Parameters

- OMKar version: xxx
- Genome assembly: hg38
- Genes: protein coding
- Breakpoint Gene Reporting Proximity: 50
- Threshold for event insertion size: 200
- Threshold for event deletion size: 200
- Supported SV types:
    - Deletion
    - Inversion
    - Single/repeated Tandem-duplication
    - Left/right Duplication-inversion
    - 2/multi-break Reciprocal-balanced-translocation
    - Nonreciprocal-balanced-translocation
    - Duplicated-insertion

Sample Id: 3

Event Cluster 1 (of 6)

SVs

1. inv(2)(q12.2q12.3). inversion on
Chr2: 106,462,406-107,833,526 (q12.2 - q12.3)

2. dup(2)(q14.3). tandem duplication on
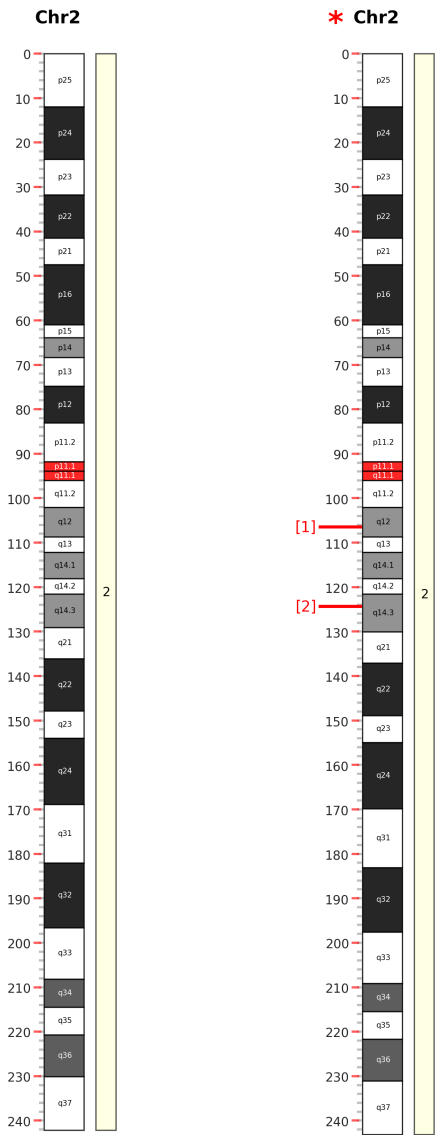Chr2: 124,318,296-125,307,772 (q14.3 - q14.3)

Impacted genes in DDG2P

None

Figure 1: karyotype 3, cluster 1

Event Cluster 2 (of 6)

SVs   **1. inv(9)(q22.32q22.33)**. inversion on Chr9: 94,438,791-96,949,317 (q22.32 - q22.33)

2. del(9)(p21.1). deletion on Chr9: 28,498,341-28,758,302 (p21.1 - p21.1)

Impacted genes in DDG2P
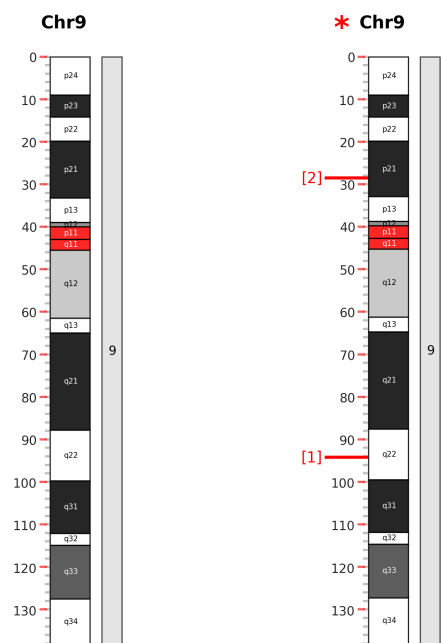
None

Event Cluster 3 (of 6)

