Medix Prize of the Minerva Foundation 2019

RESEARCH GROUP FROM THE UNIVERSITY OF HELSINKI AWARDED FOR RESEARCH INTO THE GENETIC BASES OF CHILDHOOD-ONSET CARDIOMYOPATHIES

The Minerva Foundation's Medix prize for research in clinical medicine has this year been awarded to Academy Professor **Anu Wartiovaara's** research group at the University of Helsinki.

The group, working in cooperation with the group of paediatric cardiologists headed by Docent **Tiina Ojala**, was awarded for their research into the genetic bases of severe childhood-onset cardiomyopathies. The results of the research can be utilised in diagnostics, genetic counselling, the development of therapeutic strategies and treatment decisions.

Being less studied than adult cardiomyopathies, the genetic background of paediatric cardiomyopathies has been poorly understood. Childhood cardiomyopathies are typically severe and may require cardiac transplantation. Often the initial phases of the disease are life-threatening, but sometimes the disease may spontaneously improve by school-age, in which case intensive conservative care without a cardiac transplantation is the best option.

As both disorders – the terminal and the spontaneously recovering form – are similarly life-threatening at onset, their different prognoses cannot be identified at this stage on the basis of clinical examination alone.

"In Finland, paediatric cardiac transplants are centralised at Helsinki Children's Hospital. We were therefore able to collect an extensive data on paediatric patients who were considered for cardiac transplant due to a severe cardiac disease over a time period of 21 years. The cohort is globally unique. The average age of diagnosis was four months, and 17 children out of 66 had a cardiac transplant," says Paediatric Cardiologist Tiina Ojala.

Next-generation DNA sequencing, molecular studies and protein modelling were carried out by doctoral student **Catalina Vasilescu**.

"The results indicate that the genetic background of severe childhood-onset cardiomyopathies is highly heterogenic. Nearly every family had their own specific mutation. We also discovered new genes contributing to childhood cardiomyopathies," says Academy Professor Anu Wartiovaara.

According to Catalina Vasilescu, the awarded research is an important step towards more personalised treatment.

"We managed to identify a significant number of genetic causes in cardiomyopathies of children, and show that the DNA-diagnosis has an impact on therapeutic strategies. Genetic information helps to predict prognosis and facilitates individualised decision-making, such as who will need a cardiac transplantation and who could be best helped with intensive conservative care without transplantation," she says.

In half of the patients, the mutation was new, occurring during embryonic development and was not found in the parents. In this case, the disease is not hereditary, which means that the parents do not have an elevated risk of having another child with cardiomyopathy. However, if the parents are found to be carriers of the child's gene defect, the risk for getting a second affected child depends on the type of the mutation. The DNA-diagnosis enables genetic counseling.

Also the disease-risk for siblings of a child can be tested, and if a mutation is not found, they can be released from cardiac follow-up.

The Minerva Foundation's Medix prize is a major annual award for excellent Finnish scientific research published as one article during the previous year. The study of Academy Professor Anu Wartiovaara's group was published in the Journal of the American College of Cardiology.

The authors of the awarded article Genetic Basis of Severe Childhood-Onset Cardiomyopathies. *J Am Coll Cardiol*. 2018; 72:2324-2338: Vasilescu C, Ojala TH, Brilhante V, Ojanen S, Hinterding HM, Palin E, Alastalo TP, Koskenvuo J, Hiippala A, Jokinen E, Jahnukainen T, Lohi J, Pihkala J, Tyni TA, Carroll CJ, Suomalainen Wartiovaara A.

The Minerva Foundation celebrates its 60th anniversary this year, and therefore it awarded two Medix prizes instead of the customary one. The other Medix prize was awarded in the field of biomedical basic research to the research group of Professor **Gong-Hong Wei** at the University of Oulu. The group, working in a collaboration with the group of genetic predisposition to cancer headed by Professor of Medical Genetics **Johanna Schleutker** at University of Turku, studied the mechanisms behind aggressive prostate cancer. Both prizes are €20,000.

Invitation:
The research groups receive the Medix prizes and give a lecture on 23 September 2019 at 1.15 p.m. Address: Luentosali 2, Biomedicum, Haartmaninkatu 8, Helsinki, Finland

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