The Chinese Foundation of Health, Medical Laboratory.

Newborn Screening Center

The Consent Form for Optional Newborn Screening Items

Dear parents:

In order to help early detection of congenital metabolic disorders that can be screened for, the screening center offers the following optional screening items. All tests will not increase the amount of blood drawn or burden your baby, and most of these conditions have appropriate treatment methods. Therefore, if early diagnosis is made through newborn screening and timely medical interventions are taken, it can effectively reduce harm to your baby. The test results can also serve as a reference for your future family planning and genetic counseling.

We are seeking your consent and inviting your baby to undergo testing. For more details on the diseases included in the screening and related information, please refer to the website. The results of this program will help the Health Promotion Administration, Ministry of Health and Welfare (HPA) determine whether these conditions should be included in routine screening in the future.

LSD 4-in-1 Screening

Includes **Pompe disease, Fabry disease, Gaucher disease, and Mucopolysaccharidosis Type I**.

Lysosomal storage disease (LSD) is caused by a dysfunction in the lysosome. Due to mutations in enzymes within the lysosome, defects occur, leading to the accumulation of metabolic products such as lipids, glycoproteins, and mucopolysaccharides in cells. This accumulation eventually causes cell death and organ dysfunction. Currently, some lysosomal storage diseases can be treated with enzyme replacement therapy, which are administered via injection to improve the patient's condition and delay disease progression. Since the damage caused by lysosomal accumulation is mostly irreversible, early detection and treatment are crucial.

SCID & SMA 2-in-1 Screening

Severe Combined Immunodeficiency (SCID)

SCID is caused by abnormal T lymphocyte function, resulting in deficiencies in cellular and antibody-mediated immunity. This makes patients highly susceptible to viral and bacterial infections. Patients should avoid exposure to environments prone to infections, and the incidence is approximately 1 in 70,000. It is important to note the potential adverse effects of this condition when administering live attenuated vaccines (including BCG, MMR, yellow fever, varicella, and rotavirus vaccines) to babies. Newborn screening provides an opportunity for early diagnosis and treatment.

Spinal Muscular Atrophy (SMA)

SMA is a genetic disorder caused by mutations in the *SMN1* gene, leading to the degeneration of spinal motor neurons, resulting in muscle weakness and atrophy. The incidence is approximately 1 in 10,000. The most severe form, Type 1, presents within the first 6 months of life with symptoms such as weak cry, difficulty feeding, labored breathing, swallowing difficulties, and severe weakness in the limbs and trunk. Most children with Type 1 SMA die from respiratory failure by the age of 2. Type 2 and Type 3 SMA generally present in infancy or childhood. In the past, clinical management focused on supportive care to alleviate complications, but now, there are medications available for treatment.

Early diagnosis through newborn screening and integrated medical care can reduce mortality and complications. However, this test can only detect 95% of patients, as about 5% of individuals may have normal *SMN1* gene copy numbers but abnormal function, which cannot be detected by this screening.

BD & DMD 2-in-1 Screening

Biotinidase Deficiency (BD)

BD occurs when there is a deficiency in the activity of biotinidase, an enzyme that helps the body utilize biotin effectively. Depending on the enzyme activity, the condition can be classified into profound deficiency or partial deficiency. The treatment for this condition is oral biotin supplementation, which, when taken, can prevent or eliminate biochemical metabolic abnormalities, seizures, and skin-related symptoms.

Duchenne Muscular Dystrophy (DMD)

DMD is caused by a mutation in the DMD gene located on the X chromosome, leading to a lack of dystrophin, a protein necessary for muscle function. This results in muscle atrophy. As muscle tissue deteriorates with age, individuals gradually experience symptoms such as difficulty walking, inability to stand or move, and loss of respiratory muscle function. In the later stages, complications such as heart failure can lead to death. The incidence of DMD in males is approximately 1 in 3,000 to 1 in 6,000. Female carriers are often asymptomatic, though some may exhibit mild muscle degeneration or more severe clinical progression. With the use of corticosteroids and supportive treatment, the progression of the disease can be delayed, extending survival. Gene therapy is also in the clinical trial stage.

MPS & ALD 2-in-1 Screening

Mucopolysaccharidosis (MPS)

Includes **MPS Type II, and free results for MPS Type IVA, and MPS Type VI**.

Mucopolysaccharides are essential components of the body's important organs, including the bones, blood vessels, skin, hair, and corneas. MPS patients lack the enzymes required to break down mucopolysaccharides, causing these molecules to accumulate in various organs and impair their function. MPS is currently classified into seven types. MPS Type II, MPS Type IVA, and MPS Type VI can be treated with enzyme replacement therapy, using genetically engineered enzymes administered via injection to improve the patient's condition and delay disease progression.

Adrenoleukodystrophy (ALD)

ALD is primarily caused by the inability of peroxisomes within cells to metabolize very long-chain fatty acids (VLCFA). This leads to the accumulation of VLCFA in the white matter of the brain and the adrenal cortex, damaging the myelin sheath of the central nervous system, which interferes with nerve conduction and causes developmental delays and degeneration of the central nervous system. For cases that have not yet shown neurological symptoms, Lorenzo’s oil can be used to slow the progression of the disease. In cases where early-stage symptoms are not severe, bone marrow transplantation may be considered as a treatment, though this carries risks of rejection or opportunistic infections, which could lead to death.

The above are the optional screening items provided by the screening center. As the number of screening items increases, the likelihood of your baby needing additional tests may also rise. If any abnormal results are found, it means that your baby may have a higher chance of having the condition compared to the general population. In such cases, we will arrange for a re-examination or refer you baby to a nearby genetic counseling center for further diagnosis and treatment. All data will be recorded in the medical record. Furthermore, a confirmed diagnosis may affect your baby's insurance benefits. Your baby's sample will be properly stored by the newborn screening center and will not be used for any other purposes.

In compliance with the relevant laws for personal data protection in Taiwan, the screening center collects, processes, and utilizes your personal data based on business needs. Your data will only be used within the scope of newborn screening services. By selecting to accept any of the optional screening items below, you acknowledge that you have read, understood, and agreed to the screening center's use of your personal data. After reviewing the relevant disease education materials, if you wish to proceed with the optional screening for your baby, please check the consent box below, sign, and provide the signed form to your baby's nurse or doctor.

If you have any questions, please feel free to contact us to ensure that your baby receives the timeliest medical services!
The Chinese Foundation of Health (CFOH), Medical Laboratory.
Newborn Screening Center
Phone: 02-87681020 Website: www.cfoh.org.tw

This form is retained by the Legal Representative.

Declaration of the Legal Representative:

I have carefully read the relevant information regarding the optional newborn screening items and understand their purpose, methods, and the importance of the diseases involved. Any unclear points have been fully explained by the medical staff, and I now have a clear understanding (please refer to the website for detailed information).

☐Agree ☐Disagree To allow my baby to undergo the LSD 4-in-1 Screening.
☐Agree ☐Disagree To allow my baby to undergo the SCID & SMA 2-in-1 Screening.
☐Agree ☐Disagree To allow my baby to undergo the BD & DMD 2-in-1 Screening.
☐Agree ☐Disagree To allow my baby to undergo the MPS & ALD 2-in-1 Screening

Legal Representative:_____(Signature)；Date: _____/_____/_____ (Year/Month/Day)
ID Number (Residence Certificate):_____
Please provide the fastest and most accurate contact number:_____

This form will be retained by the collecting hospital.