Supplementary Material

Genetic mosaicism in a group of patients with Cornelia de Lange Syndrome.

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**Table 1.** List of all tested genes in NGS analysis and numbers of transcripts.

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| --- | --- |
| Gene | Transcript |
| NIPBL | NM\_113433.3 |
| SMC1A | NM\_006306.2 |
| SMC3 | NM\_005445.3 |
| RAD21 | NM\_006265.2 |
| HDAC8 | NM\_018486.2 |
| STAG1 | NM\_005862.2 |
| SGOL1 | NM\_001012410.4 |
| PDS5A | NM\_001100399.1 |
| PTTG1 | NM\_001282382.1 |
| TAF6 | NM\_005641.3 |
| ESCO2 | NM\_001017420.2 |
| RECQL4 | NM\_004260.3 |
| WAPAL | NM\_015045.3 |
| CDCA5 | NM\_080668.3 |
| KMT2A | NM\_012308.2 |
| DDX11 | NM\_152438.1 |
| ESPL1 | NM\_012291.4 |
| PDS5B | NM\_015032.3 |
| PLK1 | NM\_005030.5 |
| AURKB | NM\_001284526.1 |
| ESCO1 | NM\_052911.2 |
| MAU2 | NM\_015329.3 |
| ATRX | NM\_000489.3 |
| STAG2 | NM\_001042749.2 |

**Table 2. List of primers and annealing temperatures.**

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| --- | --- | --- | --- | --- |
| Case no | Exon/ intron | Forward (5’-3’) | Revers (5’-3’) | Annealing (C) |
| NIPBL - DNA |
| CdLS02/ CdLS02M | 8-9A | CACAACTGTTACTTCTATCGA | TGTCCTGGGTACTGCACA | 63-56 |
| CdLS03/ CdLS45 | 28-29 | ACAAAAATGTCAATGTT | TTTTTCTAACTTAACACTTT | 51 |
| CdLS04 | 26 | CATCAAGCTCAAGTCTGTCTAA | ATAAATGTAAACTGTTTTCTCCTT | 63-56 |
| CdLS11/ CdLS42 | 17 | TCCACCAGTGAAAATCAAATC | TTTTGGTGCCATTTTAAGTCC | 68-61 |
| CdLS20 | 9B | AACTTACATTAAGACTTTCTC | ATGATGAAGTGGTAATAATGAG | 63-56 |
| CdLS22 | 12 | TTGTCATGGGGATTTGCTTC | CTTAAACGATAATCACTGCAC | 63-56 |
| CdLS24 | 40 | GTAAAGTTAGTATAGGTGCTC | AGTGCTGCTGCCATAAAACA | 63-56 |
| CdLS33/ CdLS55 | 35-36 | ATATAGTTTCTTTTCAGGTTTTGGAT | GCAGAATAATTTTAAAACCCTGGTA | 68-61 |
| CdLS54 | 10E | TGAATCAGAGCGACATCGAG | CCCCCAACAAATAACTTGGAA | 63-56 |
| CdLS58 | 42 | TGAAGCTAGCCTCAGAATGTAA | ACTTAATGAAGTTACTATCATATAC | 63-56 |
| CdLS62 | 32 | GGCTAAAGCATAACAAAAGTATATT | AATAAATTTTCCCTACCAAAAGAA | 63-56 |
| CdLS63/ CdLS53 | 10D | AGTGATGGGCATCCTGAAAC | CTTCGAACCCTAGACTGATC | 63-56 |
| CdLS68 | 47 | GCGTCAAGGGATTAAAAGCA | ATGTTTGCCCAACATTTCCTTT | 63-56 |
| HDAC8 - DNA |
| CdLS21 | 10 | TGTGGTTGGTTTGGAGGTTT | ATTTAAGGAAGCCCCCACTG | 60 |
| CdLS65 | 6 | CCTCCTTCTCACAGCTGCTT | TTGCATAGGATTGTGGCAGT | 59 |
| SMC1A |
| CdLS05 | 10 | CTGTCCCCCTTTCCATACTG | TCCAGTACTGAGCCTGTCCA | 62 |
| CdLS29 | 6 | CTGGGAACCCTCCCTTCTT | TCTGTGGACCATCATCCCTA | 58 |
| CdLS59 | 5 | TTTTCTGGGTGAAGGAGTGG | ATAAACAGCACGGCCTCTTG | 58 |
| KMT2A - DNA |
| CdLS09 | 7 | CCGTCGAGGAAAAGAGTGAA | GAGAGCTGATTTTCTTGAACTGAA | 58 |
| NIPBL - cDNA |
| CdLS03 | 27-31 | TTGATATTTATTTGACACAGATCCT | GAATGTTTCATTTACTAATTTCTTAA | 60 |
| CdLS11 | 17 | GGTATAATGGATAAGCTTTC | CCCATAGATGAAACCTGAA |   |
| CdLS20 | 9B | GACTTTCTCGTGTAAGGTCTTCAG | TCTGTTGTTGATTCTGTACCACA |   |
| CdLS02/ CdLS02M | 7-10 | TGAGAGTCCACTCTTCCTGC | TGAGAGTCCACTCTTCCTGC | 60 |
| CdLS24 | 38-42 | AAGCTATCATTGGTCTAGGATTT | AGTCACGTCTGTTTTTGCTG | 55 |
| CdLS62 | 30-35 | ATGAAGAGGGCATTAAGAAATT | GAAATCATTTTGCGTACTAC | 54 |
| SMC1A - cDNA |
| CdLS29 | 4-09 | CAGGAGAAAGAAGAGGCTGA | TCTTCTAGGGACTGCTTGCT | 58 |
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