PRESS RELEASE

Herman Verrelst
CEO Cartagenia
herman.verrelst@cartagenia.com

+32 16 40 40 66

www.cartagenia.com

Cartagenia partners major UK study on new prenatal diagnosis technology

Leuven, 19 June 2012 - Cartagenia today announced that it will be making its cloud-based BENCH platform for genomic variant storage and analysis available to the UK EACH (Evaluation of Array Comparative Genomic Hybridisation in Prenatal Diagnosis of Fetal Anomalies) research project, a ground-breaking project in the field of prenatal diagnosis. The lead investigators on EACH are Professor Stephen Robson (Newcastle University), Professor Lyn Chitty (University College London) and Dr John Crolla (Salisbury).

With array CGH gradually replacing conventional karyotyping in routine clinical practice in postnatal genetic diagnosis, the question is now whether it should replace conventional karyotyping in prenatal diagnosis, particularly in cases of fetal abnormality. To answer this question, a consortium of several English Fetal Medicine Units, Clinical Geneticists, and leading genetic laboratories will be conducting a 3-year study.

The EACH study, funded by the Efficacy and Mechanism Evaluation Programme will evaluate the potential for array CGH for the prenatal diagnosis of fetal anomalies, determining whether it is an acceptable, cost-effective and robust method to detect more clinically significant chromosomal imbalances in fetuses when compared with conventional karyotyping.

Major congenital anomalies are a leading cause of neonatal death and disability, resulting in substantial emotional and economic burden for families and society. Chromosomal anomalies are responsible for about a quarter of major fetal anomalies and screening shows 3% of pregnant women to be at high risk of their fetus having a chromosomal anomaly.

Cartagenia's cloud-based BENCH platform will allow researchers to collect genetic and clinical data on the study cohort. On top of its existing tools for storing large sets of patient-centric data, filtering and interpreting genetic

Herman Verrelst
CEO Cartagenia
herman.verrelst@cartagenia.com

+32 16 40 40 66

www.cartagenia.com

variants, and annotating clinical and administrative information, Cartagenia will configure the platform towards EACH needs by setting up clinical data collection user interfaces in the form of user-friendly forms for collecting patient, maternal and paternal demographics, lab information, but also important clinical information such as indications for testing or relevant fetal abnormalities. These data will be anonymised and managed according to the standards required by UK and European legislation. Thanks to Cartagenia's data entry tools based on state-of-the-art phenotype nomenclatures such as HPO and flexible clinical forms, the database infrastructure can be rapidly tailored to EACH needs.

John Crolla, who is leading and co-ordinating the molecular cytogenetics aspects of the EACH project expressed his pleasure at having Cartagenia on board: "For us Cartagenia is the obvious partner to assist us in this multi-centre project. They have profound expertise in the collection, analysis and interpretation of array CGH results and clinical patient information, and have a proven track record working with other research consortia, such as the US-based ISCA Consortium or the European FP7 CHERISH programme".

***

About Cartagenia

Cartagenia supplies diagnostic knowledge, software and database systems, and related services to genetic labs and clinicians, enabling them to perform clinically relevant genetic analyses quickly and efficiently, and offer patients and carers high-quality genetic interpretation and counselling. (www.cartagenia.com)

About EACH

EACH has been funded via the Efficacy and Mechanism Evaluation Programme (EME) co-admistered by the UK's Medical Research Council and the National Helath Service's National Institute of Health Research (NIHR). The project aims to compare 1,000 conventional karyotypes and array CGH results following invasive prenatal diagnosis (amniocentesis or chorionic villus sampling) when a

Herman Verrelst
CEO Cartagenia
herman.verrelst@cartagenia.com

+32 16 40 40 66

www.cartagenia.com

fetus has been diagnosed with an ultrasound abnormality either in the first trimester (nuchal tranlucency ≥3.5 mm) or when a structural abnormality is diagnosed in the first or second trimester. Several Fetal Medicine Units will be recruiting patients into the study from May 2012 and the array CGH will be carried out in a limited number of regional cytogenetic laboratories using the International Standard for Cytogenomic Array (ISCA) consortium's 60k oligo array CGH design.

***

Contact:

CARTAGENIA
Herman Verrelst
CEO Cartagenia
Technologielaan 3
3001 Leuven
T +32 16 40 40 66
E herman.verrelst@cartagenia.com
www.cartagenia.com