A genetic database can help further our understanding of the inheritance patterns of congenital heart disease

childhood disability LINK

Summary

Medical and surgical treatments for congenital heart diseases (CHD) have advanced greatly in recent times. However, the underlying causes remain a mystery. Scientific studies suggest that inheritable factors, including genes and systems that control their expression, play a major role. In order to further our understanding of these genetic pathways, researchers have collected a large amount of genetic data from individuals and their families affected by CHD all over Quebec. After three years of data collection, the gene database is large enough to run analyses. These results will further our understanding of the genetic mechanisms behind these diseases. The researchers in this group targeted families with at least two members affected by CHD. Genetic and phenotypic information were collected from 1603 participants from 300 families, 169 of which had more than one affected member. Equivalent data was collected from 87 healthy participants from 28 families and from 52 healthy adults. All phenotypes were analyzed by electrocardiogram, and family histories were also logged. As this is an open-ended study, data from additional congenital heart disease patients and their families will continue to be added to the collection.

What families should know

Congenital heart defects make up the largest category of malformations in Canadian children and are a significant cause of death in infants under one year old. However, improved detection, intervention, and care strategies are now allowing more individuals born with these defects to survive into adulthood and have children of their own. Since congenital heart disease has a sizeable inheritable component, there is a risk that these individuals pass on a defect to their children. Thus, a better understanding of the genetic factors that predispose an individual to congenital heart disease is essential to not just treatment but also prevention

What practitioners should know

The genetic database that has been compiled by this research group presents an ideal platform for examining the inherited causes of congenital heart disease. The database logs information mostly from individuals in families where more than one individual is affected by congenital heart disease, which makes it useful for studying how genetic linkage, or the co-inheritance of certain genetic factors, influences inheritance. Now, after three years of data collection, the database is finally large enough to accommodate meaningful studies of genetic linkage. The database will continue to be augmented with information from more patients, but it has already shown that a certain type of heart defect, left ventricular outflow tract obstruction (LVOTO), is strongly linked to the inheritance of two or more genes.

Reference

Dubé, M., Bigras, J., Thibeault, M., Bureau, N., Chetaille, P., Richter, A., et al. (2011). Design and rationale of a genetic cohort study on congenital cardiac disease: experiences from a multi-institutional platform in Quebec. Cardiology in the Young, 21, 656-664.