What is Galactosemia?

Galactosemia is a hereditary disease that is caused by the lack of a liver enzyme required to digest galactose. Galactose is a breakdown product of lactose, which is most commonly found in milk products.

Normally, when a person consumes a product containing lactose, the body breaks it down into glucose and galactose which are used by the body for energy.

How common is this disease?

1 in every 7,500 live births will have some form of galactosemia.

It is also estimated that 1 in every 40 people may be a carrier of this defective gene.

Who is a carrier?

A carrier is an individual who has one defective gene and one normal gene. This means that the person himself/herself is not affected but can pass on the defective gene to the next generation which may lead to diseased state.

What causes galactosemia?

Galactosemia is caused by mutations in the following genes:

GALT p-arm of chromosome 9
GALK1 q-arm of chromosome 17
GALE - p-arm of chromosome 1

How can galactosemic child be born to normal parents?

The gene defect for galactosemia is unknowingly passed down from generation to generation.

When two carriers have a child together there is a 25% chance that the child will be born with the disease and a 50% chance that the child will be a carrier.

How can we diagnose Galactosemia?

Galactosemia is normally first detected through newborn screening (NBS) by GALT test in which heel prick blood is collected on special graded filter paper taken within 72 hours of birth.

If newborn screening is positive for GALT, further DNA based testing can be done for confirmation.

What are the Signs and Symptoms?

Galactosemia usually causes no symptoms at birth, but jaundice, diarrhea, and vomiting soon develop and the baby refuses to feed and fails to gain weight.

If not detected in early life, it can later on result in liver disease, cataracts, brain damage (mental retardation), and even death. Bacterial infections are common in untreated galactosemic infants.

What steps need to be taken?

Breast feeding to galactosemic children must be avoided. Galactose and lactose should be strictly eliminated from the diet throughout life.

What is the alternative if the baby cannot be given milk in any regular form?

An infant can be fed with soy formula, meat-based formula or protein hydrolysate formulas which don't contain lactose and galactose.

Is there a treatment for this disease?

- Presently, there is no chemical or drug substitute for the missing enzyme and hence no treatment for galactosemia.
- However, there exists a dietary management for galactosemic patients.
- A person with galactosemia will never be able to properly digest foods containing galactose.
- Antibiotics are commonly prescribed for all galactosemic children who have ingested galactose to prevent sepsis.
- Even after a galactosemic child has been switched to a soy based formula, sepsis can still develop if the child has previously ingested galactose.
- Children suspected to have galactosemia, whether clinically diagnosed or having a positive newborn screening (NBS) result showing Galactosemia, should have a sepsis work up.