**THAMEUS Project**

**The Next-Generation Newborn Screening**

**Human Genetics Department – Pediatrics Department**

**GIGA-Research Center**

**University Hospital of Liège – University of Liège**

**Position open**:

Post-doctoral researcher for a genomic medicine project

Development of Next-Generation Sequencing protocols for newborn screening

The Human Genetics and Pediatrics departments of CHU Liège, in association with GIGA-Research Center, launch a genomic medicine project in newborn screening.

Current advances in the treatment of rare diseases lead newborn screening programs to consider a constantly growing number of disorders. For most of newly developed treatments, early or pre-symptomatic administration of the therapy is strongly correlated with higher life expectancy, avoidance of severe disabilities, and fewer complications during the course of life.

Currently, newborn screening programs face limitations in identifying disorders with no metabolic or endocrine biomarkers. In this context, the Thameus project aims to establish a dynamic genomic neonatal screening, which not only detects the commonly listed disorders but also considers the inclusion of new genetic diseases in a timely and economical fashion, in order to accelerate clinical development and maximize the effect of innovative new therapies.

**Contract**:

Project-based contract.

A two-year contract will be offered with possible extension for at least 2 years.

**Duties**:

* To participate in the development and validation of a genomic sequencing platform for newborn screening (from DNA extraction of dried blood spots to variants reporting) with respect to clinical quality standards.
* To set up bioinformatic protocols, in collaboration with our bio-informatic team, for variants, indels and CNVs detection, filtering and interpretation.
* To manage heterogenous databases (newborns, variants…) and registries.
* To organize and/or participate in molecular boards for variant interpretation and clinical reports.
* To integrate genomic data with biochemical results, when appropriate.
* To collaborate with clinicians for gene selection, data interpretation and panel evolutions (gene and variants).
* To review and publish lab SOPs.
* To ensure compliance to quality standards set by regulations, procedures and mission.
* To support science communication efforts including conferences and writing articles.

**Qualifications and abilities**:

* Post-doctoral scientist with experience in Next-Generation Sequencing in the field of human genetics, preferably constitutional genetics, and in a medical environment.
* Good knowledge of the bio-informatic tools for variant calling and interpretation. Ability to collaborate with the bio-informatic team for specific developments.
* Ability to work with a multidisciplinary team and to interact with all the partners from the hospital and the GIGA-Research unit.
* Ability to organize and prioritize work duties and to function as a team-member; an experience in project management would be a plus. Ability to encourage positive dynamics within the team.
* Good written and oral communication skills.
* Language skills: knowledge of French and English is mandatory.

**Applications**

Applications should be sent before April 30th 2020 to

* + Prof Laurent SERVAIS : laurent.servais@paediatrics.ox.ac.uk
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