VŠEOBECNÁ FAKULTNÍ NEMOCNICE V PRAZE
Gynekologicko porodnická klinika, Neonatologické oddělení
U Nemocnice 499/2, 128 08 Praha 2 | IČ: 00064165, tel.: 224 961 111

Účinnost dokumentu od: 08. 03. 2022
Edukační materiál

SCREENING PROGRAMME FOR SPINAL MUSCULAR ATROPHY AND SEVERE COMBINED IMMUNODEFICIENCY

Dear Madam,

We offer participation in a pilot programme for early detection of **spinal muscular atrophy (SMA) and severe combined immunodeficiency (SCID)** in newborns to your family and your child. This pilot screening programme (Early Detection Programme) enables early detection of these two serious rare genetic diseases in newborn babies. It is intended only for participants in public health insurance in the Czech Republic. **Screening (i.e. search) examinations in newborns, which lead to early detection of these two hereditary diseases, have not been carried out in our country so far.** Participation in this programme can provide you with important information about your child's health condition and contribute to successful treatment and, in many cases, to saving your child's life. Early detection of these diseases in their presymptomatic stage, i.e. before they manifest their typical clinical symptoms, is a prerequisite for successful treatment.

Currently, neonatal laboratory screening for 18 serious rare diseases is already underway as a standard practice, and SMA and SCID would be introduced into the national neonatal screening programme for rare genetic diseases after the expected successful completion of the pilot project (see more details – www.novorozeneckyscreening.cz).

Please read the following information to help you decide whether to take part in this programme.

What Is Spinal Muscular Atrophy (SMA) and Why Is Its Early Detection Important?

Spinal muscular atrophy (SMA) is a serious inherited neuromuscular disease that affects the part of the nervous system responsible for controlling muscles important for movement of the lower and upper limbs, head, respiratory muscles and swallowing. Children affected by this disease gradually lose the ability to move independently. The disease is manifested by slowly increasing weakness of the leg muscles, worsening mobility of the arms, and even complete loss of mobility of the whole body. In European populations, approximately one in 10,000 newborns is affected.

The advantage of participating in the programme is that the disease can be detected as soon as possible after the child's birth, which will allow to start effective treatment of the child early, before the development of any difficult-to-treat or even untreatable complications due to delayed clinical diagnosis of the disease. Effective treatments for SMA are now available and, together with specialised rehabilitation, can significantly reduce the symptoms of the disease.

What Is Severe Combined Immunodeficiency (SCID) and Why Is Early Detection Important?

Severe combined immunodeficiency (SCID) is a serious inherited disease of the immune system. Babies born with SCID usually appear healthy at birth because they are protected by antibodies from their mother, but as maternal antibodies gradually decline, the children are increasingly at high risk of serious infections. During the first few months of life, they often develop diarrhoea, pneumonia, otitis media, sepsis and skin infections. Vaccination with attenuated vaccines is also a serious risk for patients; this type of vaccine can cause serious complications and even child's death. If the disease is detected early, the success rate of

SCREENING PROGRAMME FOR SPINAL MUSCULAR ATROPHY AND SEVERE COMBINED IMMUNODEFICIENCY

targeted treatment, which is urgent bone marrow transplantation and restoration of normal immune function, is increased. In European populations, approximately one in every 50,000 new-borns is affected.

What Does It Mean That SMA and SCID Are Hereditary?

SMA and SCID are so-called recessive inherited diseases, therefore, it is reasonable to assume that the parents of the patients are so-called healthy carriers of the disease.

What Is the Screening Process for SMA and SCID?

Screening for these two diseases is done from a drop of blood taken from the heel of the newborn between 48 and 72 hours after birth. A small drop of capillary blood (max. 100 microlitres, i.e. e.g. the size of a larger pinhead) is used for screening, as part of a one-off collection of a series of small blood samples for the existing neonatal laboratory screening for rare genetic diseases (see above). For this reason, the SMA and SCID screening pilot examination will not burden your child with additional blood collections compared to current practice. This routine collection of a small blood sample from the heel of the newborn is virtually painless and does not traumatise your baby in any way.

What Can You Expect if You Participate in the SMA and SCID Early Detection Programme?

