Alberta Health Services
Alberta Newborn Screening

Talking with Parents > When talking to a parent whose baby has received an abnormal screen result, let them know that an abnormal screen result doesn't mean that the infant has the condition. It means that they have a higher chance of having the condition, and they need more tests to find out for sure. A printable sheet for parents with more condition specific information can be used to support your conversation, visit ahs.ca/nms and under Quick Reference click on *What conditions are screened for?*

Tyrosinemia Type I (TYR1)

(metabolic condition: amino acid disorder)

Information for Health Professionals

Also known as:

- fumarylacetoacetate hydrolase deficiency
- fumarylacetoacetase deficiency
- hepatorenal tyrosinemia

What are amino acid disorders?

Amino acid disorders are inherited metabolic conditions in which certain amino acids cannot be fully broken down. This leads to an accumulation of toxic metabolites in the body which can cause serious health problems.

What is TYR1?

Tyrosinemia type 1 (TYR1) is an amino acid disorder resulting from a defect in the enzyme involved in the breakdown of the amino acid tyrosine. Tyrosine is a building block of proteins. It is important in the synthesis of neurotransmitters, hormones, melanin, and several other biomolecules. The clinical features of TYR1 result from accumulation of tyrosine and its toxic metabolites in the liver, kidney, and central nervous system.

What causes TYR1?

TYR1 is caused by a deficiency of fumarylacetoacetate hydrolase (FAH), an enzyme involved in tyrosine degradation. When FAH activity is absent or decreased an alternative tyrosine degradation pathway is activated leading to accumulation of succinylacetone (SUAC) and other toxic metabolites.

How common is TYR1?

TYR1 is a rare condition with an estimated incidence of 1 in 100 000 infants born in Canada. Although TYR1 occurs in all ethnic groups, it is more common in French Canadians.

What are the clinical features of TYR1?

Infants with TYR1 appear normal at birth. Within the first few months of life they may present with feeding problems, vomiting, diarrhea, lethargy, failure to thrive, hepatitis/liver failure and renal tubular dysfunction.

What is the screening test for TYR1?

Screening for TYR1 is performed by measuring SUAC on the newborn bloodspot. Newborn screening will not detect all infants with TYR1. Infants with symptoms or signs consistent with TYR1 need timely assessment and diagnostic testing even if their screen result is normal.

How is the diagnosis confirmed?

The diagnosis of TYR1 is confirmed by measuring tyrosine and other metabolites in blood and urine. Molecular genetic analysis of the FAH gene may be performed. The Clinical and Metabolic Genetics Program will arrange diagnostic testing.

How is TYR1 treated?

TYR1 is treated with medications and a diet low in tyrosine. Individuals with TYR1 are treated and monitored by a healthcare team including a metabolic specialist and a dietician. Early treatment can prevent developmental delay and reduce mortality from acute liver and kidney complications. Treatment is lifelong.

Is TYR1 inherited?

TYR1 is inherited as an autosomal recessive disorder. Parents of a child with TYR1 are carriers of the condition and have a 1 in 4 chance of having another affected child in each subsequent pregnancy. TYR1 carriers are healthy. Genetic counselling is available to families with TYR1.

Additional resources are available through:

Clinical & Metabolic Genetics Program (Edmonton)

8-53 Medical Sciences Building
8440 – 112 St. NW
Edmonton, AB T6G 2H7
Phone: 780-407-7333
Fax: 780-407-6845

Emergency consultations:
Phone 780-407-8822 and ask for the specialist on call for metabolic diseases.

Clinical & Metabolic Genetics Program (Calgary)

Alberta Children's Hospital
28 Oki Drive NW
Calgary, AB T3B 6A8
Phone: 403-955-7587
Fax: 403-955-3091

Emergency Consultations:
Phone 403-955-7211 and ask for the specialist on call for metabolic diseases.

Early screening and follow-up care – every baby, every time

For more information about the Alberta Newborn Screening Program, visit www.ahs.ca/newbornscreening