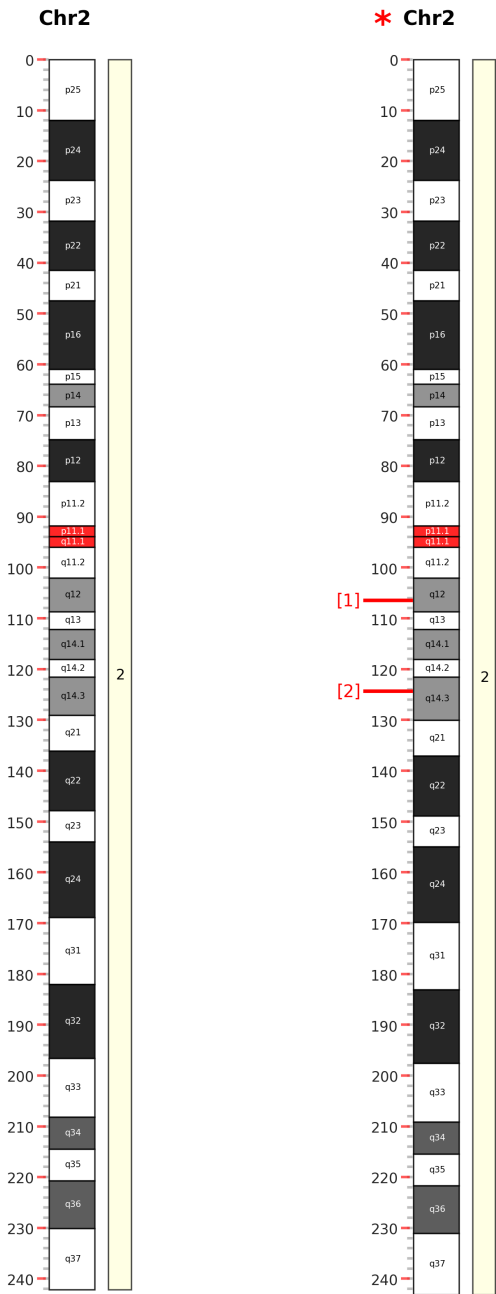


Parameters

- 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



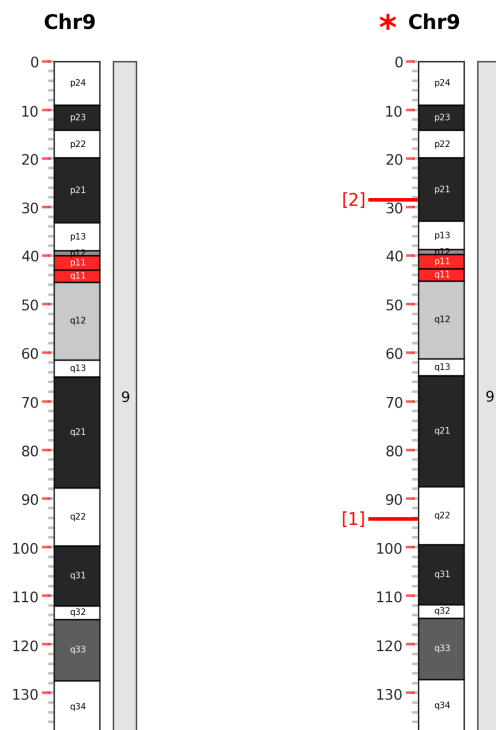
SVs

- 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

Event Cluster 2



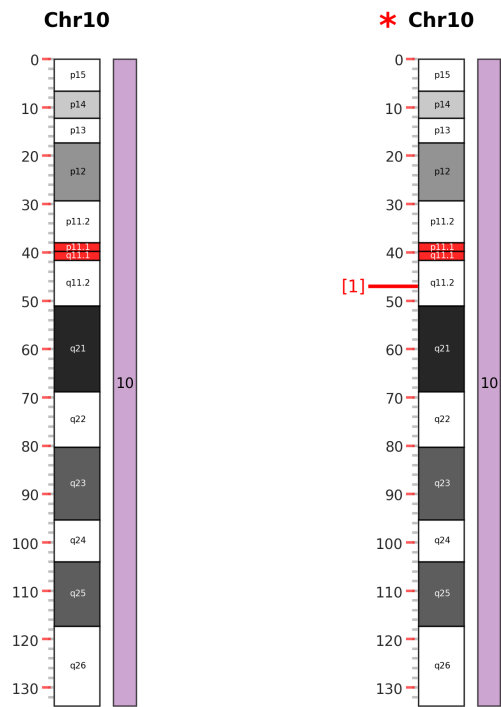
SVs

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

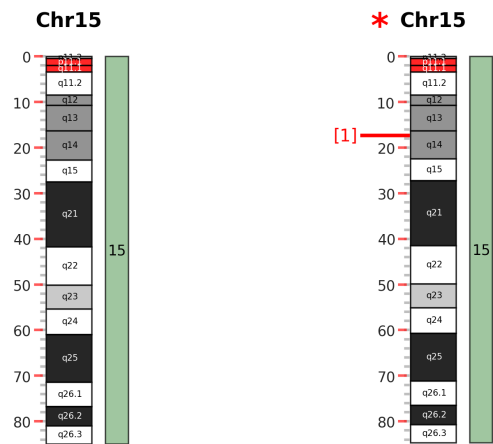


SVs
1. **inv(10)(q11.22)**.inversion on
Chr10: 46,991,593-
48,060,267 (q11.22 - q11.22)

