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Obligatory laboratory screenings

Karyotype

Principle: determination of karyotype from peripheral blood

Result: normal male karyotype 46, XY

Mutation CFTR – gene screening for cystic fibrosis

Principle: PCR, reverse hybridisation, analysed 19, or 34 mutations

19 mutations:

F508del, G542X, N1303K, W1282X, G551D, 1717-1GtoA, R553X, CFTRdele2,3(21kb), I507del, 711+1GtoT, 3272-26AtoG, 3905insT, R560T, 1898+1GtoA, S1251N, I148T, 3199del16, 3120+1GtoA, Q552X

34 mutations:

F508del, G542X, N1303K, W1282X, G551D, 1717-1GtoA, R553X, CFTRdele2,3(21kb), I507del, 711+1GtoT, 3272-26AtoG, 3905insT, R560T, 1898+1GtoA, 3120+1GtoA, A455E, 2143delT, Y1092X, 2184InsA, 34-5T, 35-7T, 36-9T, 621=1GtoT, Y122X, R347P, R1162X, 3849+10kbCtoT, 2183AA>G, 394delTT, 2789+5GtoA, G85E, 3659del1C, R117H, R334W, R347P, 2184delA, 1087delT

Result: negative

Spinal muscular atrophy (SMA)

The autosomal recessive neuromuscular disorder, which is the second most prevelant genetic reason of neonatal mortality. The affected individual is a homozygote. The heterozygote individuals do not show signs of the disease, but they can transmit this disorder to their offspring.

Gene SMN1 and SMN2

SMN1 exon 7, exon 8 – number of copies

SMN2 exon 7, exon 8 – number of copies

Principle: MLPA, fragmental analysis

Result: negative

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Hereditary Hearing Impairment (Connexin 26, GJB2 Gene)

Principle: exclusion of the presence of a mutation in the gene for Connexin 26 (GJB2; MIM: 121011). PCR amplification of the monitored region followed by fragmentation analysis

Mutation they are responsible for 60-80% of autosomal recessive non-syndromic losses

hearing. The investigation includes testing for the most common 35delG mutation (p. Gly12Valfs) GJB2 gene in the Caucasian population.

Result: negative, no pathogenic mutation was found

HBV (Hepatitis B)

A determination by the "surface antigene method" (HBsAg) and antibodies against nucleocapsid antigen HBV (anti-HBc), where positive HBsAg (identifies ongoing hepatitis – type B), positive anti – HBc reveals a past or persistent infection.

Principle: chemiluminescent immunoassay on paramagnetic microparticles (CMIA) electrochemiluminescent analysis ECLIA

Result: negative

HCV (Hepatitis C)

Principle: chemiluminescent immunoassay on paramagnetic microparticles (CMIA)

Result: negative

HIV (Human Immunodeficiency Virus)

HIV type 1,2 method determination of antibodies and an evidence of antigen p24

Principle: electrochemiluminiscent immunoassays ECLIA

Result: negative

Syphilis

TPHA – Detection of antibodies Treponema pallidum – haemagglutination test principle: CIMIA (Chemiluminescent Microparticle Immunoassay)

RRR – (Non-specific test for treponema detection, demonstrating non-specific antibodies)

Principle: aglutination on a microtiter plate

Result: negative

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Chlamydia trachomatis

Chlamydia trachomatis – urine examination.

Principle: qualitative determination of DNA Chlamydia trachomatis by a method RT-PCR on the analyser unit Cobas TaqMan

Result: negative