NEWBORN GENOME SCREENING

Why is early screening important ?

Screening involves looking for conditions in advance before they produce symptoms.

A large number of childhood onset diseases have a genetic basis. With conventional methods there is often a long delay in diagnosing genetic conditions even after the onset of symptoms. This delay results in irreversible physical and intellectual damage in children.

In many of those conditions early detection and treatment can significantly improve outcomes .

In our newborn genome screening programme we have hand picked around 400 genetic variations which will have implications on the child's health within the first 5 years of life . Around 120 of these diseases have specific treatments as well.

Compared to currently used newborn screening programmes which screen for around 50 different conditions based on chemical levels in the blood, genome screening can pick up a much larger number of conditions with a single test. Moreover the data can be reanalyzed in future for other genetic conditions if necessary

How is it done ?

A small blood sample is taken from the umbilical cord at birth or directly from the baby. DNA from the blood sample is analysed to identify genetic variations. The results will be shared with you and your healthcare provider in about 6 weeks from the test.

The technology used is called whole genome sequencing. Although the test is capable of analysing your child's entire genome, for the purpose of relevance we will be only reporting for the presence of around 400 genetic variations that will have implication on your child's health at this moment. The sequenced data can be re-analysed for other conditions any number of times in future after discussion with your doctor

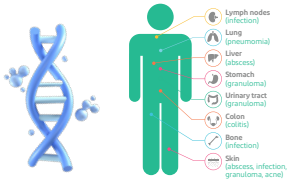
REWRITING THE PAINFUL DIAGNOSTIC JOURNEY IN RARE DISEASES

If Noor had an early genetic diagnosis .....

- Doctors could have started medications to avoid serious infections and multiple hospital admissions
- Early knowledge of this genetic condition could have helped doctors to send her for life saving bone marrow transplant before serious infections developed

This is just an example of the 400 different significant conditions that can diagnosed through Kings Newborn genome Screening programme

Noor was troubled by repeated unusual infections needing frequent hospital admissions from 6 months of life.



At 4 years of age Noor was eventually diagnosed to have a rare genetic disorder called chronic granulomatous disease due to NCF1 mutation making her white cells unable to fight bacteria or fungi properly.

Without appropriate treatment this is a life limiting condition. Appropriate preventive antibiotics can improve life expectancy and bone marrow transplant can cure this disease.

Children like Noor with rare genetic conditions go through years long painful journey before a diagnosis is finally made

Delay in time critical treatments often results in irreversible physical and intellectual damage

Few things to be aware of

UNEXPECTED FINDINGS

The screening may reveal unexpected genetic findings not related to the initial purpose of the test. Examples of such findings could include:

1. Genes associated with adult-onset conditions.
2. Carrier status for genetic disorders

But for newborn screening we minimise such situations by actively reporting a select list of conditions only.

VARIANTS OF UNCERTAIN SIGNIFICANCE (VUS)

Some genetic variations identified may be classified as Variants of Uncertain Significance (VUS). These are changes in the DNA whose impact on health is not yet known. To reduce the anxiety and uncertainty related to test results for the purpose of newborn screening we will not be actively reporting variants of uncertain significance. However, if the child develops symptoms of any particular condition we can re analyse the genetic data to see if there are any variants of uncertain significance related to those symptoms

IMPLICATIONS ON WIDER FAMILY

The results of your child's genetic screening could have implications for other family members. Certain genetic conditions are hereditary, meaning other relatives could be affected. You may be encouraged to share relevant findings with family members who could benefit from this information

IMPLICATIONS ON INSURANCE

Some insurance policies do not cover genetic conditions. This is something to be considered prior to undertaking the test. But if a child has one of the genetic conditions screened for , the benefit of early diagnosis will likely outweigh the

CONFIDENTIALITY AND DATA PROTECTION

Your child's genetic data will be stored securely and will only be accessible to authorized personnel. Results will be shared with you and your healthcare provider. The data will only be used for research with your specific separate written consent.