UPMC
LIFE CHANGING MEDICINE

# UPMC CYTOGENETICS LABORATORY

## Constitutional FISH Probe Reference List

Please contact the lab (pittcytogenetics@upmc.edu) for FISH testing using probes that do not appear on this list.

| Common Microdeletion/Duplication Syndromes | | | |
|---|---|---|---|
| **Locus** | **Target Genomic Location** | **Condition/Gene** | **OMIM # or PMID** |
| **1q21.1** | chr1:147,107,518-147,294,532 | 1q21.1 deletion syndrome | 612474 |
| | | 1q21.1 duplication syndrome | 612475 |
| **1q21.1** | chr1:145,523,005-145,685,541 | TAR syndrome | 274000 |
| **3q29** | chr3:195,907,411-196,071,593 | 3q29 deletion syndrome | 609425 |
| | | 3q29 duplication syndrome | 611936 |
| **7q11.23** | chr7:73,442,427-73,684,236* | Williams-Beuren syndrome | 194050 |
| | | Williams-Beuren region duplication syndrome | 609757 |
| **10q22.3q23.2** | chr10:85,901,343-86,099,824 | 10q22.3q23.2 deletion syndrome | 612242 |
| **15q11.2** | chr15:22,877,142-23,026,834 | 15q11.2, BP1-BP2 deletion | 615656 |
| **15q13.1** | chr15:29,936,698-30,124,815 | Prader-Willi Syndrome | 176270 |
| | | Angelman Syndrome | 105830 |
| **15q13.3** | chr15:32,219,234-32,378,020 | 15q13.3 deletion (*CHRNA7* gene) | 612001 |
| **16p13.12p13.11** | chr16:15,569,590-15,781,930 | 16p13.11 deletion | PMID:21150890 |
| | | 16p13.11 duplication | PMID:18550696 |
| **16p11.2** | chr16:15,569,590-15,781,930 | 16p11.2 deletion syndrome | 611913 |
| | | 16p11.2 duplication syndrome | 614671 |
| **17p13.3** | chr17:674,915-844,565 | Miller-Dieker lissencephaly syndrome | 247200 |
| | | 17p13.3 duplication syndrome | 613215 |
| **17p13.1** | chr17:7,397,449--7,744,913* | 17p13.1 deletion syndrome (*TP53* gene) | 613776 |
| **17p12** | chr17: 15,106,256-15,282,725 | Hereditary neuropathy with liability to pressure palsies (*PMP22* gene deletion) | 162500 |
| | | Charcot Marie Tooth Type 1A (*PMP22* gene) | 118220 |
| **17p11.2** | chr17:19,456,850-19,645,771 | Smith-Magenis syndrome (*RAI1* gene deletion) | 182290 |
| | | Potocki-Lupski syndrome (*RAI1* gene duplication) | 610883 |
| **17q11.2** | chr17:29,353,307-29,827,962* | 17q11.2 deletion syndrome (*NF1* gene) | 613675 |
| | | 17q11.2 duplication syndrome (*NF1* gene) | 618874 |

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| | | | |
|---|---|---|---|
| **17q12** | chr17:36,055,082-36,*227*,148 | 17q12 deletion/duplication syndrome<br>17q12 syndrome | 614527<br>614526 |
| **22q11.1** | chr22:17,722,544-17,910,640 | Cat Eye syndrome | 115470 |
| **22q11.2** | chr22:19,288,224-19,419,219* | 22q11.2 deletion syndrome/DiGeorge/VCF syndrome (*HIRA* gene) | 188400<br>192430 |
| | | 22q11.2 duplication syndrome (*HIRA* gene) | 608363 |
| | chr22:21,213,744-21,406,241 | Central 22q11.2 deletion/duplication syndrome (*CRKL* gene) | PMID: 26278718<br>608363 |
| | chr22:22,185,268-22,419,019 | Distal 22q11.2 deletion/duplication syndrome (*MAPK1* gene) | 611867<br>608363 |
| | chr22:24,054,783-24,251,617 | Distal 22q11.2 deletion/duplication syndrome (*SMARCB1* gene) | 611867<br>608363 |
| **Xp22.3** | chrX:7,130,472-7,352,682* | STS Deficiency (*STS* gene) | PMID: 23791652 |
| **Xq13.2** | chrX:73,000,486-73,092,588* | XIST gene | |
| **Xq28** | chrX:153,078,744-153,238,727 | MECP2 duplication syndrome (*MECP2* gene) | 300260 |

*Approximate coordinates

**For Questions about CPT codes:**

1. Billing codes will differ depending on what type of copy number variant we are assessing. In general, there is one set of codes used for deletions and one for duplications. Please see below for corresponding CPT codes
2. Please see the "Documents" Page on our website for a full list of CPT codes for cytogenetic testing.
3. With any additional questions, please contact us at PittCytogenetics@upmc.edu

| TEST REQUESTED | CPT Codes |
|---|---|
| **FISH for Deletion** | 88230, 88273, 88271 (x2) |
| **FISH for Duplication** | 88230, 88274, 88271 (x2) |