

# STRUCTURAL VARIATION

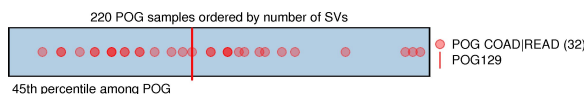
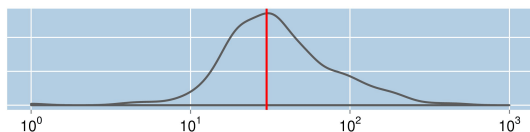
This section provides details of structural variations detected from de novo assembly of the tumour genome and transcriptome. Look in this section for details of the potentially relevant large-scale genomic rearrangements including gene fusions, duplications and deletions.

## STRUCTURAL VARIATION

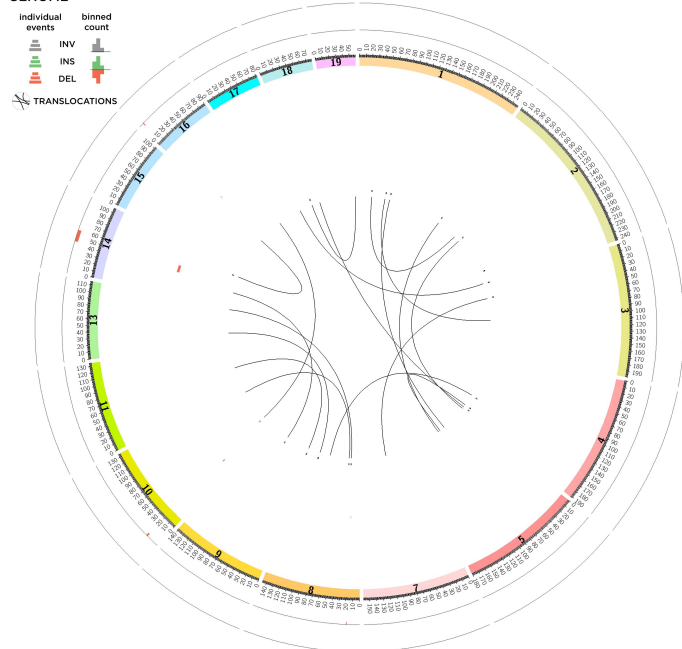
### SUMMARY OF STRUCTURAL EVENTS

Structural Variants  
[Expressed Fusions]

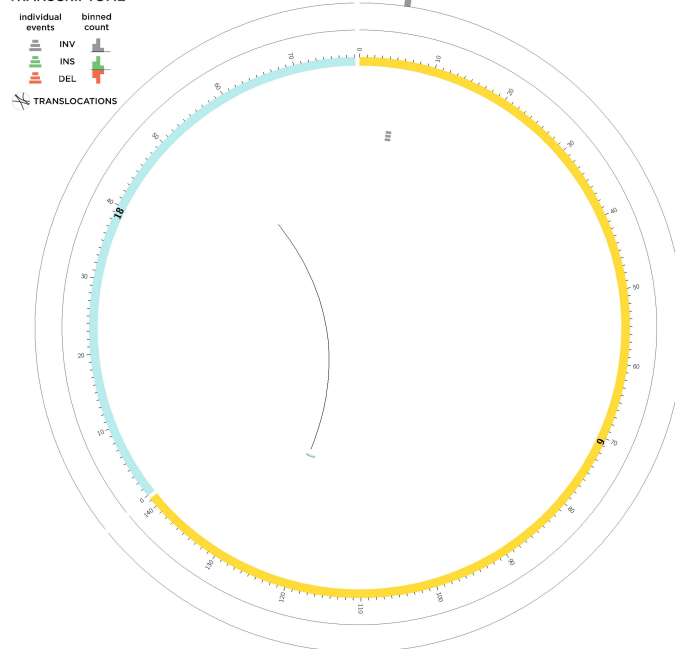
30 [5]



#### POG129 GENOME



#### POG129 TRANSCRIPTOME



### STRUCTURAL VARIATION: GENOMIC DETAILS

#### GENE FUSIONS OF POTENTIAL CLINICAL RELEVANCE WITH GENOME AND TRANSCRIPTOME SUPPORT

No gene fusions of potential clinical relevance were found from the analysis.

#### GENE FUSIONS OF PROGNOSTIC AND DIAGNOSTIC RELEVANCE

No gene fusions of prognostic or diagnostic relevance were found from the analysis.

