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The genetic background of human neural tube development: Quest to a personalized prophylaxis

Congenital Neural Tube Defects (NTDs) are common malformations both as an isolated form and a part of genetic syndromes. Extraordinarily fast development of molecular genetics confirms that almost all NTDs are genetically dependent in terms of aberrations in different regions of a chromosome or single gene mutations. On the other hand, NTDs are an important component of diverse genetic diseases, including monogenic and metabolic disorders with mutations (often called polymorphism) genes responsible for the condition of the MTHFR gene. 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 NT development are based on the balance between apoptosis, proliferation and migration. Crucial genes controlling fetal development, including the creation of neural tube and the forming of vertebral continuity are primary "homeobox" genes grouped in 4 clusters HOX1-4. Other genes condition the forming of different structures. The most important pathways are Shh, Wnt, FGF, Notch and BMP. These pathways are closely connected with other structures of the body like conus heart, thymus, intestinal tract and skin or symphatic nervous system. The most complicated is closing of column. On the one hand, this process does not depend on one but on numerous genes, especially Pax3, Pax7 and on the other hand, it depends on proper work mainly of Folic Acid Path, as well as Vitamin B_{12} and Choline. However, it is of great importance to know the real FA level, which is not reflected in the serum. Neural development is also affected by the imprinting (about 30 genes) and the inactivation of the X chromosome in day 21st of embryo development. In our daily prenatal practice we are able to find specific NTDs as soon as 12th week of gestation but our target is to confirm if NTDs may be of truly isolated nature or non-specific mild ultrasound co-markers. As you can see above, we have a lot of information and we can prevent many open NTDs, but still affected children are born. It means that our knowledge about it is not yet complete. Presently, we have some possibilities to help the baby in uterus to close peripheral open NT if it's not too big and has isolated nature.

Biography

Krzysztof Piotrowski is a Specialist in Clinical Genetics. He has 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. For the last nine years he was the Manager of Cytogenetic Unit for Pomeranian Medical University, Szczecin, Poland. Since 2012, he established a new and independent Genetic Center named as DIAGEN co., which includes Cytogenetic and Molecular laboratories. He was the Vice-President of the prenatal diagnosis section of the Polish Society of Human Genetics for six years. He also participated in many investigation programs.

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