

Poster

Implication of the non-canonical Wnt pathway in neural tube defects



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ABSTRACT

Motivation: Neurulation is the embryonic process that leads to the development of the neural tube, precursor of the brain and spinal cord. Failures in this process cause neural tube defects (NTD), such as spina bifida and other lethal defects. There are two types of spina bifida, open spina bifida and spina bifida occulta (SBO). The most important form of SBO is lipomyelomeningocele, which is characterized by a subcutaneous lipoma that is generally located in the lumbar or sacral region. A major knowledge in understanding the genetic basis of neurulation has been the discovery of the crucial role of the non-canonical Wnt signalling pathway (Wnt-PCP). In this project, we are going to investigate the role of the Wnt-PCP pathway in the final step of the closing of the posterior neuropore. For this purpose, we will use mutant mice for Vangl-2 gene (Loop-tail), a member of the Wnt-PCP pathway. In parallel, we will carry on genetic studies to correlate these genetic alterations in lipomyelomeningocele patients.

Methods: In the mouse part of this study, standard histological techniques is been used to determine the incidence of caudal malformations in wild-type and Loop-tail heterozygous embryos. In addition, gene expression by in situ hybridization in the whole embryos is been done to analyse candidate genes, as well as at the protein distribution by immunostaining on cryostat sections. In the human study, we are analysing Vangl-2 gene in lipomyelomeningocele patients to detect possible genetic alterations compared to control patients.

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