



## INFORMATION FOR PARENTS/CARERS RESAMPLE REQUESTED FOR CITRULLINE

### What is the Newborn Screening Programme?

Your baby had a blood sample taken from the heel at about three days of age. The blood was absorbed into a special card and sent to the NSW Newborn Screening Laboratory in Sydney where it was tested for several disorders.

### Why does my baby need a second test for Citrulline?

One of the test results from your baby's sample was slightly abnormal and the laboratory needs a repeat sample just in case your baby has a mild disorder. In the majority of babies the second result is **quite normal**.

If, however, your baby's test result remains abnormal, your doctor will arrange for your baby to have further blood and urine tests.

### Why are babies screened for Citrulline disorders?

The NSW Newborn Screening Programme now has new technology that can rapidly detect more than 20 rare disorders. Two of these disorders cause an abnormal citrulline result. One is called Citrullinaemia or ASAS for short.

The second is called Arginino-succinicaciduria or ASAL for short. Both are extremely rare disorders. Only one in every 1:100,000-200,000 babies has one of these disorders.

In the first days, newborn babies with ASAS or ASAL are not obviously different from unaffected babies. An early sign might be a tendency to oversleep. Delay in diagnosis and treatment may lead to more serious problems.

### What are Citrullinaemia (ASAS) and Arginosucciniacidemia (ASAL)?

ASAS and ASAL are enzyme deficiencies. These enzymes are part of the system of chemical reactions in the body that help to dispose of extra nitrogen. If the enzymes work too slowly, ammonia may accumulate. High levels of ammonia inside the body are toxic. Babies with deficiencies of these enzymes benefit from dietary treatment.



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For further information please contact your paediatrician or the NSW Newborn Screening Programme  
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