

Newborn screening

All babies in Australia have a screening test done within a few days of birth if their parents agree. The test is done by pricking the baby's heel, getting some blood and sending it to a laboratory.

At the laboratory, the blood is tested for about 25 different conditions. The most common ones are:

- Congenital hypothyroidism, in which not enough thyroid hormone is produced and which causes intellectual disability if untreated
- Phenylketonuria, which causes intellectual disability if untreated
- Cystic fibrosis, which affects the lungs and gastrointestinal system
- Galactosaemia, which causes serious problems such as poor growth, liver disease and intellectual disability if untreated.

These conditions are serious and sometimes life-threatening. Diagnosing them early can make an enormous difference to a child's life.

More than 99 babies out of 100 will have normal results. Parents are not contacted if the results are normal.

Occasionally, the testing is unclear. Parents will be contacted so that the blood test can be re-done and there may be other tests. It can take a couple of weeks to sort things out.

Only about one baby in 1000 is found to have any of the problems being looked for. Parents are contacted and an appointment will be made to see a specialist. More tests will be done to confirm that the problem is present and treatment will be started.

Please note: like every test, this one is not perfect. It is possible for a child to have one of these conditions, but for the testing to miss it. So even if your baby has had these tests and you have been told all is well, the tests do not provide a guarantee. If you are worried about your child, see a doctor.

Contacts and further information

- All states and the ACT have familial cancer services. Contact them through your local state or territory health department.
- Australasian Genetic Alliance at <http://www.australasiangeneticalliance.org.au>
- MyDr at <http://www.mydr.com.au>
- The Centre for Genetics Education at <http://www.genetics.edu.au>
- HealthInsite at <http://www.healthinsite.com>
- MedicineNet at <http://www.medicinenet.com>
- For other related fact sheets, you can contact the Gene Technology Information Service on **free call Australia-wide 1800 631 276** or email gtis-australia@unimelb.edu.au or visit Biotechnology Australia's website at <http://www.biotechnology.gov.au>