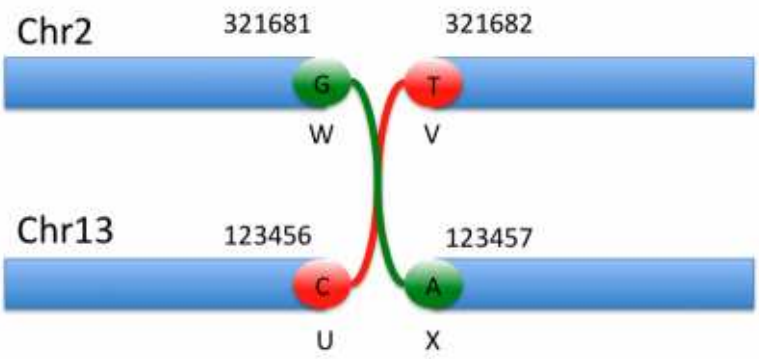

Figure 7: Rearrangements

Figure 8: Inversion

5.4.7 Inversions

Similarly an inversion such as in Figure 8:
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 lines represent the most extreme possible recombination events which are allowed by the sequence evidence available. We therefore place both U and V arbitrarily within the interval of possibility:

#CHROM	POS	ID	REF	ALT	QUAL	FILTER	INFO
2	321681	bnd_V	T	T]13 : 123462]	6	PASS	SVTYPE=BND;MATEID=bnd_U;CIPOS=0,6
13	123456	bnd_U	A	A]2 : 321687]	6	PASS	SVTYPE=BND;MATEID=bnd_V;CIPOS=0,6

Note that the coordinate in breakend U's ALT string does not correspond to the designated position of breakend V, but to the position that V would take if U's position were fixed (and vice-versa). The CIPOS tags describe the uncertainty around the positions of U and V.

The fact that breakends U and V are mates is preserved thanks to the MATEID tags. If this were a reciprocal translocation, then there would be additional breakends X and Y, say with X the partner of V on Chr 2 and Y