#### GENE FUSIONS WITH BIOLOGICAL RELEVANCE

Genes 5'::3'	Exons 5'/3'	Breakpoint	Event Type	Sample	Cytogenetic Description	RPKM 5'/3'	Fold Change vs. unknown	TCGA %ile
EWSR1::FLI1	e7:e4	22:29683123   11:128675261	translocation	RNA	t(22;11)(q12.2;q24.3)	10.96/6.37	-1.4/-1.29	0/56

#### GENE FUSIONS WITH GENOME AND TRANSCRIPTOME SUPPORT

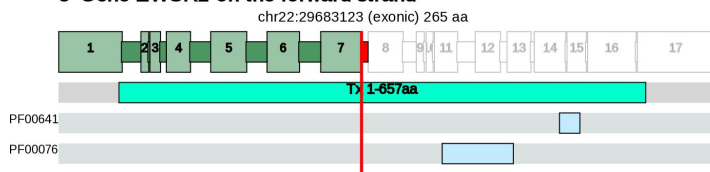
## STRUCTURAL VARIATION

Genes 5'::3'	Exons 5'/3'	Breakpoint	Event Type	Sample	Cytogenetic Description	RPKM 5'/3'	Fold Change vs. unknown 5'/3'	TCGA %ile 5'/3'
TRIM28::NA	e2:e	19:59056904   19:59096255	inversion	RNA	i(19)(q13.43)	93.05/na	2.09/na	56/na
WIPF3::IQCF4	e:e	7:29929023   3:51851986	translocation	RNA	t(7;3)(p14.3;p21.2)	15.39/na	2.37/na	100/na

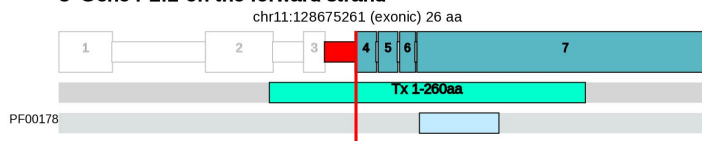
**STRUCTURAL VARIATION**

**GENE FUSION FIGURES**

**5' Gene EWSR1 on the forward strand**



**3' Gene FLI1 on the forward strand**



**EWSR1/FLI1 Fusion Gene**

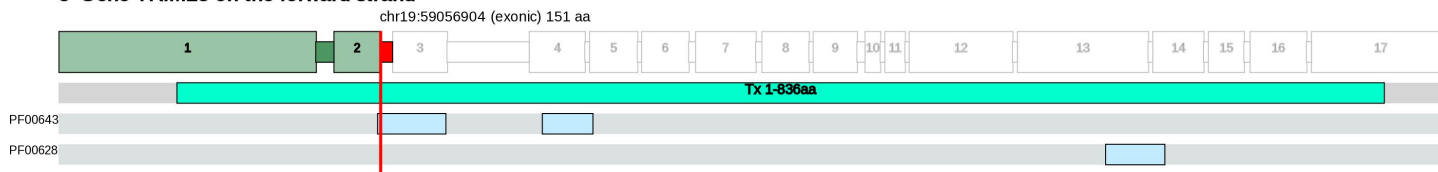


Gene ID: **EWSR1::FLI1** Type: **translocation** Predicted: **in-frame**

Position	HGNC Gene	Ensembl Gene	Ensembl Transcript	Exon	Strand	Breakpoint
5'	EWSR1	ENSG00000182944	ENST00000397938	e7	forward	22:29683123
3'	FLI1	ENSG00000151702	ENST00000525560	e4	reverse	11:128675261

Putative Fusion-translocation event. The 5' (in the fusion) transcript EWSR1-001(ENST00000397938) from the gene EWSR1(ENSG00000182944) on the forward strand is drawn top left with its corresponding breakpoint at 22:29683123. The 3' (in the fusion) transcript FLI1-004(ENST00000525560) from the gene FLI1(ENSG00000151702) on the forward strand is drawn top right with its corresponding breakpoint at 11:128675261. Exons are drawn to scale relative to other exons in the same drawing. Introns are scaled to make up approximately 1/4th of the final drawing. This is predicted to be an IN-frame fusion. Domain(s) featured in the above figure(s) are labelled by their various external identifiers as follows: Ets(PF00178); RRM\_dom(PF00076); Znf\_RanBP2(PF00641).

**5' Gene TRIM28 on the forward strand**



Gene ID: **TRIM28::NA** Type: **inversion** Predicted: **not determined**

Position	HGNC Gene	Ensembl Gene	Ensembl Transcript	Exon	Strand	Breakpoint
5'	TRIM28	ENSG00000130726	ENST00000253024	e2	forward	19:59056904
3'	NA	<na>	<na>	e	reverse	19:59096255

Transcript TRIM28-201(ENST00000253024) from gene TRIM28(ENSG00000130726) on the forward strand. Breakpoint shown is 19:59056904. Exons are drawn to scale relative to other exons in the same drawing. Introns are scaled to make up approximately 1/4th of the final drawing. Domain(s) featured in the above figure(s) are labelled by their various external identifiers as follows: Znf\_B-box(PF00643); Znf\_PHD-finger(PF00628).