| Trizomie | Senzitivita vyšetření | Dvojčata |
|---|---|---|
| Trizomie 21 (Downův syndrom) | 99.12% | ANO |
| Trizomie 18 (Edwardsův syndrom) | | ANO |
| Trizomie 13 (Patauův syndrom) | | ANO |
| **Další možnosti testování** | | |
| Stanovení pohlaví | 98% | NE |
| Trizomie 9 | není validováno | NE |
| Trizomie 16 | | NE |
| Trizomie 22 | | NE |
| **Aneuplodie pohlavních chromozomů** | | |
| X0 (Turnerův syndrom) | 95% | NE |
| XXY (Klinefertův syndrom) | | NE |
| XXX | | NE |
| XXY | | NE |
| X-M | | NE |
| **Delece/Duplikace** | | |
| (viz. tabulka) | není validováno | NE |

| | |
|---|---|
| 11q11-q13.3 duplication Syndrome | distal arthrogryposis 2B type(DA2B) |
| 12q14 microdeletion Syndrome | Smith-Magenis Syndrome |
| 14q11-q22 deletion Syndrome | Holoprosencephaly 4 type(HPE4) |
| 15q26 overgrowth Syndrome | Feingold Syndrome |
| 16p11.2-p12.2 microdeletion Syndrome | Diaphragmatic hernia, congenital (HCD/DIH1) |
| 16p11.2-p12.2 microduplication Syndrome | Dyggve-Melchior-Clausen Syndrome(DMC) |
| 17q21.31 deletion Syndrome | Holoprosencephaly 6 type(HPE6) |
| 17q21.31 duplication Syndrome | Jacobsen Syndrome |
| 1p36 microdeletion Syndrome | Langer-Giedion Syndrome(LGS) |
| 1q21.2 deletion Syndrome | Prader-Willi-like Syndrome |
| 1q21.2 duplication Syndrome | Rieger Syndrome1 type (RIEG1) |
| Wilms tumor 1 (WT1) | Van der Woude Syndrome (VWS) |
| 2q33.1 deletion Syndrome | Cat-eye Syndrome(CES) |
| 5q21.1-q31.2 deletion Syndrome | Monosomy 9p Syndrome |
| 8p23.1 deletion Syndrome | Orofaciodigital Syndrome |
| 8p23.1 duplication Syndrome | Panhypopituitarism, X-linked |
| Alpha Thalassemia, Mental Retardation Syndrome | Potocki-Lupski Syndrome (17p11.2 duplication Syndrome) |
| Androgen insensitivity Syndrome(AIS) | Leukodystrophy with 11q14.2- q14.3 |
| AngelmanSyndrome/Prader-Willi Syndrome | Mental retardation X-linked growth horm. Def (MRGH) |
| Aniridia II & WAGR Syndrome | Saethre-Chotzen Syndrome(SCS) |
| Bannayan-Riley-Ruvalcaba Syndrome(BRRS) | Sensorineural deafness and male infertility |
| BranchlootorenalSyndrome(BOR)/Melnick-Frazer Syndrome | Duchenne muscular dystrophy (DMD);Duchenne/Becker mascular dystrophy (DMD/BMD) |
| Microphthalmia Syndrome6 type, pituitary hypoplasia | Split-Hand/Foot Malformation 5 type (SHFM5) |
| Chromosome 10q deletion Syndrome | Split-hand/foot malformation-3 type(SHFM3) |
| Chromosome 10q22.3-q23.31 microdeletion Syndrome | Trichorhinophalangeal Syndrome1 type(TRPS1) |
| Chromosome 18p deletion Syndrome | Trichorhinophalangeal Syndrome I type |
| Chromosome 18q deletion Syndrome | Microphthalmia with linear skin defects |
| Cornelia de Lange Syndrome(CDLS) | 1q41-q42 microdeletion Syndrome |
| Cowden Syndrome(CD) | Cri du Chat(5p deletion)Syndrome |
| X-linked lymphoproliferative Syndrome(XLP) | Xp11.22-p11.23 microduplication Syndrome |
| Dandy-Walker Syndrome(DWS) | DiGeorge Syndrome2 type (DGS2) |
| | Holoprosencephaly 1 type (HPE1) |