Figure 2: karyotype 3, cluster 2

SVs

1. inv(10)(q11.22). inversion on
Chr10: 46,991,593-48,060,267 (q11.22 - q11.22)
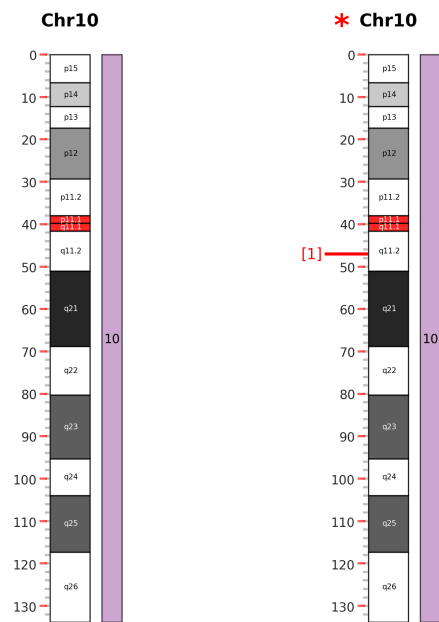
Impacted genes in DDG2P

None

Figure 3: karyotype 3, cluster 3

Event Cluster 4 (of 6)

SVs

1. del(15)(q14). deletion on
Chr15: 34,346,216-34,614,876 (q14 - q14)

Impacted genes in DDG2P

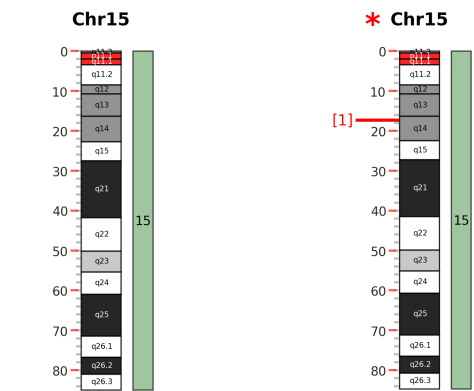| SV	Rationale	Gene Name	Gene Omim
1	breakpoint proximal	NOP10	606471
1	breakpoint proximal	SLC12A6	604878
1	breakpoint proximal	OTUD7A	612024
1	CN-1	NOP10	606471
1	CN-1	SLC12A6	604878

Figure 4: karyotype 3, cluster 4

Event Cluster 5 (of 6)

SVs

1. inv(16)(p12.2). inversion on
Chr16: 21,502,978-22,436,123 (p12.2 - p12.2)
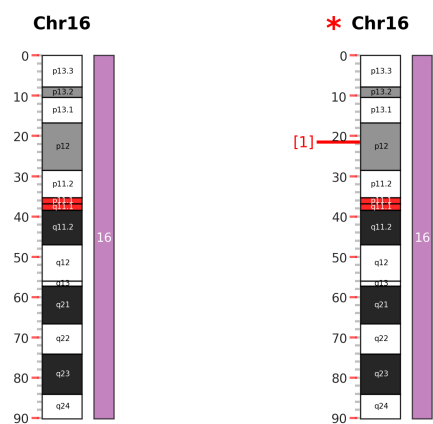
Impacted genes in DDG2P

None

Figure 5: karyotype 3, cluster 5

Event Cluster 6 (of 6)

SVs

1. inv(17)(p11.2). inversion on
Chr17: 16,806,184-18,509,508 (p11.2 - p11.2)
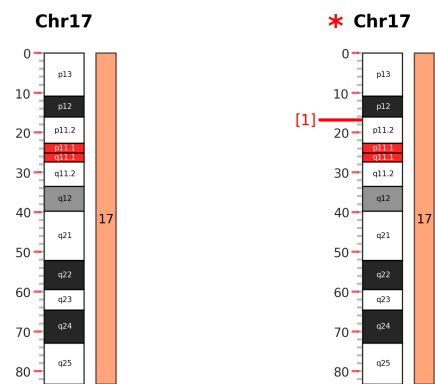
Impacted genes in DDG2P

None

Figure 6: karyotype 3, cluster 6

Sample Id: 12

Event Cluster 1 (of 4)

