Current Constitutional FISH tests provided by the Clinical Cytogenetics Department

Please note that microarray analysis will identify all imbalances detected by FISH and if negative for a specific FISH test, microarray may identify another imbalance of clinical significance.

Disorder	Chromosomal location/probe
Subtelomere deletion syndromes, including: Monosomy 1p36 syndrome Monosomy 2q37 syndrome Monosomy 6p25 syndrome Kleefstra syndrome (monosomy 9q34.3) Monosomy 22q13.3	• 1p36.13~33 • 2q37 • 6p25 • 9q34.3 • 22q13.3
Wolf Hirschhorn syndrome	• 4p16
Cri du Chat syndrome	• 5p15
Saethre-Chotzen	• 7p21.1
Williams syndrome	• 7q11.23
7q11.23 microduplication syndrome	• 7q11.23
Langer Giedion syndrome/Tricho-Rhino-Phalangeal syndrome	• 8q24.11-q24.13
Patau syndrome (Trisomy 13)	• 13
Prader Willi microdeletion*/microduplication	• 15q11.2
Angelman microdeletion syndrome*	• 15q11.2
Smith Magenis syndrome	• 17p11.2
17p11.2 Potocki-Lupski microduplication syndrome	• 17p11.2
Miller Dieker syndrome	• 17p13.3
Edwards Syndrome (Trisomy 18)	• 18
Alagille Syndrome	• 20p12 JAG1
Down Syndrome (Trisomy 21)	• 21
DiGeorge/Velo-Cardio-Facial syndrome	• 22q11
Kallman syndrome	• Xp22.3
Turner syndrome (Monosomy X)	• X centromere
X-linked ichthyosis	• Xp22.3 STS
Leri-Weill dyschondrosteosis Langer mesomelic dysplasia	• Xp22 SHOX
Assess X chromosome for inactivation functionality	• Xq13 XIST
Testis determining factor	• Yp11.3 SRY

*For first line see molecular testing

Please note that this is not an exhaustive list. Please contact the Clinical Cytogenetics Department if you require additional information.

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