Zellweger Syndrome

Also called cerebrohepatorenal syndrome, peroxisomal biogenetic disorder, Zellweger spectrum . It is a congenital (autosomal recessively inherited) progressive encephalopathy , which was first described by Hans Zellweger in 1964.

Cause

The essence is a mutation in any of the 12 genes (see *peroxin-1 (PEX1), peroxin-12 (PEX12)*), controlling the proper function of peroxisomal enzymes. The consequence of this mutation is reduced function or a complete deficit of peroxisomes in brain, liver and kidney cells. The function of peroxisomes is to catalyze the conversion of peroxide to oxygen and water by the enzyme catalase. Thanks to which they degrade fatty acids with long chains and break down harmful substances.

Frequency

The most in Canada − 1:12,000 versus the least in Japan − 1:500,000.

Symptoms

Manifested since birth by morphological and functional deviations:

- craniofacial dysmorphia (high forehead, hypoplastic supraorbital arches, flattened nose, small mandible, large anterior fontanelle),
- visual impairment (glaucoma , cataract , nystagmus),
- hearing impairment
- mental retardation,
- hepatomegaly associated with reduced liver function and subsequent jaundice,
- impaired kidney function due to cystic dysplasia,
- convulsions,
- general muscle hypotonia making mobility and food intake impossible,
- bleeding in the GIT,
- increased levels of copper and iron in the blood.

Treatment and prognosis

It is a congenital disorder, so it is not possible to avoid it by prevention. There is currently no cure. Only symptomatic and supportive treatment is used.

Prognosis : children die around the age of 1.

Links

References

Použitá literatura

-
-
-

Template:Navbox - dědičné metabolické poruchy

Kategorie:Patobiochemie Kategorie:Genetika Kategorie:Pediatrie