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PROCEEDINGS FROM MEETINGS

Minutes of the European POmpe Consortium (EPOC) Meeting

March 27 to 28, 2015, Munich, Germany

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In September 2014, twenty-two experts in the field of Pompe disease from nine European countries assembled in Naarden, The Netherlands, for a workshop on Pompe disease, the 208th ENMC International Workshop "Formation of a European Network to develop a European data sharing model and treatment guidelines for Pompe disease" (1). Here, the European POmpe Consortium (EPOC) was funded. To continue our work, the consortium met for the second time, March 27 to 28, 2015 in Munich, Germany.

Friday March 27, 2015
Introduction and summary

The meeting was opened by Benedikt Schoser who welcomed all attendees to Munich and summarized the agenda.

Ans van der Ploeg gave an overview of the first meeting of the consortium in September 2014 in Naarden. At this first meeting the EPOC consortium was founded, a minimal dataset was agreed for adult patients, as well as criteria for starting and stopping enzyme replacement therapy (ERT) in adult patients. She elaborated shortly on the scope of the consortium: i.e. via concerted actions of the partners we aim to i) improve prospects for patients by combining efforts on understanding the disease process, existing therapies and development of innovative treatment strategies; ii) to provide guidance on treatment, care and outcome measures, iii) to harmonize the views on access and reimbursement of therapies in Europe in order to create equal chances for patients with Pompe disease across Europe.

After the first meeting in Naarden, the steering group for the Naarden and Munich meetings proposed the name European POmpe Consortium (EPOC). All attendees agreed to adopt this name.

Michelle Kruijshaar presented the progress on publications resulting from the Naarden meeting. A lay summary was posted on the ENMC website in September 2014. The full workshop report is now available via the Journal "Neuromuscular disorders" (1). Finally, a separate manuscript is being prepared to describe criteria for starting and stopping ERT in adult Pompe patients. The proposed start and stop criteria for adult patients were reviewed. A few remarks from the group were made regarding the phrasing of certain stop criteria, which will be incorporated in the manuscript that is being prepared.

Consortium agreement

Benedikt Schoser presented a proposal of the steering group for the organizational structure of the consortium.

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This was discussed in detail. The following was agreed:

1. General Assembly (GA):
   a. Is comprised of all approved members of the consortium.
   b. Meets at least every one or two years.
   c. At the meeting, members can vote board members and approve new members of the consortium.
2. Members:
   a. Have to be professionals (clinicians and scientists) with experience on Pompe disease working in Europe (or Switzerland).
   b. Have voting rights.
   c. One IPA representative shall be a member, i.e. with voting rights.
3. Board:
   a. Acts for the GA.
   b. Is comprised of 5 members: a chair, a co-chair, a secretary/treasurer, and two general board members.
   c. Board members are elected by the GA. Every 2 years, 2 board members shall be substituted. It is possible to be re-elected once.
   d. Proposals for new members of the Consortium will be put to the board before being agendized for the GA meeting.

Next, an election was held for the first steering board by secret ballot (i.e. on paper not by raising hands). The first elected board is:

- Benedikt Schoser (chair)
- Ans van der Ploeg (co-chair)
- Pascal Laforêt (secretary)
- Antonio Toscano
- Pieter van Doorn

Finally, a discussion followed on the need for, and content of, a consortium agreement and statutes for the consortium. Example documents were circulated in advance of the meeting. It was agreed that a consortium agreement should be made first; then the EPOC statute should be provided. The content should be similar to the elements included in the example document, specifying the organizational structure of the consortium, and specifying that at present there is no fee attached to membership.

How to collect data

To further our understanding of the disease processes and existing therapies the consortium wants to/ will collect data from the different countries. Antonio Toscano and Pieter van Doorn chaired the session on how data could be organized. They presented a number of decisions that have to be made when considering data sharing. The first point, whether or not there should be a central database, received a lot of discussion. Sabrina Sacconi expressed concern that data would be collected without sufficiently important research questions driving this data collection. Other members from Italy and France supported the need for data collection to be driven by specific research questions. Another point that was made was the importance of having a clear publication policy that includes all centers that provide data. Last, it appeared that rules regarding sharing data and requirements for ethics differ between countries. As next steps it was decided to first focus on the research questions ("in a project by project" way), and that an inventory of the rules and requirements for the different countries should be made.

National presentations and scientific ideas

Pascal Laforet and Antonio Toscano chaired the session on national presentations and scientific ideas. Presentations by the individual countries (of about 5 minutes) were given, including the total number of patients, number of centers and distance to the centers, as well as the main clinical and scientific interests in these countries.

Adding up the entire patient numbers of the 9 countries (Italy, Spain, France, the Netherlands, the UK, Denmark, Switzerland, Austria and Germany), our consortium will cover at least 1250 Pompe patients of all ages. This gives a prevalence of Pompe disease of about 1 in 283,000 (1250 / 354 million inhabitants; given that all the patients included in the presentations are still alive).

Presentations from the UK and the Netherlands indicated that infusions of myozyme are commonly done in the home setting. Other countries were interested in the experience with home infusions and it was suggested that it would be helpful if these experiences were published.

