**Table S1**. Coding genes in the 15q26.3-qter and in the 5p13.3-pter regions involved in our patients’ rearrangement that were predicted as likely pathogenic or pathogenic by the AnnotSV tool and their respective haploinsufficiency or triplosensitivity scores.

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| Region | Gene | AnnotSV Score | OMIM Phenotype | HI (gnomAD) | HS(ClinGen) | pLI(gnomAD) | TS(ClinGen) |
| 15q26.3 | *MEF2A* | Likely Pathogenic | Coronary artery disease 1 (608320) | **6.03%** | - | **0.990** | NA |
| *ADAMTS17* | Pathogenic | Weill-Marchesani 4 syndrome (613195) | 44.61% | AR | 0.000 | NA |
| *CERS3* | Pathogenic | Ichthyosis congenital 9 (615023) | 62.25% | AR | 0.001 | NA |
| *ASB7* | Likely Pathogenic | - | 29.00% | - | **0.980** | NA |
| *CHSY1* | Pathogenic | Temtamy preaxial brachydactyly syndrome (605282) | 54.01% | AR | 0.710 | NA |
| *SNRPA1* | Likely Pathogenic | - | **10.49%** | - | **0.990** | NA |
| 5p15.33 | *TERT* | Likely Pathogenic | Dyskeratosis congenita (613989), Pulmonary fibrosis and/or bone marrow failure, telomere-related (614742) | NA | NA | NA | 0 |
| *SLC6A3* | Likely Pathogenic | Parkinsonism-dystonia (613135) | NA | NA | NA | - |
| 5p15.31 | *MTRR* | Likely Pathogenic | Homocystinuria-megaloblastic anemia (236270) | NA | NA | NA | 0 |
| 5p15.2 | *DNAH5* | Pathogenic | Ciliary dyskinesia (608644) | NA | NA | NA | 0 |

HI: Haploinsufficiency Index (lower values indicate that a gene is more likely to exhibit haploinsufficiency); pLI: higher values indicate a higher probability that a gene is intolerant to a heterozygous loss of function; HS: Haploinsufficiency score; AR: Gene associated with an autosomal recessive phenotype (HS=30); TS: Triplosensitivity (Score =0 indicates that there is no evidence for dosage pathogenicity); NA: Not applicable.