

THE LIST OF TESTED CHROMOSOMAL DISORDERS*

1p36 microdeletion syndrome
1q21.1 susceptibility locus for thrombocytopenia-absent radius (TAR) syndrome
Van der Woude syndrome 1; VWS1
Hypoparathyroidism-retardation dysmorphism syndrome; HRD
2p15-16.1 microdeletion syndrome
Hypotonia-cystinuria syndrome, homozygous 2p21 deletion syndrome
2p21 microdeletion syndrome
Holoprosencephaly 2; HPE2
Feingold syndrome
Nephronophthisis, familial juvenile NPH1
Mowat-Wilson syndrome; MOWS
Synpolydactyly 1; SPD1
2q33.1 microdeletion syndrome
2q33.1 deletion syndrome
Chromosome 2q37 deletion syndrome
Holoprosencephaly 6; HPE6
2q37 monosomy (2q37 deletion syndrome)
Von Hippel-Lindau syndrome; VHLS
Blepharophimosis, ptosis and epicanthus inversus; BPES
Dandy-Walker syndrome; DWS
3q29 microdeletion syndrome
3q29 microduplication syndrome
Wolf-Hirschhorn syndrome; WHS
Chromosome 4q21 deletion syndrome
Axenfeld-Rieger syndrome, type 1; RIEG1
Cri-du-Chat syndrome (5p deletion)
Cornelia de Lange syndrome 1; CDLS1
Familial adenomatous polyposis; FAP1
Adult-onset autosomal dominant leukodystrophy (ADLD)
5q35 microduplication syndrome
Parietal foramina 1; PFM1
Sotos syndrome; SOTOS1
Adrenal hyperplasia, congenital due to 21-hydroxylase deficiency
Branchio-oculo-facial syndrome; BOFS
Axenfeld-Rieger syndrome, type 3; RIEG3
Chromosome 6pter-p24 deletion syndrome
Prader-Willi-like phenotype
Diabetes mellitus, transient, neonatal 1; TNDM1
Silver-Russell syndrome; SRS
Greig cephalopolysyndactyly syndrome; GCPS

Saethre-Chotzen syndrome; SCS
Williams-Beuren syndrome (WBS)
7q11.23 duplication syndrome
Split-hand/foot malformation 1 (SHFM1)
Speech-language disorder 1; SPCH1
Holoprosencephaly 3; HPE3
Currarino syndrome
Kallman syndrome 2; KAL2
8p23.1 duplication syndrome
8p23.1 deletion syndrome
CHARGE syndrome
8q21.11 microdeletion syndrome
Nablus mask-like facial syndrome; NMLFS
Chromosome 9p deletion syndrome
Chromosome 9p24.3 deletion syndrome
Basal cell nevus syndrome; BCNS
Nail-patella syndrome; NPS
9q subtelomeric deletion syndrome; Kleefstra syndrome 1; KLEFS1
Hypoparathyroidism, sensorineural deafness and renal disease; HDR Del
Di George syndrome/velocardiofacial syndrome complex 2
Chromosome 10q22.3-q23.2 deletion syndrome
Juvenile polyposis syndrome; JPS
Split-hand/foot malformation 3; SHFM3
Maternal mosaic 10q25-ter deletion
Potocki-Shaffer syndrome
WAGR 11p13 deletion syndrome
Aniridia 1; AN1
Beckwith-Wiedemann syndrome; BWS
Exudative vitreoretinopathy 1; EVR1
Jacobsen syndrome; JBS
12q14 microdeletion syndrome, Buschke-Ollendorff syndrome; BOS
Noonan syndrome 1; NS1
Peters plus syndrome; PTRPLS
Chromosome 13q14 deletion syndrome
Retinoblastoma; RB1
Holoprosencephaly 5; HPE5

*** PLEASE NOTE:** The listed disorders/syndromes can be detected with regards to the use of TRISOMY test only in cases caused by a mutation in the genetic material due to a deletion or duplication of the corresponding part of the chromosome. Other types of changes in genetic material (such as point mutation, methylation change, uniparental disomy, reciprocal translocation, inversion) cannot be detected using TRISOMY test. This is also the reason why the sensitivity of high-risk detection for individual disorders/syndromes cannot be determined explicitly. At the same time, with regards to the sensitivity of detection of deletions or duplications of a part of the chromosome, test limitations apply as referred to in the current Information (www.trisomytest.sk/en/information-concerning/TTC) taking into account specific fetal fraction levels in combination with the size of deletion or duplication found in the analysis of the biological sample examined.