SVs

1. dup(4)(p16.1). tandem duplication on
Chr4: 8,929,610-9,144,574 (p16.1 - p16.1)
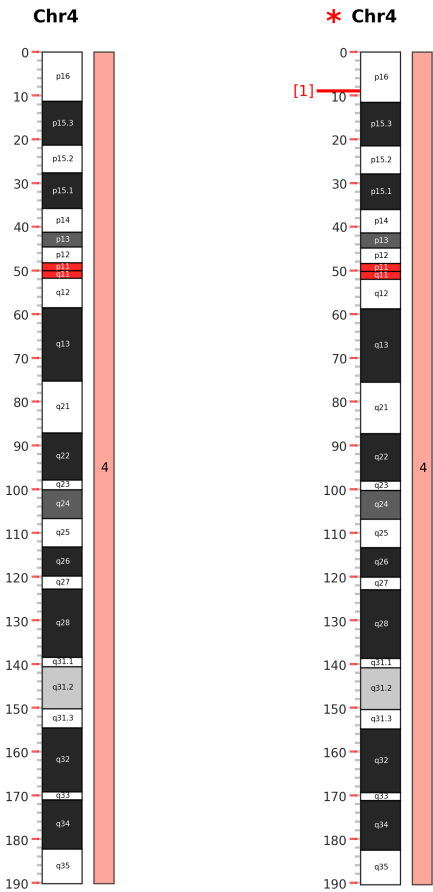
Impacted genes in DDG2P
None

Figure 7: karyotype 12, cluster 1

Event Cluster 2 (of 4)

SVs

1. inv(7)(q11.23). inversion on
Chr7: 73,061,114-75,291,965 (q11.23 - q11.23)
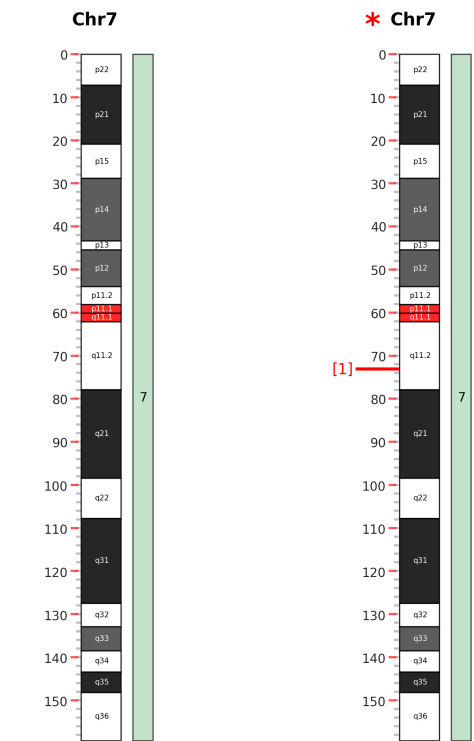
Impacted genes in DDG2P

None

Figure 8: karyotype 12, cluster 2

Event Cluster 3 (of 4)

SVs

1. dup(17)(p11.2). tandem duplication on
Chr17: 21,366,522-21,692,279 (p11.2 - p11.2)
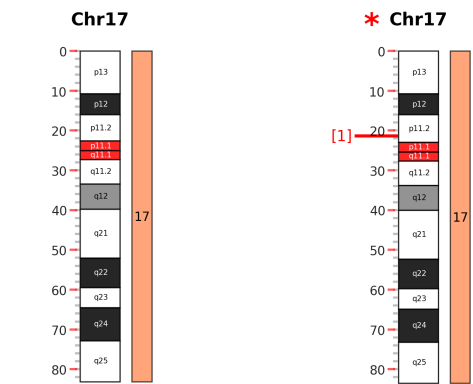
Impacted genes in DDG2P

None

Figure 9: karyotype 12, cluster 3

Event Cluster 4 (of 4)

SVs

1. del(22)(q11.23q12.1). deletion on
Chr22: 25,127,033-25,515,764 (q11.23 - q12.1)

