Short Communication

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# The genetic background of human neural tube development in the aspect of prenatal diagnosis

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## Abstract

Congenital neural tube defects (NTD) are common malformations both as an isolated form and a part of genetic syndromes. Extraordinarily fast development of molecular genetics confirms that almost all NTD are genetically dependent in terms of aberrations in different regions of a chromosome or single gene mutations. On the other hand, NTD 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, skin or sympathetic 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 and Pax7 and on the other hand, it depends on proper work mainly of folic acid path, as well as vitamin B12 and choline. 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 NTD as soon as 12th week of gestation but our target is to confirm if NTD 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 NTD, 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

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## 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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