Impacted genes in DDG2P

None

Event Cluster 4



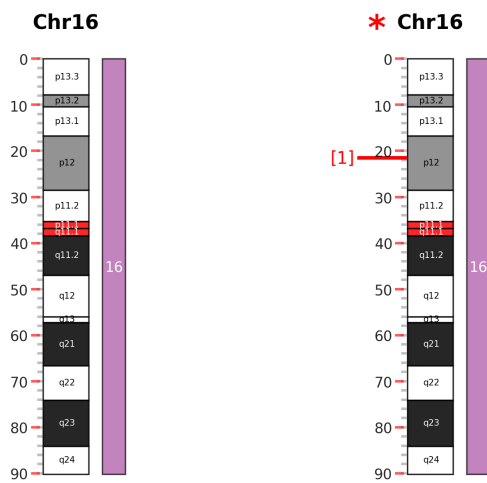
SVs

- 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

Event Cluster 5



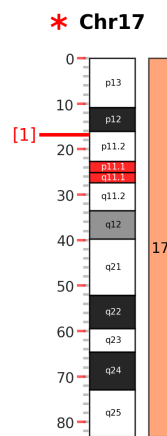
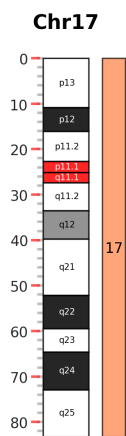
SVs

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

Impacted genes in DDG2P

None

Event Cluster 6



SVs

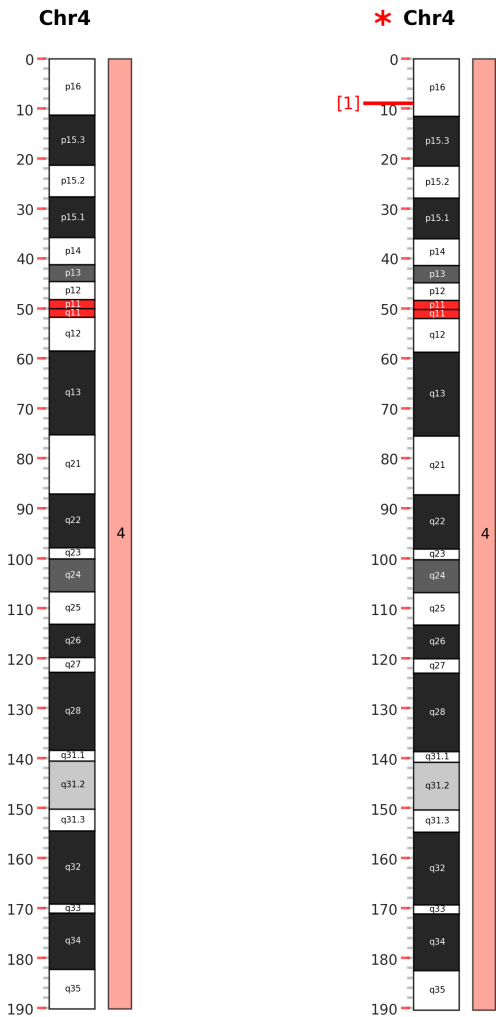
1. **inv(17)(p11.2)**. inversion on
Chr17: 16,806,184-
18,509,508 (p11.2-p11.2)