Impacted genes in DDG2P

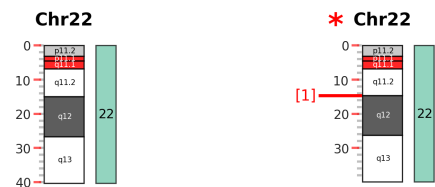| SV	Rationale	Gene Name	Gene Omim
1	CN-1	CRYBB2	123620
1	CN-1	CRYBB3	123630

Figure 10: karyotype 12, cluster 4

Sample Id: 39

Event Cluster 1 (of 3)

SVs

1. t(1;17)(q43;q25.3). balanced translocation between Chr1 and Chr17,
between segments Chr1: 237,668,707-248,943,333 (q43 - q44) and Chr17: 80,544,491-83,246,392 (q25.3 - q25.3)
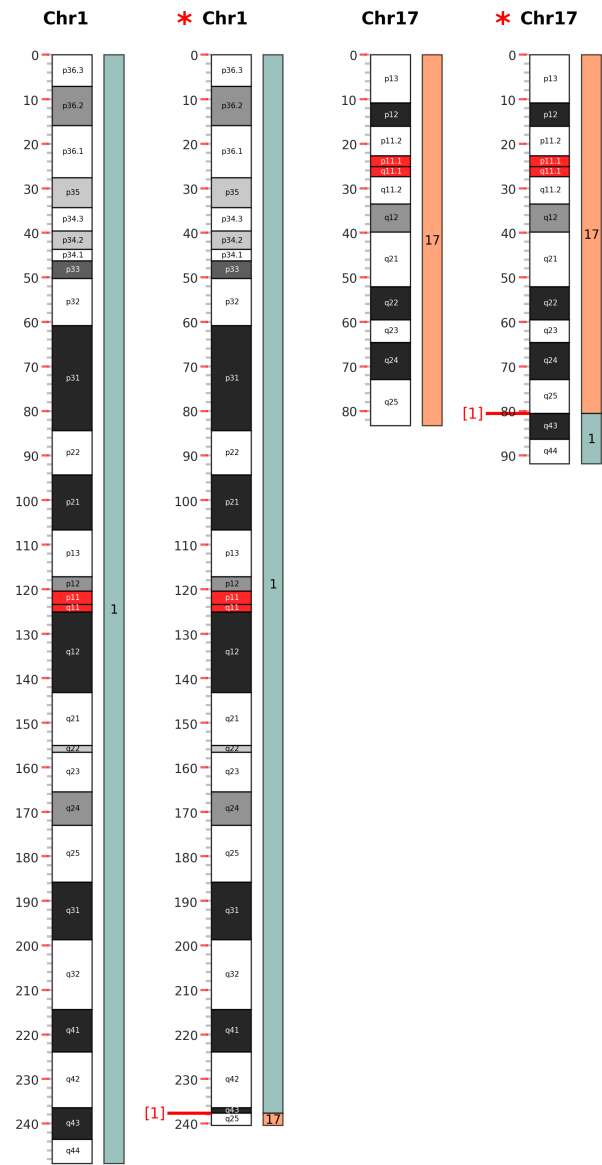
Impacted genes in DDG2P
None

Figure 11: karyotype 39, cluster 1

Event Cluster 2 (of 3)

SVs

1. inv(9)(p13.3p13.1). inversion on
Chr9: 33,582,925-38,561,385 (p13.3 - p13.1)
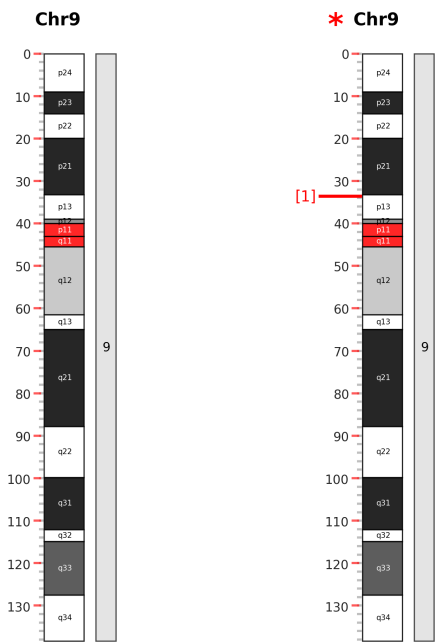
Impacted genes in DDG2P

