees (PhDs/post-docs) – November 1 deadline
    c. Cultural event planned for one night with dinner
    d. Uli to examine hotels to limit to 3 to promote interactions between members
    e. Brief operational/applied sessions to review the needs for systematic data collection i.e. Minimal Data Set (MDS) & Slims
    f. There was discussion about holding a bioinformatics workshop for genetics investigators
    g. Day 1 (Thursday March 6): symposium + plenary session 4-5 pm + posters and light food & drink
    h. Day 2 (Friday March 7): symposium + evening cultural event (dinner & museum)
    i. Day 3 (Saturday March 8, morning only afternoon departure) : COST business and Management Committee decisions

COST MEETING: Berlin March 5-8, 2014

Wednesday March 5

17:00-19:00
Management Committee Meeting

Thursday March 6

09:00-11:30
Symposium 1: Novel clinical insights into HH (N. Pitteloud)
Spectrum of IHH and new clincial markers (J. Young)
Neonatal gonadotropin therapy (C. Bouvattier/M. Maghnie)
Overlapping syndromes including HH (C. Dode/Dattani/J. Leger/I. Netchine)
Genetic background of constitutional delayed of puberty (L. Dunkel)
Novels genes involved in CHH (**TBA**)

13:00-15:30
Abstract Session

16:00-17:00
Plenary Session: proposed speakers Tom Südhof (Stanford)/Reinhard Jahn (Max Planck Inst)

17:00-19:00
Poster session

Friday March 7

09:00-11:30
Symposia 2: Kisspeptin neuron biology (V. Prévot)
Kisspeptin uncover GnRH pulses in HA (W. Dhillo)
Kisspeptin and epigenetics (S. Ojeda)
Electrophysiology of the Kisspeptin neurons (A. Herbison)

13:00-15:30
Symposium 3: Regulation of GnRH neuron circuits (M. Tena-Sempere)
Role of NO in GnRH (V. Prévot)
Genetics approach to GnRH neuron circuits (U. Boehm)
Genetics of synaptic proteins in GnRH deficiency (N. De Roux)
AMH regulation of GnRH neurons (P. Giacobini)

17:00-19:00
Cultural event (dinner & museum)

Saturday March 8

09:00-11:30
Symposium 4: Novel approaches to study the HPG axis (N. De Roux)
Pituitary development (P. Mollard)
Circadian rythms and endocrine (S. Lightman)
MiRNA and regulation of the HPG axis (M. Tena-Sempre)
Hormonal profiling in mice (D. Steyn, Australia)

11:30-12:00
Meeting closes

5. Review of Aims/Objectives for year 1

a. Establishing rules for database → September meeting (**see action items**)

b. WORKING GROUP 1: Clinical
   i. Finalize Minimal Data Set (MDS) – see notes above
   ii. Finalize & post Patient Materials – Andrew to modify per discussion above and review with WG1 leaders and Patient Advocates
   iii. Reach consensus on treatment and evaluation (idea of Opinion Paper)

c. WORKING GROUP 2: Genetics & Bioinformatics
   i. Nicolas rasied the point of defining variants/mutations and how the goals for developing genetic counseling could be accomplished given the growing complexity
   ii. Brian explained different filtering programs/approaches
   iii. There was discussion of storing exome data in a cloud for collaboration and sharing
   iv. There was a good deal of discussion on the specific national regulations and guidelines for data sharing as well as the Europea n Society of Genetics recently published recommendations for handling exome data
   v. A fall meeting (September, September 6th in Lille - TBA) will be planned to discuss the databse rules, exome data sharing, genotypes, and ethical considerations (**see action items**)

d. WORKING GROUP 3: Basic Science
    i. Discussion touched upon the point raised at the last meeting regarding the importance of identifying canadidates now for basic scientists to be able to generate models/KOs and publish before the end of the COST grant
    ii. Uli pushed the idea of putting together a showcase publication (i.e. junior people would all share co-first and senior investigators would share co-last authorship) with murine models distributed to the various basic groups to contribute to the biology of the clinical and genetic findings.
    iii. The idea was to have genetic groups come to present candidate genes among which several will be selected for further *in vivo* studies by WG3 - this will be the basis for a joint COST publication (**see action items**)

e. WORKING GROUP 4: Training & Education
    i. The idea of having a meeting following ESPE (September 22nd) to further discuss plans for the next training school with Mohammad (**see action items**)
    ii. Review of need to have instructors and students register for e-COST to be reimbursed for training school
    iii. Richard raised the idea of assigning homework assignments to the students prior to the training school
    iv. Without funds to bring patient advocates to the Training School in Prato the possibility was raised of requesting funds to bring them to the March meeting
    v. Focus for next training school would be translational i.e. combined clinical-genetics-basic and a shorter duration (3.5 days)
    vi. Target of 20-25 students
    vii. Idea of possible joint presentations by students & mentors
    viii. Location TBA (i.e. Prato, Les Diablarets, others?)
    ix. The need to identify industry support to cover down payments and additional costs was underscored
    x. Next planning steps for Training School 2014
        1. Set dates for early September 2014
        2. Determine site
        3. Finalize 3.5 day program
        4. Invite speakers
        5. Solicit sponsors

KEY ACTION ITEMS:

1. Link Action website to all websites of COST investigators
2. Nicolas, Leo, & Mohammed to investigate beginning a ESPE working group on puberty
3. Uli & Manuel to discuss and initiate Marie Curie joint PhD training platform
4. Finalize clinical and genetic centers of excellence and patient materials for website
5. Examine rules for using database sent by J. Leger
6. For March Meeting: chairs to contact plenary and symposia speakers (July-August)
7. Next Core WG leader meeting September 6th (Lille) to discuss the database rules, exome data sharing, genotypes, and ethical considerations, opinion papers, next training school, and candidate genes form exome sequencing for WG3