# ERN ITHACA Cooperation with "Prenatal HPO Working Group"

Invitation to a Webinar

*08/06/2021 at 14:00*

**Abstracts**

# Extending the terms of the Human Phenotype Ontology to include prenatal phenotypes – **Peter Robinson**

- The Human Phenotype Ontology (HPO) provides a standardized vocabulary of phenotypic abnormalities encountered in human disease. The HPO is planning on extending the HPO to add terms to comprehensively cover prenatal disease and will develop computational disease models (annotations) for diseases with prenatal manifestations. These resources will be made freely available to all under the HPO license (https://hpo.jax.org/app/license). In this presentation I will present a short introduction about the HPO and will discuss our proposed strategies for developing prenatal terms. We are inviting colleagues to participate in this project by working with us to (1) generate a comprehensive representation of prenatal phenotypic features in the HPO; (2) Specify the diagnostic modalities that can be used to ascertain these features; (3) to enable integration with sonographic software for partial automatic generation of HPO terms; and (4) to develop HPO disease annotations for prenatal manifestations of Mendelian and other diseases. We are planning a series of community workshops over the coming year. Additionally, the HPO team is participating in a project of the Fetal Sequencing Consortium (https://www.columbiaobgyn.org/fetal-sequencing-consortium) to improve database resources for prenatal medicine and we are seeking collaboration in the community. I will briefly present our plans to use Global Alliance for Genomics and Health (GA4GH) standards to enable data exchange with the Fetal Sequencing Consortium.

# European collaboration project – **Gijs Santen**

- Increasingly, exome sequencing is performed after the identification of ultrasound anomalies during pregnancy. Every geneticist involved in fetal exome sequencing will have experienced the lack of data out there to counsel patients. Important knowledge gaps include (1) prenatal phenotypes of genetic disorders, (2) the posterior risk of a genetic disease when exome sequencing is negative, (3) the optimal analysis strategy to balance the risk of VUS and incidental findings versus the risk of a missed diagnosis, and (4) lack of research into uniquely prenatal disease genes. Many of these knowledge gaps can be solved relatively easily by collecting clinical data (genetic data as well as HPO-coded prenatal phenotypes). The more we collaborate, the quicker we can collect data and answer these questions. Therefore, in the past 1.5 years we have been trying to get some funding for a collaborative project in the Netherlands and we have finally succeeded. I will present the outline of the study we hope to start soon and hope to spark interest for a larger European collaboration with many of you to increase the speed of data collection even further.

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- For expressions of interest to join the working group, please fill this Form


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