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An outline of the genetic aspect of human heart development in prenatal diagnosis

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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 micro aberrations in different regions of a chromosome or single gene mutations, and many formerly recognized as teratogenic are actually gene-dependent. Many mechanisms of heart development are based on the balance between apoptosis, proliferation and migration. The genes participating therein are located nearly on each chromosome, mainly on pathways, along with ligand genes and co-factors, transcription factors or individually. There are at least 80 such confirmed genes. Crucial genes controlling fetal development, including the creation of heart tube and, e.g. the forming of left and right ventricular outflow (LVOT and RVOT), are primary "home box" genes grouped in 4 clusters. Other genes condition the forming of different structures. Moreover, in numerous functional disorders, e.g. the long Q-T conducting, and most intrauterinal fetal and sudden infant deaths, the reason is also genetic, namely the mutation of ion-channel gene placed in 6 chromosomes. Many genes of cardio genesis were identified through the investigation of other genetic disorders, e.g. PTPN11 in Noonan and Holt-Oram Syndromes. Heart development is also affected by imprinting and the inactivation of the X chromosome in the 21st day of development. We propose a classification of genetic pathologies connected to CHD – often the sole syndrome confirmed by USG in prenatal diagnosis. The above genes and mechanisms constitute a mere representation of the complex issue of cardio genesis.

Biography

Krzysztof Piotrowski, a specialist in Obstetrics, Gynecology and Clinical genetics, completed his PhD with a dissertation on fetal echocardiography. Putting his knowledge into practice, he performs about 3,000 USG investigations of gravidas annually for prenatal diagnosis.He has published many scientific papers and chapters covering prenatal diagnosis. Having introduced the BACs-on-BEADsTM 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. Lately, he has founded a new independent genetic centre, DIAGEN.

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