Impacted genes in DDG2P

None

Sample Id: 12

Event Cluster 1

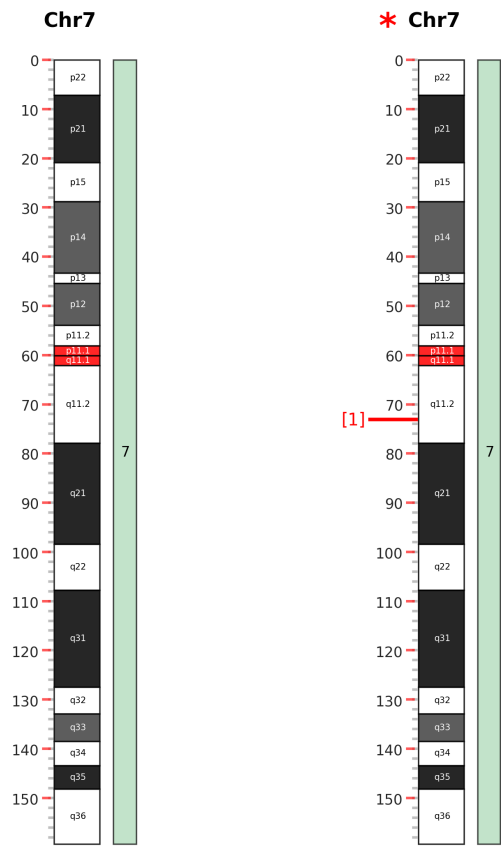


SVs
1. **dup(4)(p16.1)**. tandem duplication on
Chr4: 8,929,610-9,144,574 (p16.1 -
p16.1)

Impacted genes in DDG2P

None

Event Cluster 2

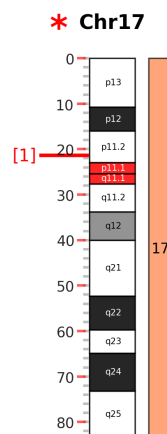
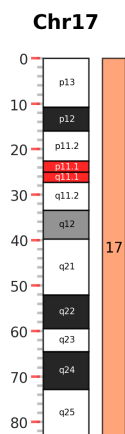


SVs
1. **inv(7)(q11.23)**.inversion on
Chr7: 73,061,114-
75,291,965 (q11.23 - q11.23)

Impacted genes in DDG2P

None

Event Cluster 3



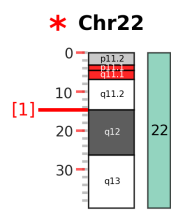
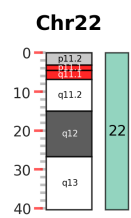
SVs

