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

HCV (Hepatitis C)

principle: chemiluminescent immunoassay on paramagnetic microparticles (CMIA)

result: negative

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

HIV

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

Chlamydia trachomatis – urine examination

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

result: negative