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**Update #17 on the AIDNPC clinical programme**

*(arimoclomol in treatment of Niemann-Pick disease type C)*

**Interim information bulletin (following the Sept. 28th call)**

SUMMARY

* 001 Study: Conclusions and initial learnings
* 002 Study: Status and timelines
* Under-2-year-olds in study
* Key assessment of study outcome
* Orphazyme to present at INPDA meeting

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***The AIDNPC clinical trial programme consists of two studies:***

* ***The ‘-001’ Observational Study, recruiting closed.***
* ***The ‘-002’ Interventional Study, recruiting closed.***

001 Study: Conclusions and initial learnings

The study was recently completed in May 2017. A total of 35 patients were enrolled across 12 sites in 8 countries.

The primary objective of the 001 study was to characterize the individual patient’s disease progression profile through clinical, biological and quality-of-life measures recorded prospectively, as well as the historic disease information collected from patient medical records.

The 001 study has already provided us with some valuable learnings: The NPC-CSS disease score were completed by patients with a good compliance, the skin biopsies and blood tests have been valuable for development and conformation of biomarkers, and we found that two-thirds of patients were treated with miglustat as part of their standard of care. Once all data have been analysed, we expect to gain insight about biomarkers and how the disease evolves over time in greater detail

002 Study: Status and timelines

The first patient in the 002 study (a phase 2/3 trial) enrolled in June 2016, and the last patient enrolled in May 2017. A total of 50 patients enrolled and were randomized, of which a total of 27 patients from the 001 study were rolled over into the ongoing 002 study. We expect to have results from this study in the autumn of 2018. As each patient has his/her last visit in the blinded part of the study, he/she will be offered to enter into an open-label extension of the study where all patients will receive only the active substance, arimoclomol, as treatment (i.e., no one will receive placebo during the open-label extension of the study).

The following sites are participating in the 002 Study:

Copenhagen, Denmark

Birmingham, UK

Great Ormond St, UK

Mainz, Germany

Münich, Germany

Paris, France

Montpellier, France

Barcelona, Spain

Warsaw, Poland

Udine, Italy

Rome, Italy

Rochester, MN-USA

Oakland, CA-USA



Under-2-year-olds in study

Orphazyme is planning an arimoclomol open-label sub-study in NPC patients below 2 years of age to assess safety, tolerability and pharmacokinetics as well as to explore effectiveness in this age group.

Key assessment of study outcome

Orphazyme is re-assessing the NPC Disease Severity (NPC-CSS) scoring tool as it was originally published by Yanjanin et. al in 2010, which includes 9 domains of assessment. We are currently concluding efforts (i.e., discussions with regulators; a survey of expert physicians, patient organisations and NPC caregivers; a responder analysis; and a rater-reliability study) to establish a simplified 5–Domain NPC-CSS scoring tool, with the intention to improve tracking and scoring of NPC patients in the future. The 5 remaining domains in this NPC-CSS scoring tool will be ambulation, fine motor skills, swallowing, speech and cognition.

Orphazyme to present at the INPDA meeting in October

Orphazyme will present and be present at the 5th Biennial INPDA Face-to-Face Meeting in Toronto, Canada (13th–15th October 2017).

Clinical design overview

The graphic below illustrates the design of the 002 Study, including number and timing of patients visits to the sites. An escape route is provided for patients that experience an unacceptable rate of progression of the disease.



***We encourage the sharing of above information with the patient community.***

Next call:

The next AIDNPC call is schedule for Thursday December 28th 2017 at 15h EDT.