

Trizomie	Senzitivita vyšetření	Dvojčata
Trizomie 21 (Downův syndrom)		ANO
Trizomie 18 (Edwardsův syndrom)	>99,9%	ANO
Trizomie 13 (Patauův syndrom)		ANO
Trizomie		
Stanovení pohlaví	99,53%	NE
Trizomie 9		NE
Trizomie 16	>99,9%	NE
Trizomie 22		NE
Aneuploidie pohlavních chromozomů		
X0 (Turnerův syndrom)		NE
XXY (Klinefertův syndrom)	>99,9%	NE
XXX		NE
XYY		NE
Delece / Duplikace		
(viz. tabulka)	Copy Number Variations>3M / >90%	NE

Suspected disease
Chromosome 7q deletion
Chromosome 18q deletion syndrome
Chromosome 9p deletion syndrome
Chromosome 14q11-q22 deletion syndrome
Chromosome 1p36 deletion syndrome
Chromosome 6q11-q14 deletion syndrome
Chromosome 8q12.1-q21.2 deletion syndrome
Chromosome Xq21 deletion syndrome
Chromosome 1q41-q42 deletion syndrome
Chromosome 6q24-q25 deletion syndrome
Dandy-Walker syndrome
Chromosome 18p deletion syndrome
Chromosome 10q26 deletion syndrome
Chromosome 3pter-p25 deletion Syndrome
Chromosome 2p12-p11.2 deletion syndrome
Chromosome 5q14.3 deletion syndrome
Chromosome 13q14 deletion syndrome
Chromosome 10q22.3-q23.2 deletion syndrome
Levy-Shanske syndrome
Chromosome 15q26-qter deletion syndrome
Chromosome 6pter-p24 deletion syndrome

Suspected disease
Split-hand/foot malformation 5
Chromosome Xq27.3-q28 duplication syndrome
Holoprosencephaly 6
Chromosome Xp21 deletion syndrome
Cri du Chat syndrome
WAGRO syndrome
Chromosome 4q21 deletion syndrome
Yuan-Harel-Lupski syndrome
Cat-Eye syndrome
Jacobsen syndrome
DiGeorge syndrome 2
Langer-Giedion syndrome
Chromosome 2p16.1-p15 deletion syndrome
Chromosome 4q32.1-q32.2 triplication syndrome
Chromosome 16p12.2-p11.2 deletion syndrome
Chromosome Xp11.23-p11.22 duplication syndrome
Chromosome 2q31.1 duplication syndrome
Chromosome 2q33.1 deletion syndrome
Chromosome Xq28 deletion syndrome
Chromosome 22q11.2 duplication syndrome
Chromosome 16p deletion syndrome
Chromosome 16p13.3 deletion syndrome
Chromosome 16p11.2-p12.2 microduplication syndrome
Chromosome 5q12 deletion syndrome
Chromosome 1p32-p31 deletion syndrome
Chromosome 1p31 duplication syndrome
Chromosome 16q22 deletion syndrome
Frias syndrome
Chromosome 15q11-q13 duplication syndrome
Chromosome 16p deletion syndrome
Chromosome 16p13.3 deletion syndrome
Chromosome 16p11.2-p12.2 microduplication syndrome
Chromosome 5q12 deletion syndrome
Chromosome 1p32-p31 deletion syndrome
Chromosome 1p31 duplication syndrome

Suspected disease
Chromosome 16q22 deletion syndrome
Frias syndrome
Chromosome 15q11-q13 duplication syndrome
CHDM
Chromosome 15q14 deletion syndrome
Chromosome 17q12 duplication syndrome
Chromosome 17q12 deletion syndrome
Chromosome 3q29 duplication syndrome
Chromosome 8q22.1 duplication syndrome
Chromosome 8q22.1 deletion syndrome
Prader-Willi/Angelman syndrome
Holoprosencephaly 1
WAGR syndrome
Chromosome 7q11.23 deletion syndrome
Chromosome 7q11.23 duplication syndrome
Potocki-Shaffer syndrome
HCD
Chromosome Xq22.3 telomeric deletion syndrome
Wolf-Hirschhorn syndrome
Chromosome 17q21.31 duplication syndrome
Chromosome Xp11.3 deletion syndrome
Chromosome 3q13.31 deletion syndrome
Chromosome 8p23.1 deletion syndrome
Chromosome 8p23.1 duplication syndrome
Chromosome 12q14 microdeletion syndrome
Chromosome 17q23.1-q23.2 deletion syndrome
Potocki-Lupski syndrome
Smith-Magenis syndrome
Chromosome 17p13.3 duplication syndrome
Chromosome 17p13.3 deletion syndrome
Chromosome 19q13.11 deletion syndrome
Chromosome 2q35 duplication syndrome
Chromosome 15q25 deletion syndrome
Chromosome 22q11.2 deletion syndrome
DiGeorge syndrome