

Table S1: Inclusion Criteria and Genomic Coordinates Used to Indicate Copy Number Variant Confirmation Testing

Copy Number Variant	Criteria	Genomic Coordinates (GRCh38/hg38; Mb)
1q21.1 (<i>GJA5</i>) del*	Size >50% of critical region	chr1:147.11-147.92
1q21.1 (<i>GJA5</i> + <i>TAR</i>) del*	Size >50% of critical region	chr1:145.63-147.92
7q11.23 (<i>ELN</i>) del	Size >50% of critical region	chr7:73.33-74.73
15q13.3 (<i>CHRNA7</i>) del	Size >50% of critical region	chr15:30.84-32.15
15q24 (<i>SIN3A</i>) del	At least 1 Mb between the A-E intervals	chr15:72.67-75.68
16p13.11 (<i>MYH11</i>) del	Size >50% of critical region	chr16:15.42-16.20
16p11.2 (<i>TBX6</i>) del	Size >50% of critical region	chr16:29.64-30.19
17q11.2 (<i>NF1</i>) del	Size >50% of critical region, affecting <i>NF1</i>	chr17:30.78-31.94
17q12 (<i>HNF1B</i>) del	Size >50% of critical region	chr17:36.46-37.85
22q11.2 del (proximal, A-B)#	Size >50% of critical region	chr22:18.92-20.29
22q11.2 del (proximal, A-D)#	Size >50% of critical region	chr22:18.92-21.11

* - included as 1q21.1 deletions; # - included as 22q11.2 deletions