Frias syndrome
Chromosome 15q11-q13 duplication syndrome
Angelman syndrome (Type 1)
Prader-Willi syndrome (Type 1)
Prader-Willi syndrome (Type 2)
Angelman syndrome (Type 2)
15q13.3 microdeletion syndrome
15q15.3 deafness and infertility syndrome
Chromosome 15q24 duplication syndrome
15q24 recurrent microdeletion syndrome
Witteveen-Kolk syndrome; WITKOS
15q26 overgrowth syndrome
Chromosome 15q26-qter deletion syndrome
16p11.2 microduplication syndrome
Recurrent 16p12.1 microdeletion (neurodevelopmental susceptibility locus)
16p11.2-p12.2 microdeletion syndrome
16p11.2-p12.2 microduplication syndrome
ATR-16 syndrome
Rubinstein-Taybi syndrome, chromosome 16p13.3 deletion syndrome, proximal
Fanconi anemia, complementation group A; FANCA
Smith-Magenis syndrome
Potocki-Lupski syndrome (17p11.2 duplication syndrome)
Charcot-Marie-Tooth syndrome type 1A (CMT1A)
Hereditary liability to pressure palsies (HNPP)
Chromosome 17p13.3, centromeric, duplication syndrome
Miller-Dieker syndrome (MDS)
Lissencephaly 1; LIS1
NF1 microduplication syndrome
NF1 microdeletion syndrome, neurofibromatosis, type 1; NF1
Renal cysts and diabetes syndrome; RCAD
17q21.31 recurrent microdeletion syndrome (Koolen de Vries syndrome)
Van Buchem disease; VBCH
Chromosome 17q23.1-q23.2 deletion syndrome
Chromosome 17q23.1-q23.2 duplication syndrome
Holoprosencephaly 4; HPE4
Chromosome 18q deletion syndrome

Pitt-Hopkins syndrome; PTHS
Diamond-Blackfan anemia 1; DBA1
Alagille syndrome 1; ALGS1
Pseudohypoparathyroidism, type 1A; PHP1A
Early-onset Alzheimer disease with cerebral amyloid angiopathy
Mental retardation, autosomal dominant 7; MRD7
Holoprosencephaly 1; HPE1
Cat-eye syndrome (Type I)
22q11 duplication syndrome
22q11.2 distal deletion syndrome
22q11 deletion syndrome (Velocardiofacial / DiGeorge syndrome)
Neurofibromatosis, type II; NF2
22q13 deletion syndrome, Phelan-McDermid syndrome; PHMDS
Xp11.22-linked intellectual disability
Xp11.22-p11.23 microduplication
Chromosome Xp21 deletion syndrome
Glycerol kinase deficiency; GKD
Adrenal hypoplasia, congenital; AHC
Muscular dystrophy, Becker type; BMD
Muscular dystrophy, Duchenne type; DMD
Albinism, ocular, type 1; OA1
Linear skin defects with multiple congenital anomalies 1; LSDMCA1
Steroid sulphatase deficiency (STS)
Hypogonadotropic hypogonadism 1 with or without anosmia; HH1, Kallmann syndrome 1; KAL1
Chondrodysplasia punctata 1, X-linked recessive; CDPX1
Léri-Weill dyschondrosteosis (LWD) - SHOX deletion 1
Léri-Weill dyschondrosteosis (LWD) - SHOX deletion 2
Agammaglobulinemia, X-linked; XLA
Pelizaeus-Merzbacher disease
Lymphoproliferative syndrome, X-linked, 1; XLP1
Mental retardation, X-linked, with panhypopituitarism
Fragile X syndrome; FXS
Xq28 microduplication
Rett syndrome; RTT
MECP2 duplication syndrome
Deafness, dystonia and cerebral hypomyelination; DDCH
SRY translocation
SRY deletion
AZFa
AZFb+AZFc