

### A Public Interest Initiative



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### **New Born Screening**



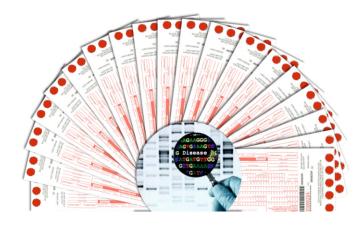


## Screen and save

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### **New Born Screening**

It all began when a 5 year ICMR study of new born screening concluded with alarming results. 30000 babies from Chennai were screened for Congenital Hypothyroidism and Congenital Adrenal Hyperplasia and the incidence was relatively very high as CH-1/727; CAH 1/2036. Both the disorders are treatable and treatment, though lifelong is affordable to many.



The need of public screening program was strongly felt and the medical team of FCRF under the proactive leadership of Prof. Suresh decided to bring it to public use and benefit. Inferring from the referrals to MediScan's genetic clinic three more disorders are added to the program by our geneticist Dr. Sujatha Jagadeesh.

### What is New Born Screening?

It is a simple blood test to screen babies for 5 inherited metabolic disorders.

- © **Congenital Hypothyroidism** which if untreated leads to mental retardation
- Congenital Adrenal Hyperplasia, if undetected male babies may die and female babies may develop external ambiguity of gender.
- Galactosaemia babies do not assimilate even mother's milk and if not intervened with special diet may die
- © Glucose 6 Phosphate Dehydrogenase deficiency results in red blood cell disintegration
- O Cystic fibrosis which if not treated affects the lungs, pancreas and intestine.

### Isn't it only for sick babies? My baby looks healthy...

Most babies are healthy when they are born. Screening should be done for all babies because a few babies look healthy but have rare health problem. With special lab tests the baby with any one of the 5 disorders can be identified. This will alert the parents to take specialized medical care the infant would need. Early detection helps to give treatment before the baby becomes ill.

## The family bloodline is healthy... There is no previous history of inherited disease...

One may not know. Inherited disorders may surface after a few generations also. Any one's baby will be picked at random by screening and saved from death or from a lifetime of mental or physical disability.

### How is the screening test done?

All 5 tests are performed on a few drops of blood obtained by pricking the baby's heel. The sample is usually taken 72 hours after birth. This method can be used until the baby is 15 days old. After that the test can be done only on the blood serum. The sample is collected on a filter paper card, air dried and baby's details like parents name birth weight are filled, and then sent to the lab for testing.

### What if the screening 'tests' positive?

The baby has to undergo a second test (retest). This can be done only within I5days of life. It is done in the same way like the previous test (filter paper)

# What if again the test result is positive? Does it confirm that the baby definitely has problem?

You will be advised to have another confirmatory test for the baby. This test is done using a different method. And only if this test proves the disorder, treatment will be initiated. If the test result is positive the respective hospital doctor who is in-charge of screening will be informed and asked to further guide the parent about follow up and treatment.

### If the test result is negative, does it mean the baby is healthy?

If the results are screen negative the baby will not have the 5 disorders screening is done for (an error of 1 to 5% may happen).

#### What if my baby has one of these disorders? Can it be cured?

None of these disorders can be cured. However, the serious effects can be lessened – and often be prevented completely – if a special medical treatment is started early. Most of these disorders are very complicated to treat and medical care should be coordinated by a specialist in the specific disorder

