



Meeting Minutes

BM1105 WORKING GROUP LEADERS MEETING: September 6th, 2013 - CHRU, Lille FR

- 1. Nelly reviewed that the 3 main objectives for the meeting:**
 - a. finalizing the plans for the Berlin Meeting in March
 - b. review and edit the charter draft defining rules for using the database
 - c. selecting genes to be used for a joint cross-COST manuscript

- 2. Discussion of Berlin Meeting → March 5-8, 2014 (Uli Boehm) – tentative agenda attached**
 - a. An email will be circulated to all COST members to identify approximate numbers
 - i. 100 rooms (80 singles + 20 doubles) may be reserved (Motel One Hauptbahnhof) this would help promote interaction between participants
 - ii. Size of the meeting – the NeuroCure center is nearby in Berlin and we may have possible walk-in participants
 - iii. Uli has arranged for a cultural event – a private, after-hours, guided museum tour (the fee would be 2'600€ for 125 people – approximately 20€ each)
 - b. Young investigators are strongly encouraged to come: 500€ travel grants will be provided, both clinical and basic trainees from COST groups should submit abstracts for oral presentations and/or posters → deadline November 1 (decisions by December 1). Topics ranging genetics, bioinformatics, GnRH biology, and clinical studies/challenging cases (i.e. diagnosis or treatment).
 - c. Berlin meeting will be a joint Management Committee meeting, Scientific Meeting, and Training School (tentative agenda attached)
 - d. Additional discussion included a planned Bioinformatics workshop for the fall of 2014 in Lausanne combined with Management Committee meeting

- 3. Budget: (Nelly Pitteloud)**
 - a. The proposed budget for the next funding period was reviewed and discussed (attached)
 - b. This is to be sent to the Management Committee for review and comment

- 4. Training School Review (Vincent Prevot):**
 - a. Vincent reviewed the training school held in Prato: positives included the setting and the quality of the speakers/faculty, drawbacks included that a number of students were not specific to GnRH biology, a week was deemed too long, and the financing was challenging
 - b. There was discussion about adding more clinical focus to the next training schools
 - c. 2014 Training School will be folded into the planned Berlin Meeting
 - d. 2015 Training School → The final training School will be targeted for Easter Break 2015 in Prato. This would be a joint venture between Monash and our COST action. We will initiate seeing additional funds to support the program now (including support for bringing patient advocates). The framework will be to pair 20-30 young basic scientists with 20-30 young clinicians to make the focus on collaborative translational research.

- 5. Website, Publicity, SLiMS Database: (Virginia Hughes)**
 - a. WEBSITE: Virginia reviewed the website and the pages in development - to be launched soon
 - i. Clinical Centers of Excellence (launched)

- ii. Genetics Centers of Excellence (launched): - Nicolas to write a brief blurb about genetic screening for KS and introduction
 - iii. We still needs content for genetic studies (email sent soliciting studies - no responses were received)
 - iv. WG3 has been updated with photos → WG3 Leaders to generate 20 keywords for searching
 - v. Gene curation - access to Cardiff gene database is pending. Nicolas raised the question about what genes should be included and overlapping syndromes? This was yet to be finalized.
- b. PUBLICITY: This is a key metric to evaluate the Action
- i. Participants are reminded to link their individual website with the COST action website
 - ii. Notify Virginia of publications involving participants form 2 COST countries.
 - iii. Acknowledge the COST Action BM1105 in manuscripts i.e. “This work was supported by COST Action BM1105”
 - iv. Nicolas suggested it would b beneficial to publish what we have done so far
 - 1. Goal is to get something published (and referenced in PubMed) by the Berlin Meeting
 - 2. Nicolas to contact Philippe Bouchard about potential to publicize via ESE and to contact the editor of Hormone Research in Pediatrics – October 1
 - 3. Manuel to contact Bob Millar about Neuroendocrinolgy – October 1
 - v. Structure: what is the action about?, what are objectives?, what has been done?, and what are the future directions... - draft to be circulated mid October
- c. DATABASE: SLiMS
- i. Virginia showed representative screenshots of what the database looks like and how queries will look online
 - ii. The database belongs to the COST action but SLiMS was purchased by the Pitteloud Lab and it is hosted at the CHUV (who has contributed 20'000 CHF per year for the server/tech support to support it)
 - iii. The importance of local ethics approval was underscored
 - iv. There is a critical need to link the SLiMS unique identifier with the identifier of the local biobank to ensure that there are no “lost” cases
 - v. Further research needs to be done regarding the European Ethics Standards for the data handling and confidentiality as it relates to de-identified/anonymous registries

6. Charter for utilizing and querying the patient registry (Nelly Pitteloud)

- a. Nelly reviewed the charter for the database – which was a prior registry charter provided by Juliane Leger
- b. It was proposed that the governance of the access/use of data be overseen by a panel that includes the WG leaders and Juliane Leger (given her expertise)
- c. Members will sign a confidentiality agreement protecting he intellectual property and ideas of those who submit proposals for accessing/using the data
- d. Summary statements will be provided on the website outlining the number of cases provided by each county
- e. Review of applications will occur at 2-month intervals and responses will be given within 2-weeks
- f. Authorship decisions were discussed – those contributing cases will be co-authors, we will investigate citing COST as an author (i.e. consortium) so that all are credited but only contributing authors are listed on the main author listing.