The reason for this screening test and the method of taking a small sample of your baby's blood will be explained to you by the doctor at the maternity hospital. Your possible consent to participate in this pilot programme will be recorded both in the screening card and in your child's medical records of the relevant healthcare provider (healthcare facility). This information is subject to all legal regulations and the principle of medical confidentiality. Selected screening laboratories will perform genetic analysis focused exclusively on these two diseases.

Since these are very rare diseases and all newborns born in the Czech Republic are tested, the result is only communicated to the family if there is a positive detection of SMA or SCID. If the laboratory test is negative – i.e. the disease is not detected, the result is not sent to the paediatrician or maternity hospital, similar to the national neonatal laboratory screening for selected genetic diseases (see above). In the very rare event of a repeated blood sample collection needed, e.g. because of an unclear screening result or incomplete blood collection in the maternity hospital, you will be asked to visit the paediatrician's surgery or have a repeat blood sample taken in the maternity hospital.

How Will the Result Be Communicated to You

Due to the very rare occurrence of both diseases, i.e. approximately ten children with one of these diseases per year in the Czech Republic, the examining laboratories do not inform a) the child's guardians, b) the general practitioner for children and adolescents (paediatrician) who has taken your child into care about the normal (negative) findings. On the other hand, if your child is found to have SMA or SCID (positive findings) or if the presence of one of these diseases is suspected, you will be informed about the result of the examination by the general practitioner for children and adolescents (paediatrician) who has taken your child under his/her care and recommended further specific diagnostic and treatment processes.

The positive result of the screening test is always verified at a specialised workplace within one of the university hospitals and long-term treatment is carried out at specialised workplaces of selected hospitals.

SCREENING PROGRAMME FOR SPINAL MUSCULAR ATROPHY AND SEVERE COMBINED IMMUNODEFICIENCY

What Does It Mean That You Have Not Been Contacted/Informed by the Paediatrician about Your Child's SMA and SCID Results

If you are not contacted by the registering GP for children and adolescents (paediatrician), this means that your child's screening test is negative (i.e. no SMA or SCID is suspected).

Can I Refuse SMA and SCID Screening? Will I Then Have to Pay for the Examination?

Screening for SMA or SCID can be declined, but in accordance with the opinion of experts in the respective medical fields, considering such a step is recommended. Similar pilot projects are currently underway in other developed countries and the Czech Republic is joining the group of developed European countries focusing on early detection of rare genetic diseases. This pilot project is also in line with relevant domestic and international expert recommendations. At the same time, the screening examination can provide you with important information about your child's health and thus significantly influence the quality of his or her future life. The SMA and SCID examination is free of charge for your child and this project is covered by the special preventive funds of the public health insurance. If you still decide to refuse only one of the tests (i.e. either SMA or SCID), it will not be possible to test for either disease, as the diagnostic kits in the screening laboratory are only used to detect both diseases simultaneously.

What Happens to the Results of the Screening Tests?

The results of the screening tests are stored by health care providers, are part of medical records and are handled in accordance with the applicable legislation of the Czech Republic.

Once the analysis is complete, the DNA sample (deoxyribonucleic acid, which is isolated from the blood sample) is immediately discarded. The screening card is archived in accordance with Decree No. 98/2012 Coll., on Medical Documentation, as amended, and after the expiry of the archiving period it is disposed of in accordance with the provisions of Act No. 499/2004 Coll., on Archiving and Records Management and on the Amendment of Selected Acts, as amended.

For the purpose of programme evaluation, the results will be processed in the National Health Information System (NHIS) in accordance with Act No. 372/2011 Coll., on Health Services and Conditions of Their Provision, as amended, and will be handled in full compliance with Regulation (EU) No. 2016/679 of the European Parliament and of the Council of 27 April 2016, on the Protection of Natural Persons with Regard to the Processing of Personal Data and on the Free Movement of Such Data and Repealing Directive No. 95/46/EC (General Data Protection Regulation), also known as GDPR. All persons who will have access to your child's data are bound by a duty of confidentiality. The publication of the results of the programme will be done in aggregate, i.e. without any individual data of the persons involved.

General Information

Providing Informed Consent to participate in this screening programme is completely voluntary. By agreeing, you are not waiving any of your legal rights and refusing the screening examination will not affect your child's treatment for the disease in question.