Fact Sheet: Paediatric and Childhood Alpha-1 Antitrypsin Deficiency

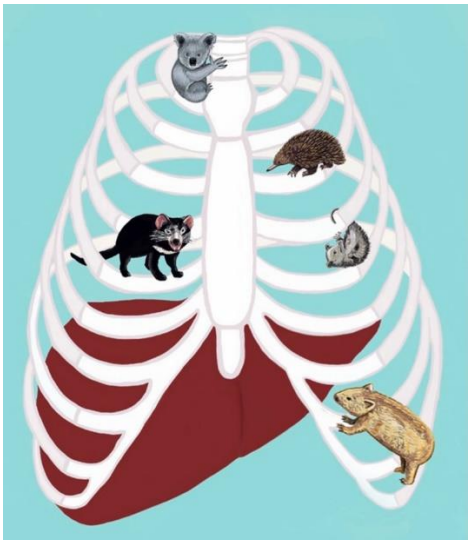
Background

Alpha-1 antitrypsin deficiency (alpha-1) is a genetic disorder, passed down from parents to children. The liver makes a protein called alpha-1 antitrypsin which normally leaves the liver via the bloodstream to protect the lungs and other organs. If there isn't enough alpha-1 antitrypsin in the bloodstream (due to low levels being made or destroyed, or if it is stuck in the liver due to a genetic mutation), children have the disorder known as alpha-1.

The normal version of alpha-1 is called PiMM genotype. Babies and children with two abnormal PiZZ alpha-1 genes are at higher risk of childhood liver disease from alpha-1. Another version of the mutated gene is called the S gene but it doesn't seem to cause childhood liver disease.

About ten percent of children will have severe alpha-1 liver problems causing liver disease. Some PiZZ children will be born with liver disease or may develop it later in their childhood. Unfortunately, out of these children, 2% (usually those with PiZZ phenotype) will have liver failure and will need a liver transplant in the first two years of life. A liver transplant cures alpha-1 and children do well after liver transplants.

Some children can develop neonatal cholestasis (where the flow of bile from the liver is reduced or blocked), liver dysfunction, liver failure or cirrhosis. Liver problems are usually obvious during the first few months of life.



Liver problems may be called neonatal hepatitis, but it is not contagious hepatitis. Many children with PiZZ genotype will have abnormal liver function test results but results return to normal during their childhood. Some alpha-1 infants will have jaundice for a lot longer than non-alpha-1 infants.

What are the Symptoms of Disease?

Some of the signs and symptoms are:

- Jaundice (yellowing of the whites of the eyes / skin)
- Poor weight gain
- Darker urine
- Pale poo (stools)
- Ascites (collection of fluid in the tummy)
- Portal hypertension (high blood pressure in the blood vessels around the liver).
- Bleeding

What if my Child has Alpha-1?

Your child will be closely monitored. Doctors will be looking at how well your child's liver is working. Monitoring may include blood tests, scans, a liver biopsy, and growth and development.

Liver function monitoring looks for any complications and quick treatment may be required. The clinical course varies across children. Unfortunately, there are no prevention strategies or specific treatments to stop liver problems. Most children become stable, with liver enzymes returning to normal through time.

Treatments or a Cure?

There is no cure for alpha-1 liver disease however, clinical trials are underway. The medical team looking after your baby or child will monitor and manage any symptoms caused by alpha-1. Your child's milk, diet or number of kilojoules consumed may need to be changed, in consultation with the health team. Dietary changes may be required because babies and children with damaged livers have poor bile production and they don't absorb food as well, which can lead to weight loss or difficulty in gaining weight. Some children may need vitamins, especially vitamin K, to help with blood clotting.

Who should be Tested for Alpha-1?

- Brothers, sisters and biological parents of affected children

Seeking Support or Counselling?

Having a sick child is very distressing and many parents find that joining an alpha-1 support group is helpful. There are Australian and international Alpha-1 Facebook groups and the Alpha-1 Organisation Australia has a video support group. There is also a child-specific Facebook group for families affected by alpha-1.

You may want to speak to a counsellor, or a genetic counsellor if you are considering having further children.

References

Pinto RB, Schneider ACR, de Silveira. Cirrhosis in children and adolescents: An overview. *World Journal of Hepatology*. 2015. 7(3): 392-405.

Children's Hospital of Pittsburgh. www.chp.edu.

More Information or Support?

Contact Alpha-1 Organisation Australia : email contactus.a1oa@gmail.co

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This brochure is one in a series of information sheets produced by the Alpha-1 Organisation Australia (A1OA). This information is designed to be a guide only and does not replace advice given by your health professional. Any treatment information or brand names are correct at the time of printing. If the information raises concerns or if you have further questions please consult your doctor.