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

Impacted genes in DDG2P

None

Event Cluster 4



SVs

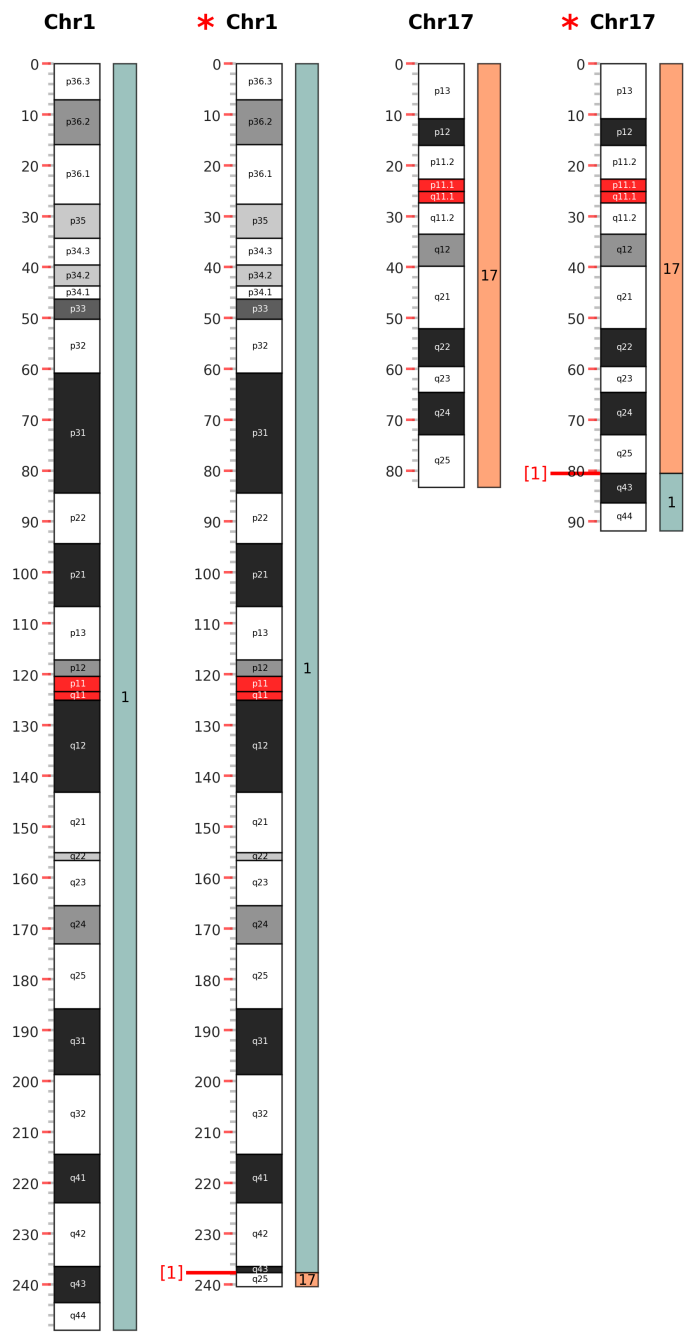
- 1. **del(22)(q11.23q12.1)**.deletion on Chr22: 25,127,033-25,515,764 (q11.23 - q12.1)