None

Figure 12: karyotype 39, cluster 2

Event Cluster 3 (of 3)

SVs

1. dup(15)(q21.3). tandem duplication on
Chr15: 53,582,371-54,147,634 (q21.3 - q21.3)

Impacted genes in DDG2P

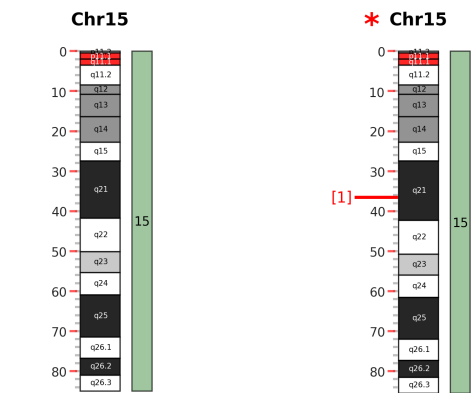| SV	Rationale	Gene Name	Gene Omim
1	breakpoint proximal	HERC2	605837

Figure 13: karyotype 39, cluster 3

Sample Id: 45

Event Cluster 1 (of 1)

SVs

1. t(1;15)(q41;q25.1). balanced translocation between Chr1 and Chr15, between segments Chr1: 220,322,626-248,943,333 (q41 - q44) and Chr15: 78,010,675-101,976,509 (q25.1 - q26.3)
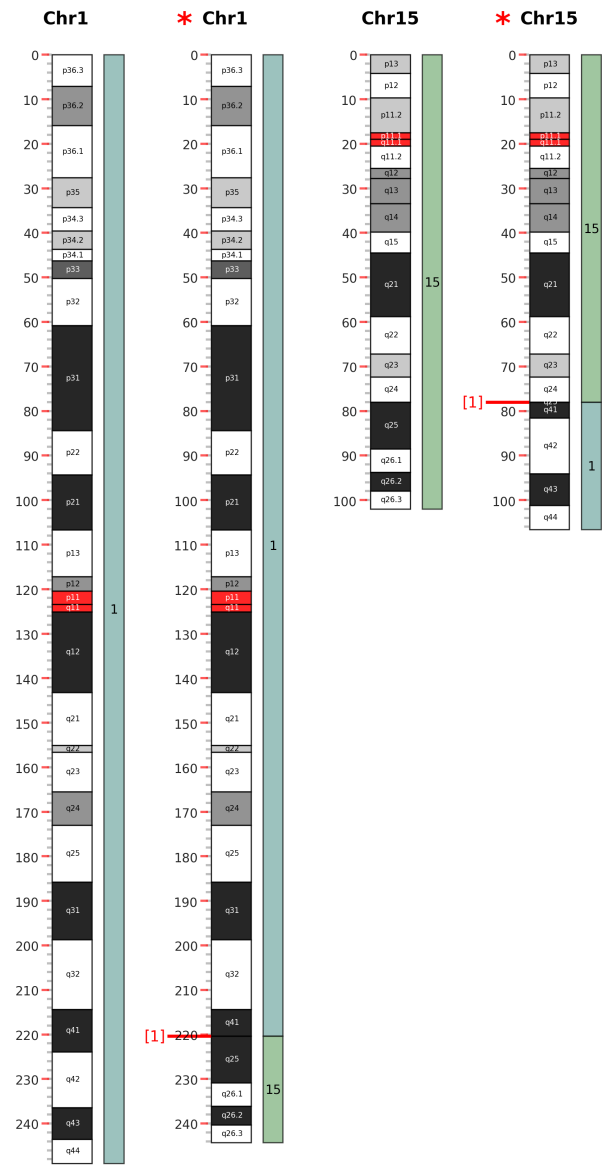
Impacted genes in DDG2P

None

Figure 14: karyotype 45, cluster 1