5.4.6 Event modifiers

As mentioned previously, a single rearrangement event can be described as a set of novel adjacencies. For example, a reciprocal rearrangement such as in Figure 7:

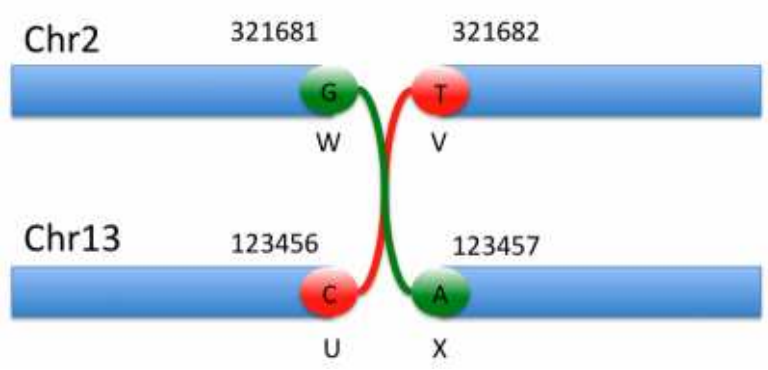

Figure 7: Rearrangements

would be described as:

#CHROM	POS	ID	REF	ALT	QUAL	FILTER	INFO
2	321681	bnd_W	G	G[13 : 123457[	6	PASS	SVTYPE=BND;MATEID=bnd_X;EVENT=RR0
2	321682	bnd_V	T	]13 : 123456]T	6	PASS	SVTYPE=BND;MATEID=bnd_U;EVENT=RR0
13	123456	bnd_U	C	C[2 : 321682[	6	PASS	SVTYPE=BND;MATEID=bnd_V;EVENT=RR0
13	123457	bnd_X	A	]2 : 321681]A	6	PASS	SVTYPE=BND;MATEID=bnd_W;EVENT=RR0

5.4.7 Inversions

Similarly an inversion such as in Figure 8:

Figure 8: Inversion

can be described equivalently in two ways. Either one uses the short hand notation described previously (recommended for simple cases):

#CHROM	POS	ID	REF	ALT	QUAL	FILTER	INFO
2	~~321682~~ 321681	INV0	~~T~~ G	<INV>	6	PASS	SVTYPE=INV;END=421681

or one describes the breakends:

#CHROM	POS	ID	REF	ALT	QUAL	FILTER	INFO
2	321681	bnd_W	G	G]2 : 421681]	6	PASS	SVTYPE=BND;MATEID=bnd_U;EVENT=INV0
2	321682	bnd_V	T	[2 : 421682[T	6	PASS	SVTYPE=BND;MATEID=bnd_X;EVENT=INV0
2	421681	bnd_U	A	A]2 : 321681]	6	PASS	SVTYPE=BND;MATEID=bnd_W;EVENT=INV0
2	421682	bnd_X	C	[2 : 321682[C	6	PASS	SVTYPE=BND;MATEID=bnd_V;EVENT=INV0

5.4.8 Uncertainty around breakend location

It sometimes is difficult to determine the exact position of a break, generally because of homologies between the sequences being modified, such as in Figure 9. The breakend is then placed arbitrarily at the left most position, and the uncertainty is represented with the CIPOS tag. The ALT string is then constructed assuming this arbitrary breakend choice.

The figure above represents a nonreciprocal translocation with microhomology. Even if we know that breakend U is rearranged with breakend V, actually placing these breaks can be extremely difficult. The red and green dashed