3rd International Conference on
Neonatal Care and Clinical Pediatrics
April 16, 2022 | Webinar
https://clinicalpediatricsummit.conferenceseries.com/

Current Pediatric Research
ISSN: 0971-9032

https://www.alliedacademies.org/current-pediatrics/

Title: The Genetic Aspect Of Human Heart Development Quest To A Personalised Prophylaxis

Krzysztof Piotrowski
Genetic Centre for Family, Szczecin, Poland

Received: May31, 2022, Editor assigned: June 01, 2022, Reviewed: June 10, 2022, QC No. Q-00005;
Published: April 16, 2022 Invoice No. NCCP-000F5

Biography

Krzysztof Piotrowski, a specialist in Clinical Genetics and in Gynecology and Obstetrics also, completed his PhD with a dissertation on fetal echocardiography. Putting his knowledge into practice, he performs about 3500 USG investigations of gravidas annually for prenatal diagnosis. He has published many scientific papers and chapters covering prenatal diagnosis. Having introduced the BACs-on-BEADs TM technology to Polish diagnostics (presently he use the highest quality array CGH by ThermoFisher-Germany), at moreover present he is focused on applying molecular genetics prenatally.

Abstract

Congenital Heart Diseases are the most common malformations both as an isolated form and a part of genetic syndromes. Extraordinarily fast development of molecular genetics confirms that almost all CHD are genetically dependent in terms of microaberrations in different regions of a chromosome or single gene mutations. On the other hand, CHD are an important component of diverse genetic diseases, including monogenic, metabolic and mitochondrial disorders, most often as secondary cardiomyopathies.

The genes participating therein are located nearly on each chromosome, mainly on pathways, along with ligand genes and co-factors, transcription factors or individually. Many mechanism on heart development are based on the balance between apoptosis, proliferation and migration. Crucial genes controlling fetal development, including the creation of heart tube and the forming of left and right ventricular outflow are primary "homeobox" genes grouped in 4 clusters HOX1-4 . Other genes condition the forming of different structures. The key process for activating consecutive genes is methylation. Methyl groups originate from the metabolic cycle of folic acid, where the main gene is MTHFR. However, it is of great importance to know the real FA level, which is not reflected in the serum!

Moreover, in numerous functional disorders, for example the arrhythmia or block, the reason is also genetic, namely the mutation of ion- channel gene placed in 6 chromosomes. Now we now over 1500 mutations.

Many genes of cardiogenesis were identified thanks to the investigation of other genetic disorders, for example PTPN11 gene in Noonan syndrome. The gene is also responsible for the development of pulmonary valves or TBX5 gene in Holt-Oram Syndrome. Presently the most promising method is NGS technology, where we can perform hundreds of mutations at one time.

Heart development is also affected by