

Supplemental Table 3**CNVs found in DGV with partial overlap of the three duplicated regions of interest (1q41, 2q37.3, 8q24.3)**

Number of described CNVs (examined cases in total)	Deletion/Duplication	Complete or partial overlapping regions	Size	References
chr1:215,945,774-216,077,064				
1 (1)	Duplication	partial	3,216 bp	¹
1 (1)	Deletion	partial	313 bp	¹
1 (1)	Duplication	partial	1 bp	²
1 (1)	Deletion	partial	314 bp	²
1 (1)	Deletion	partial	807 bp	³
chr2:241,202,666-241,227,781				
1 (270)	Deletion/Duplication*	complete	223,751 bp	⁴
1 (30)	Duplication	partial	7,321 bp	⁵
1 (1)	Duplication	partial	1,121 bp	⁶
3 (39)	Deletion	partial	1,121 bp	⁷
1 (1)	Duplication	partial	1 bp	²
chr8:145,012,210-145,132,100				
4 (30)	Duplication	partial	18,827 bp	⁸
1 (30)	Deletion	partial	18,827 bp	⁸
3 (1854)	Deletion	complete	288,952 bp	⁹

1 (1)	Duplication	partial	12,354 bp	10
1 (1)	Duplication	partial	19,170 bp	10
1 (1)	Duplication	partial	13,136 bp	10
1 (1)	Duplication	partial	12,032 bp	10
1 (1)	Duplication	partial	6,612 bp	10
1 (1)	Duplication	partial	2,895 bp	10
1 (30)	Duplication	partial	3,543 bp	8
4(30)	Duplication	partial	1,509 bp	8
1 (1)	Duplication	partial	1,069 bp	10
1 (1)	Duplication	partial	5,392 bp	10
1 (1)	Duplication	partial	1 bp	2
1 (36)	Deletion	partial	121 bp	11
1 (30)	Duplication	partial	38,482 bp	8
1 (30)	Duplication	partial	5,615 bp	8
1 (30)	Duplication	partial	1,646 bp	8
1 (485)	Deletion	partial	131,328 bp	12
2 (485)	Deletion	partial	651,337 bp	12
3 (485)	Deletion	partial	420,471 bp	12
8 (485)	Deletion	partial	183,427 bp	12
5 (485)	Deletion	partial	89,147 bp	12
8 (2026)	Deletion	partial	67,323 bp	13
1 (30)	Deletion	partial	776 bp	8

2 (2026)	Deletion	partial	52,239 bp	¹³
2 (2026)	Deletion	partial	11,168 bp	¹³
1 (112)	Deletion	partial	11,168 bp	¹⁴
3 (30)	Duplication	partial	6,171 bp	⁸
1 (30)	Deletion	partial	6,171 bp	⁸
1 (1)	Deletion	partial	6,072 bp	¹⁵

* Number of duplications or deletions not specified.

References

- 1 Pang AW, MacDonald JR, Pinto D *et al*: Towards a comprehensive structural variation map of an individual human genome. *Genome Biol* 2010; **11**: R52.
- 2 Levy S, Sutton G, Ng PC *et al*: The diploid genome sequence of an individual human. *PLoS Biol* 2007; **5**: e254.
- 3 Ahn SM, Kim TH, Lee S *et al*: The first Korean genome sequence and analysis: full genome sequencing for a socio-ethnic group. *Genome Res* 2009; **19**: 1622-1629.
- 4 Redon R, Ishikawa S, Fitch KR *et al*: Global variation in copy number in the human genome. *Nature* 2006; **444**: 444-454.
- 5 Perry GH, Ben-Dor A, Tselenko A *et al*: The fine-scale and complex architecture of human copy-number variation. *Am J Hum Genet* 2008; **82**: 685-695.
- 6 Ju YS, Hong D, Kim S *et al*: Reference-unbiased copy number variant analysis using CGH microarrays. *Nucleic Acids Res* 2010; **38**: e190.

- 7 Conrad DF, Pinto D, Redon R *et al*: Origins and functional impact of copy number variation in the human genome. *Nature* 2010; **464**: 704-712.
- 8 Park H, Kim JI, Ju YS, *et al*: Discovery of common Asian copy number variants using integrated high-resolution array CGH and massively parallel DNA sequencing. *Nat Genet* 2010; **42**: 400-405.
- 9 Itsara A, Cooper GM, Baker C *et al*: Population analysis of large copy number variants and hotspots of human genetic disease. *Am J Hum Genet* 2009; **84**: 148-161.
- 10 Kidd JM, Cooper GM, Donahue WF *et al*: Mapping and sequencing of structural variation from eight human genomes. *Nature* 2008; **453**: 56-64.
- 11 Mills RE, Luttig CT, Larkins CE *et al*: An initial map of insertion and deletion (INDEL) variation in the human genome. *Genome Res* 2006; **16**: 1182-1190.
- 12 Jakobsson M, Scholz SW, Scheet P *et al*: Genotype, haplotype and copy-number variation in worldwide human populations. *Nature* 2008; **451**: 998-1003.
- 13 Shaikh TH, Gai X, Perin JC *et al*: High-resolution mapping and analysis of copy number variations in the human genome: a data resource for clinical and research applications. *Genome Res* 2009; **19**: 1682-1690.
- 14 Wang K, Li M, Hadley D *et al*: PennCNV: an integrated hidden Markov model designed for high-resolution copy number variation detection in whole-genome SNP genotyping data. *Genome Res* 2007; **17**: 1665-1674.
- 15 Kim JI, Ju YS, Park H *et al*: A highly annotated whole-genome sequence of a Korean individual. *Nature* 2009; **460**: 1011-1015.