

Pilot project for newborn mass screening tests

Participation in the pilot project for additional testing of spinal muscular atrophy (SMA) and severe combined immunodeficiency (SCID) to general newborn mass screening tests

The Aichi Prefectural Government and the City of Nagoya will participate in the national pilot project for the newborn mass screening tests conducted by the Children and Families Agency, Government of Japan. This is to determine if two additional diseases; spinal muscular atrophy (SMA), and severe combined immunodeficiency (SCID), should be included in the newborn mass screening tests, which currently include tests for 20 diseases.

The test results for the two additional diseases will be provided to the Children and Families Agency and its study group led by Dr. Go Tajima (scientific research on children and families) without identifying the number of tests performed and positive cases found, to determine the way of enabling all babies born in Japan to be tested for the two diseases.

1. Additional diseases tested for the mass screening

There are two diseases that have been newly added to the 20 diseases that are currently tested.

- Spinal muscular atrophy (SMA)
- Severe combined immunodeficiency (SCID)

SMA is a disease that causes muscle weakness throughout the body and affects 1 in 20,000 people. Whereas SCID affects 1 in 50,000 people and makes patients susceptible to serious infections because their immune systems do not function. If they are not treated, there is a possibility of death by the age of one or two. In recent years, SMA can be controlled, and motor function can be improved with the early administration of medicine. SCID can be almost completely cured with hematopoietic cell transplantation (bone marrow or cord blood transplantation), which produces immune-supporting cells. For more information on the diseases, please visit the following websites.

- Spinal muscular atrophy (SMA) <https://www.sma-rt.org/sma.html>



- Severe combined immunodeficiency (SCID) <https://pidj-nbs.jp/scid.html>



2. Screening process

The blood sample for the general newborn mass screening is used to test for the two additional diseases so there would be no additional blood sample taken from your baby.

Your baby's birth hospital would take a blood sample and send it to a laboratory for testing. The lab tests the sample and sends the results to the birth hospital.

- If there are no abnormalities,
 - the birth hospital will report the results to the guardians (end of process)
- If suspected abnormalities are found,
 - the birth hospital will report the results to the guardians
 - the guardians will be advised to bring the newborn to a hospital for further testing
 - the hospital will report the results to the guardians

3. Fees

The test fee of the test for those who participate in the pilot project is borne by the Aichi Prefectural Government or the City of Nagoya if the medical institution is in Nagoya. Please ask your medical practitioner about any other costs such as taking and transporting the blood sample.

4. Reporting the results of the newborn mass screening tests to the study group from the Children and Families Agency and protection of personal information

The data of the children who underwent the screening tests for the SMA and SCID will be shared with the Children and Families Agency and its study group led by Dr. Go Tajima, for the purpose of verifying the efficacy of the screening tests without identifying the number of tests performed and positive cases found, and the result of the detailed examinations such as names of the diseases and number of patients. We take the utmost care in protecting personal information and assure that it will not be used for any purpose other than for the newborn screening. The test results will be statistically processed and no personal information will be identified on the final research paper when it is released.

5. Please note that:

- Not all SMA and inherited immunodeficiencies can be found through the newborn mass screening tests.
- Immunodeficiencies other than SMA and SCID may be found through the newborn mass screening tests.
- This is a screening test. Even if your children are found to require detailed examinations, no diseases may be found in the result of detailed examinations.
- You may be contacted by a local health center in your area depending on the findings of detailed examinations.