

Postnatal Constitutional Chromosome Analysis

Test	Description	Turn Around	CPT codes
Blood Chromosome analysis, Routine	Chromosome analysis of PHA stimulated peripheral blood leukocytes	5-8 days	88230, 88262, 88291
Blood Chromosome Analysis, High resolution	High resolution chromosome analysis of PHA stimulated peripheral blood leukocytes	5-8 days	88230, 88262, 88289, 88291
Specimen requirements: 5-6 ml whole blood in sodium heparin (green topped) tubes. (Minimum of 2 ml for pediatric patients)			
Tissue Chromosome analysis	Chromosome analysis of fibroblast cultures	2-4 weeks	88233, 88262, 88291
Specimen requirements: Skin biopsy, 3x3mm, in sterile transport medium or sterile saline			

Postnatal Constitutional FISH Analysis

Test	Description	Turn Around	CPT codes
Aneuploidy Screen	Interphase analysis to screen for numerical anomalies of chromosomes X, Y, 13, 18, and 21	24-48 hours	88275, 88271(x5), 88291
1p36 Microdeletion	1p36	4-6 days	88275, 88271(x2), 88291, 88230*
Angelman	15q11q13	4-6 days	88275, 88271(x4), 88291, 88230*
Cri du Chat	5p15.2	4-6 days	88275, 88271(x2), 88291, 88230*
DiGeorge / VCFS	22q11.2	4-6 days	88275, 88271(x2), 88291, 88230*
Distal 22q11.2 [BCR gene] duplication / deletion	22q11.23	4-6 days	88275, 88271(x2), 88291, 88230*
Kallmann	Xp22.3	4-6 days	88275, 88271(x2), 88291, 88230*
Miller-Dieker	17p13.3	4-6 days	88275, 88271(x2), 88291, 88230*
Prader-Willi	15q11q13	4-6 days	88275, 88271(x4), 88291, 88230*
Retinoblastoma [RB1 gene]	13q14.2	4-6 days	88275, 88271(x2), 88291, 88230*
Smith-Magenis	17p11.2	4-6 days	88275, 88271(x2), 88291, 88230*
SRY	Yp11.3	4-6 days	88275, 88271(x2), 88291, 88230*
Steroid Sulfatase [STS]	Xp22.3	4-6 days	88275, 88271(x2), 88291, 88230*
Williams	7q11.23	4-6 days	88275, 88271(x2), 88291, 88230*
Wolf-Hirschhorn	4p16.3	4-6 days	88275, 88271(x2), 88291, 88230*
Specimen requirements: 5-6 ml whole blood in sodium heparin (green topped) tubes. (Minimum of 2 ml for pediatric patients)			
* No additional blood is necessary if chromosome studies are also being processed.			