Abstract:

Meckel-Gruber syndrome (MKS) is rare multisystemic, autosomal recessive hereditary disorder, which appears in different places around the world. MKS is classified as a ciliopathy. These disorders are caused by defects of primary cilium, cell´s signaling organelle, during embryogenesis. Meckel-Gruber syndrome represents the most severe form of ciliopathy in human population. MKS is caused by mutation in several genes, involved in correct formation of primary cilium. Until this day, 13 genes have been confirmed. As a result we distinguish 13 types of MKS. More genes are also included in MKS origin, but they do not define solo type of MKS. They are called MKS-related genes. The syndrome was recognized mainly on the basis of clinical cases. A big amount of cases was described in consanguineous families. The MKS is characterized by occipital encephaloce, polycystic kidney disease and polydactyly of hands and feet. These symptoms are common with all 13 types. Syndrome also goes with disorder of nervous and renal system, face defects or undefined genitals.

Key words:

Meckel-Gruber syndrome, ciliopathy, primary cilium, autosomal recesive, hereditary disease, consanguineous origin, occipital encephaloce, polycystic kidney disease, polydactyly