Centre de recherche
CHU Sainte-Justine
Le centre hospitalier universitaire mère-enfant

Université de Montréal

Genetic Study of Congenital Scoliosis

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Description du projet :

Congenital scoliosis is a condition characterized by a spinal curvature due to vertebral malformations and affecting about 1 in 1000 live births. In many cases, these vertebral anomalies are associated with other malformations (cardiac, gastrointestinal, etc.). While some patients receive a diagnosis of a genetic condition explaining the vertebral anomalies, a majority of them do not have any explanation for it.

The aim of this project is to enhance knowledge about the role of genetics in the development of congenital scoliosis. More specifically, the objective is to identify specific mutations associated to this condition by using exome sequencing.

To achieve this, the first step is to perform a review of medical records of patients treated at CHU Sainte-Justine for congenital scoliosis. Information is thus obtained about other associated malformations, genetic diagnoses and genetic test already performed. This information will be used to select candidate patients for exome sequencing. These candidates would be patients without genetic diagnoses explaining their vertebral malformations, and significant vertebral malformations.

Then, the next step will be to enroll patients for exome sequencing. They will first be contacted by phone to evaluate their interest in participating in this project. According to their interest, they will next be met to explain more precisely the project and to sign the consent. Finally, blood samples will be collected to perform exome sequencing. It is unlikely we will have the data in time for the summer trainee to participate in the analysis, but she might be able to if she does future internships in the laboratory. IRB approval has been obtained for both parts of this project.

This project is important to provide information about a condition the causes of which are often unknown. Identifying new genes associated to congenital scoliosis would first allow a better understanding of the development of the vertebrae. Also, vertebral malformations have a significant impact for the patients, especially if they are associated to other anomalies. To

eventually provide a genetic diagnosis in these patients could improve their management because associated problems could be anticipated, and familial counseling could be provided.

Mots clés :

Maladies génétiques, génétique médicale, scoliose

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