Impacted genes in DDG2P

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

Sample Id: 39

Event Cluster 1



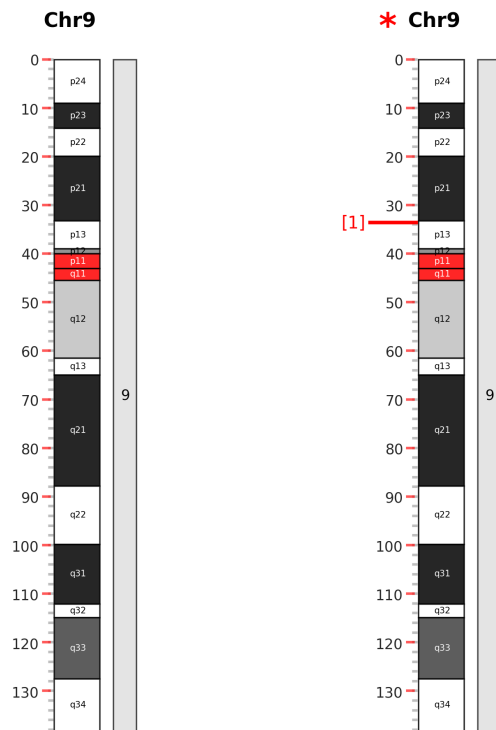
SVs

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

Event Cluster 2



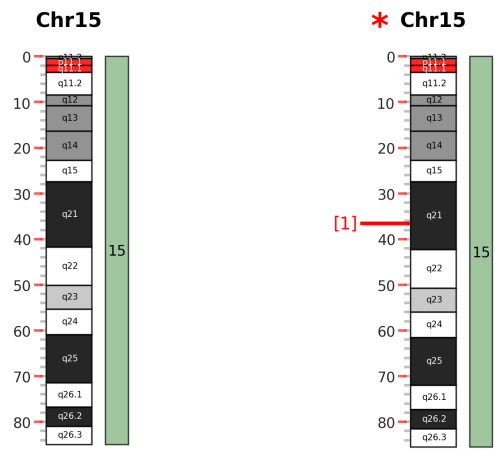
SVs

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

Impacted genes in DDG2P

None

Event Cluster 3



SVs

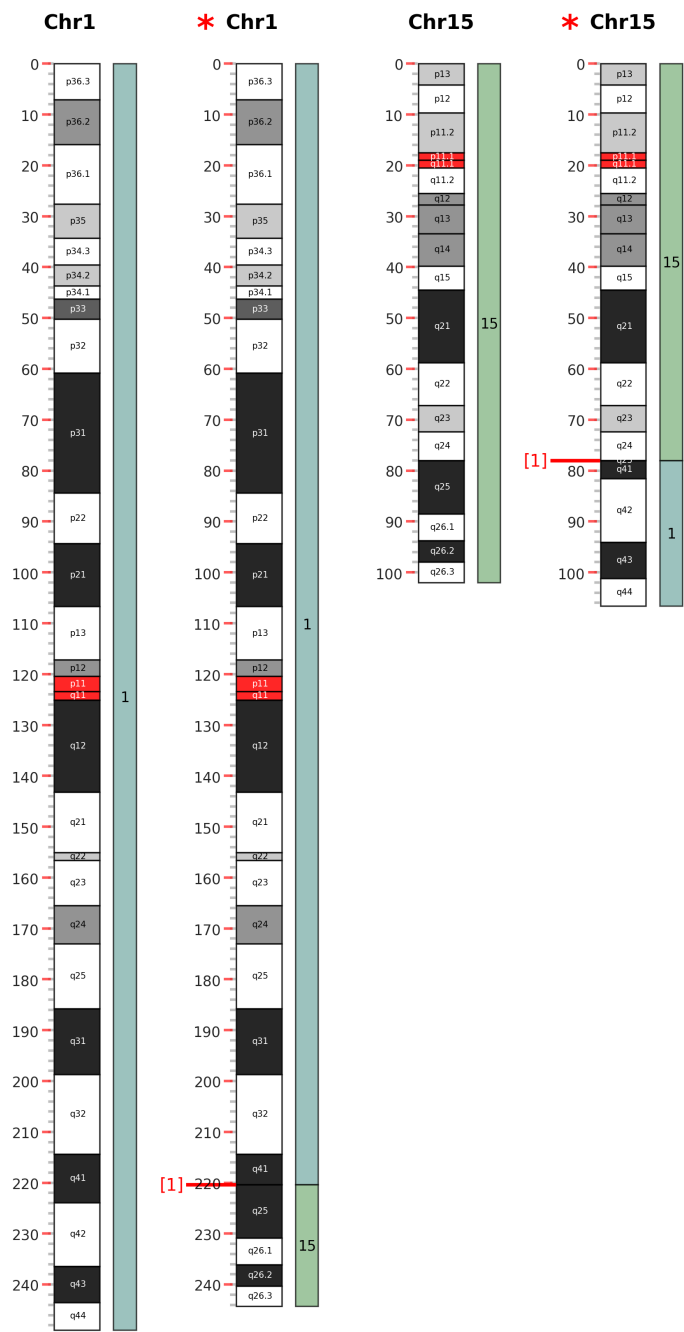
- 1. **dup(15)(q21.3)**. tandem duplication on Chr15: 53,582,371-54,147,634 (q21.3 - q21.3)

Impacted genes in DDG2P

SV	Rationale	Gene Name	Gene Omim
1	breakpoint proximal	HERC2	605837

Sample Id: 45

Event Cluster 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