7. Candidate Genes for an action-wide collaborative manuscript (Nelly Pitteloud)

- a. All present agreed that it will be a COST-wide paper with authorship in alphabetical order
- b. Ideas for candidate genes will come from genetic studies on patients (exome sequencing) to be validated in animal/cell systems or from basic scientists who wish to see if human mutations exist in the exome sequencing of patients
- c. Groups shared their experience with exome sequencing and how many exomes they have screened
- d. Nelly reviewed an Excel list of candidates from her exome sequencing and the bioinformatic approaches to identify them which stimulated discussion
- e. Next steps:
 - i. Marco Bonomi to forward the list of candidates from the Italian Group
 - ii. Other investigators will see if candidates were also observed among their exome results
 - iii. Lists will be reviewed and initial expression studies will be performed
 - iv. Meeting by Skype early November to select best candidates from 3 different pathways for expanded Sanger sequencing screening among CHH cohorts (familial cases)

ACTION ITEMS:

Berlin Meeting:

Response requested for attendance by September 23
Symposium chairs to invite speakers
Plenary speakers (2) to be invited and confirmed
Launch call for abstracts – November 1
Travel grants decided – December 1
Identify additional industry support for the meetings (2014-2015)

Budget:

Submit for MC committee review/approval

Website:

Link Action website to all websites of COST investigators
Manuel & Uli to generate 20 keywords for searching profiles
Post patient advocacy information

Publicity:

Nicolas to contact Philippe Bouchard (ESE) publicity & Hormone Research in Pediatrics – October
Manuel to contact Bob Millar about Neuroendocrinology – October 1
Paper draft to be circulated - mid October (Andrew to draft)
Nicolas, Leo, & Mohammed to inquire about starting a ESPE working group on puberty (Paris Mtg)
Uli & Manuel to investigate Marie Curie joint PhD training platform - pending Horizon 2020 (Paris Mtg)

Patient Registry:

Document circulated for review, comment, and approval
Governing body to be finalized

COST Candidate Gene Project:

Italian candidate genes to be added and list to be circulated
Expression studies then decision of top 3 → November
Identify who can contribute aspects to the joint paper



COST MEETING: Berlin, March 5-8, 2014

- **Management Committee Meeting**
- **Scientific Meeting**
- **Training School**

Wednesday, March 5 : Business Meeting

16:00-19:00 Management Committee Meeting

Thursday, March 6 : Day 1 of Scientific Meeting & Training Program

09:00-11:30 WELCOME & SYPOSIUM 1 “Novel clinical insights into HH”
 1. Spectrum of HH and new clinical markers
 2. Syndromes overlapping with HH
 3. Genetic background of constitutional delayed of puberty
 4. Exome Sequencing in Clinical Practice
 5. Future Directions: What’s Next?

13:00-15:30 Oral Communication Session

16:00-17:00 Plenary Session: proposed speaker Reinhard Jahn (Max Planck Inst)

17:00-19:00 Poster session (Clinical Cases, Genetics, GnRH biology) with food & drinks

Friday, March 7 : Day 2 of Scientific Meeting & Training Program

09:00-11:30 Symposium 2: “Kisspeptin neuron biology”
 1. Kisspeptin uncovers GnRH pulses in HA
 2. Kisspeptin and epigenetics
 3. Electrophysiology of the Kisspeptin neurons
 4. TBA
 5. Future Directions: What’s Next?

13:00-15:30 Symposium 3: “Regulation of GnRH neuron circuits”
 1. Role of NO in GnRH
 2. Genetics approach to GnRH neuron circuits
 3. Genetics of synaptic proteins in GnRH deficiency
 4. AMH regulation of GnRH neurons
 5. Future Directions: What’s Next?

16:00-17:00 Plenary Session: proposed speaker Steven Kliewer (UT Southwestern)

19:00-21:00 Cultural event (dinner & museum)

Saturday, March 8: Day 3 of Scientific Meeting & Training Program

09:00-11:00	Symposium 4: "Novel approaches to study the HPG axis" <ol style="list-style-type: none">1. Pituitary development2. Circadian rhythms and the endocrine system3. MiRNA and regulation of the HPG axis
11:00-12:00	Summation – Panel Discussion: Future directions and horizons
12:30	Close of Meeting