



## Pediatric Testing CPT Codes

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CYTOGENETICS/MOLECULAR CYTOGENETICS TESTING		
Microarrays and Karyotype		
Test #	Test Title	CPT Code(s)
100	Rapid microarray (CGH and SNP)	81229 x1
200	Standard karyotype	88230 x2, 88262 x1, 88291 x1
204	Karyotype for mosaicism	88230 x2, 88263 x1, 88291 x1
210	5-cell karyotype + microarray bundle	81229 x1, 88230 x1, 88261 x1, 88291 x1, 88285 may be added if additional cells are analyzed
Familial Studies		
230	5-cell karyotype (parent or family member of proband)	88230 x1, 88261 x1, 88291 x1, 88285 may be added if additional cells are analyzed
330	Locus-specific FISH (parent or family member of proband)	88230 x1, 88271 x2, 88273 x2, 88291 x1
MOLECULAR TESTING		
Exome Testing		
Test #	Test Title	CPT Code(s)
8910	Whole Exome Via NGS (Proband only)	81415 x1
8911	Whole Exome via NGS (Trio with proband report only)	81415 x1, 81416 x2
8912	Whole Exome via NGS (Trio with full parental reports)	81415 x1, 81416 x2
8913	Reflex from Panel to Whole exome via NGS (Proband only)	81415 x1
8915	Whole exome Via NGS (Duo with full parental report)	81415 x1, 81416 x1
Single Gene Testing		
8560	Known mutation evaluation	Dependent upon testing performed.
8830	Single gene sequencing	Dependent upon testing performed.
8835	Single gene sequencing and deletion/duplication analysis	Dependent upon testing performed.
Phenotype-Based Panels		
8230	Congenital hypotonia panel	81400 x1, 81401 x1, 81331 x1
8230R	Congenital hypotonia panel – Reflex to Southern blot for <i>DMPK</i> (ordered if initial panel is positive for <i>DMPK</i> )	81239 x1
8280	Custom NGS panel	Dependent upon testing performed.
Syndrome-Based Testing		
8115	Beckwith-Wiedemann syndrome: H19 & LIT1 methylation	81401 x1
8380	Fragile X CGG repeat analysis	81243 x1
8381	Fragile X CGG repeat analysis for buccal specimens	81243 x1
8750	Prader-Willi/Angelman syndrome methylation testing	81331 x1