

DISEASE	REGION	SYNONYM	METHOD	SAMPLE	AMOUNT	TURN AROUND TIME
Alagille Syndrome, AGS	20p12 deletion	Arteriohepatic Dysplasia	FISH	Blood <sup>Hep</sup>	5 ml	1 Month
Angelman Syndrome, AS	15q11 - q13 deletion	Happy Puppet Syndrome	FISH	Blood <sup>Hep</sup>	5 ml	20 Days
Beckwith-Widemann Syndrome, BWS	11p15.1 - p15.2 duplication	Exomphalos-Macroglossia-Gigantism	FISH	Blood <sup>Hep</sup>	5 ml	1 Month
Charcot-Marie-Tooth Type 1, CMT1A	17p11.2 duplication		FISH	Blood <sup>Hep</sup>	5 ml	1 Month
Cri du Chat Syndrome	5p15.2 deletion	Cat Cry Syndrome	FISH	Blood <sup>Hep</sup>	5 ml	1 Month
Digeorge Syndrome, DGS Velocardiofacial Syndrome, VCF SHPRINTZEN CATCH22	22q11.2 / 22q13 deletion		FISH	Blood <sup>Hep</sup>	5 ml	20 Days
Ichthyosis Due To Steroid Sulfatase Deficiency	Xp22.32 deletion	Steroid Sulfatase Mikrodeletion	FISH	Blood <sup>Hep</sup>	5 ml	1 Month
Kallmann 1 Syndrome, KAL1	Xp22.3 / Xp11.1-q11.1 deletion	Hypogonadotropic Hypogonadism And Anosmia	FISH	Blood <sup>Hep</sup>	5 ml	1 Month
Kallmann 2 Syndrome, KAL2	8p11.2 deletion	Hypogonadotropic Hypogonadism And Anosmia	FISH	Blood <sup>Hep</sup>	5 ml	1 Month
Langer-Giedon Syndrome, LGS	8q24 deletion	Tricho Rhino Phalangeal Syndrome Type 2	FISH	Blood <sup>Hep</sup>	5 ml	1 Month
Mental Retardation and Congenital Heart Defect	8p23.1 deletion		FISH	Blood <sup>Hep</sup>	5 ml	1 Month
Mental Retardation	1p36 deletion		FISH	Blood <sup>Hep</sup>	5 ml	1 Month
Miller-Dieker Syndrome, MDLS, MDS	17p13.3 deletion	Lissencephaly	FISH	Blood <sup>Hep</sup>	5 ml	1 Month
Neurofibromatosis Type1, NF1	17q11.2 deletion		FISH	Blood <sup>Hep</sup>	5 ml	1 Month
Prader Willi Syndrome, PWS	15q11 - q13 deletion		FISH	Blood <sup>Hep</sup>	5 ml	20 Days
Rubinstein-Taybi Syndrome, RSTS	16p13.3 deletion	Broad Thumb-Hallux Syndrome	FISH	Blood <sup>Hep</sup>	5 ml	1 Month
Short Stature, SS	Xpter - p22.32 deletion		FISH	Blood <sup>Hep</sup>	5 ml	1 Month
Smith-Magenis Syndrome, SMS	17p11.2 / 17p13.3 deletion		FISH	Blood <sup>Hep</sup>	5 ml	20 Days
SOTOS	5q35 deletion	Cerebral Gigantism	FISH	Blood <sup>Hep</sup>	5 ml	1 Month



## BURC GENETIC DIAGNOSTIC CENTER MICRODELETIONS TEST LIST (2010)

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TSC2 - PKD1 DELETION	16p13.3 - p13.12 deletion		FISH	Blood <sup>Hep</sup>	5 ml	1 Month
WAGR	11p13 deletion	Wilms tumor, wt1,	FISH	Blood <sup>Hep</sup>	5 ml	1 Month
WILLIAMS-BEUREN, WBS	7q11.23 / 7q31 deletion	Williams	FISH	Blood <sup>Hep</sup>	5 ml	1 Month
Wolf-Hirschhorn Syndrome, WHS	4p16.1 / 4p11-q11 deletion		FISH	Blood <sup>Hep</sup>	5 ml	1 Month