Zellweger Syndrome

Zellweger syndrome is associated with the reduction or absence of functional peroxisomes in the cells of affected individuals. This syndrome is inherited in an autosomal recessive pattern with mutations in PEX genes. At birth, newborns with Zellweger syndrome can have hypotonia. Other clinical features include seizures, hepatomegaly, and polycystic kidney disease. Diagnosis is made when increased levels of very long-chain fatty acids are present in the blood. Currently, affected individuals with this syndrome are managed by supportive care as there is no cure for Zellweger syndrome. Unfortunately, this disease carries a poor prognosis with most infants expiring within the first year of life.



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Pathophysiology

Peroxisome Dysfunction

Dead Pear-Ox

Zellweger syndrome is associated with the reduction, absence, or dysfunction of peroxisomes in the cells of affected individuals. Normally, peroxisomes play important roles in metabolism, lipid biosynthesis, reactive oxygen species detoxification, and signaling.

Autosomal Recessive

Recessive-chocolate

This syndrome is inherited in an autosomal recessive pattern. In order to express an autosomal recessive disease, you must inherit two mutated genes, one from each carrier parent.

PEX Gene Mutation

Pez Jean-Mutant

Zellweger syndrome is associated with peroxisomal disorders caused by defects in PEX genes. PEX genes encode proteins called peroxins that are involved in different stages of peroxisomal protein import and/or the biogenesis of peroxisomes. Defects of functional peroxisomes can result in several metabolic abnormalities.

Clinical Features

Hypotonia

Floppy Hippo-baby

Newborns affected with Zellweger syndrome can present with hypotonia. These patients have decreased muscular tone throughout the body.

Seizures

Caesar

Patients with Zellweger syndrome may develop seizures. This is due to neurological damage brought on by the accumulation of defective peroxisomes.

Hepatomegaly

Liver-balloon

Enlargement of the liver can be seen in patients with Zellweger syndrome. Hepatomegaly may be present due to the accumulation of very long chain fatty acids (VLCFA). Due to dysfunctional peroxisomes, VLCFAs are not properly metabolized by ?-oxidation.

Polycystic Kidney Disease

Polly-parrot with Cystic Kidneys

Zellweger syndrome may be associated with polycystic kidney disease (PKD). PKD causes numerous fluid-filled cysts to grow in the kidneys. These cysts cause the kidneys to decline over time and eventually leads to kidney failure.

Diagnosis

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Increased Very Long-chain Fatty Acids (VLCFA)

Up-arrow Very Long Bacon & Chain with Acid-lemon

Patients with Zellweger syndrome have dysfunctional peroxisomes and cannot metabolize VLCFAs. Therefore, there will be an increase in VLCFAs, which will accumulate in nerves and various organs.

Management

Supportive Care

Supportive IV bags

There is no cure for Zellweger syndrome, and supportive care is currently the best option for affected individuals.

Considerations

Poor Prognosis

Gravestone

There is no cure for Zellweger syndrome and the prognosis is very poor. Most infants with this syndrome do not survive past the first 6 months of life.

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