Saturday March 28, 2015 Discussion of possible grant proposals

Ans van der Ploeg informed all participants of an E-Rare proposal that has been sent out. E-Rare aims to stimulate international, interdisciplinary projects. There are several limitations to the proposal, including which countries are eligible for participating. A proposal was sent in under the lead of Ans van der Ploeg, supported by Pascal Laforet, Benedikt Schoser, Antonio Toscano, and Beril Talim (all E-Rare eligible countries) for a project called PrEDIcTION (Pompe disease EDiting by Treatment OptimizatION). The proposal has successfully passed the first evaluation round, and is presently in the second round of the E-Rare evaluation process.

Other calls that may be interesting for the consortium include: i) COST program of EU (funds meetings); ii) apply for an AMDA grant for a specific EPOC project, iii) German organization for rare disease database (model registry).

Progress on workgroups

Andreas Hahn and Alexander Broomfield presented the progress of the infant workgroup on behalf of the four countries (Germany, UK, Italy and The Netherlands). The workgroup has chosen to focus on a specific research question concerning the dosing regimens that are applied in Europe. Data from two of the four participating countries has already partially been collated and all four countries have already made some effort to collate their national data. A total of around 130 infant patients are expected to be available after combining these results. The preliminary data show that different dosing regimens are applied in Europe and gave a first impression what can be done with the concerted action of the consortium.

Nadine van der Beek presented the progress on the R-PACT project. A traditional assessment scale does not always have interval properties (i.e. the difference between response options is not necessarily the same and/or a sum score of 3 may identify people with different clinical problems). The Rash method can help to develop scales with interval properties. In Rotterdam, a scale to measure activity and participation of Pompe patients was developed using this methodology. In the original study it was developed and validated for the Dutch and English language. For the present project it has been translated into French, German and Italian, and other translations can be made. Pascal Laforet, Eugen Mengel and Jordi Díaz Manera have indicated that they are interested to participate in this project.

Nadine van der Beek also briefly presented the Rasch-built MRC score. This has only 4 levels, compared to the traditional MRC score which has 6 levels. Some centres may be interested to assess both MRC scores, but it was decided that at least the traditional MRC score remains part of the minimal dataset.

Next steps

The next steps of the network will be to discuss which specific research projects the network will focus on and how data can be shared for these projects. The consensus on criteria for starting and stopping ERT in adult patients will be harmonized and published. Finally, outcome measures and start and stop criteria for infants and children will be discussed.

Next meeting

It was proposed that a meeting of about 3 hours would be held at the World Muscle Conference in Brighton (September 30, 2015). The steering board will send out a meeting invitation and organize a meeting room in Brighton. Pascal Laforet offered Paris as the location for the next EPOC meeting in 2016.

List of participants (European Pompe Consortium, EPOC, members):

France: Pascal Laforêt *(Paris)*, Claude Desnuelle *(Nice)*, Sabrina Sacconi *(Nice)*
Spain: Ignacio Pascual Pascual *(Madrid),* Jordi Díaz Manera *(Barcelona)*
Italy: Antonio Toscano *(Messina),* Tiziana Mongini *(Turin),* Corrado Angelini *(Venice),* Giancarlo Parenti *(Naples)*
UK: Mark Roberts *(Manchester),* Alexander Broomfield *(Manchester)*
Switzerland: Kai Rösler *(Bern),* Oliver Findling *(Aarau)*
Germany: Andreas Hahn *(Giessen),* Eugen Mengel *(Mainz),* Benedikt Schoser *(Munich),* Wolfgang Müller-Felber *(Munich),* Angela Schüller *(Munich),* Federica Montagnese *(Munich/Messina),* Stephan Wenninger *(Munich)*
Netherlands: Ans van der Ploeg *(Rotterdam)* Nadine van der Beek *(Rotterdam),* Pieter van Doorn *(Rotterdam),* Michelle Kruijshaar *(Rotterdam),* Pim Pijnappel *(Rotterdam)*

Members with cancellation for the Munich meeting:
Denmark: John Vissing / Nicolai Preisler *(Copenhagen)*
Turkey: Beril Talim *(Ankara)*
Austria: Christian Eggers *(Linz),* Thomas Stulnig *(Vienna)*
UK: Ashok Vellodi, Robin Lachmann, Ros Quinlivan *(London)*
Belgium: Peter van den Bergh *(Brussels)*
Switzerland: Thomas Hundsberger *(St. Gallen),* Marianne Rohrbach *(Zurich)*
Germany: Ursula Plöckinger *(Berlin),* Peter Young, Matthias Boentert *(Münster),* Rudolf Kley *(Bochum),* Cornelia Kornblum *(Bonn),* Julia Hennermann *(Mainz)*
Patient representative (IPA): Thomas Schaller *(Weingarten)*

References

1. Schoser B, Laforêt P, Kruijshaar ME, et al. 208th ENMC International Workshop: Formation of a European Network to develop a European data sharing model and treatment guidelines for Pompe disease Naarden, The Netherlands, 26-28 September 2014. Neuromuscul Disord 2015;25:674-8.