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The genetic aspect of human heart development in aspect of prenatal diagnosis

Congenital heart diseases (CHD) 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 mechanisms 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. 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 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 next-generation sequencing (NGS) technology, where we can perform hundreds of mutations at one time. Heart development is also affected by the imprinting (about 30 genes) and the inactivation of the X chromosome in day 21 stage of embryo. We propose, e.g. a practical classification could refer to specific CHD characteristic of particular disorders, which might prove helpful in daily practice because in prenatal diagnosis CHD is often the sole syndrome confirmed by USG scan, which may depend on truly isolated nature or non-specific mild ultrasound co-markers.

Biography

Krzysztof Piotrowski is a Specialist in Clinical Genetics and has completed his PhD with dissertation on Fetal Echocardiography. Putting his knowledge into practice, he performs about 3500 Ultrasonography (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, at present he is focused on applying molecular genetics prenatally. For the last nine years, he was the Manager of Cytogenetic Unit for Pomeranian Medical University, Szczecin, Poland. Since 2012, he has established a new independent genetic centre, DIAGENCo, which includes a cytogenetic and molecular laboratory. For 4 years, he was the Vice President of the Prenatal Diagnosis Section of The Polish Society of Human Genetics. He participates in many investigated programs.

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