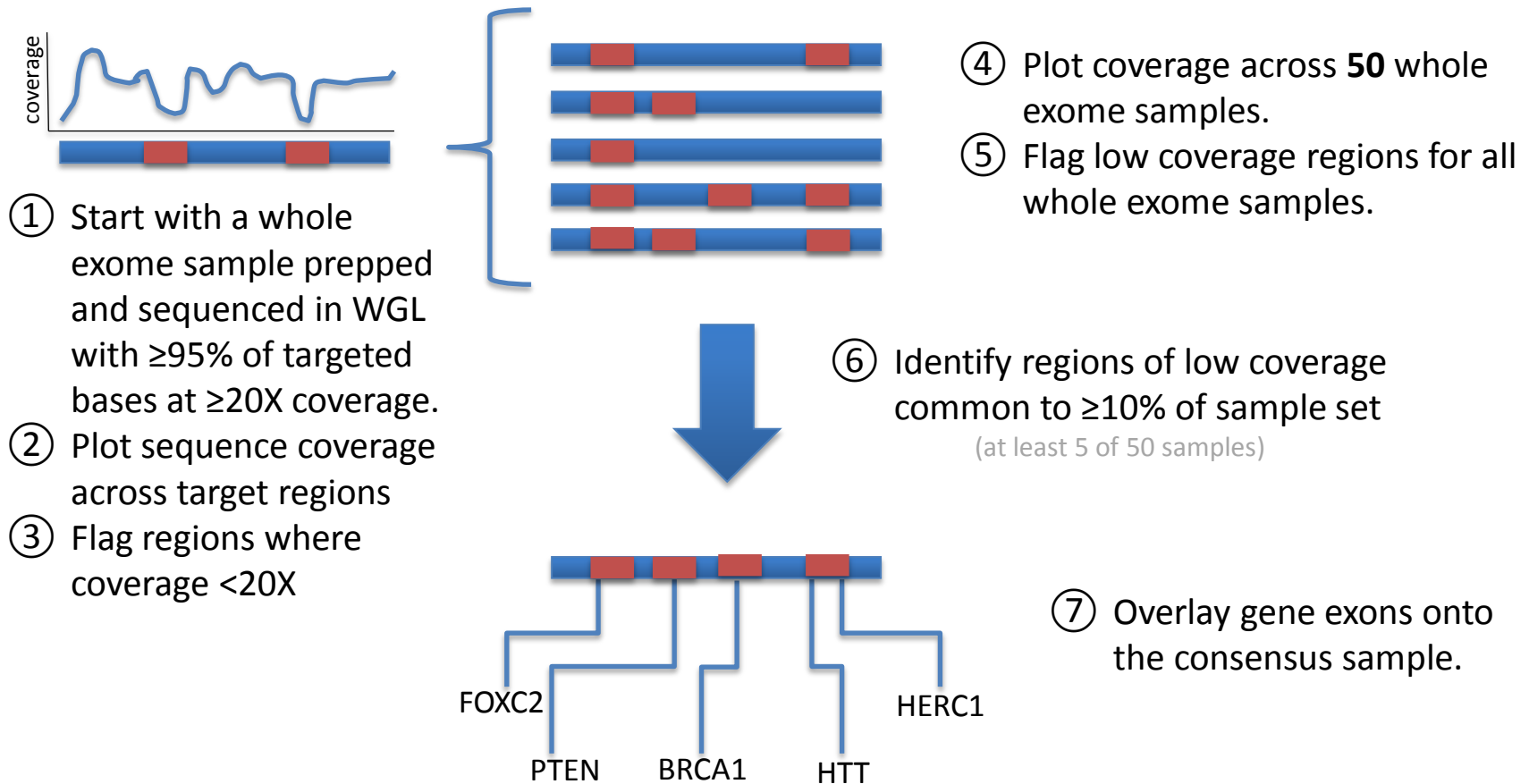




Whole Exome Coverage Search

Instructions and Coverage Calculations

Low coverage audit of WES version 3 targets



Current coverage search website

Whole Exome Coverage Search

[Instructions and Coverage Calculations](#)

Select an area to search: Gene Transcript Enter keyword: Search

Enter gene or transcript here.

Please note: This representation of sequence coverage of specific coding exons was generated from a dataset of samples undergoing exome sequencing, the actual coverage for individual samples may vary.

Click for NCBI gene description

Click for an exon-by-exon view of the transcript.

2 matches identified for your search.

Click on the transcript number to view coverage details. If you do not see a gene listed, please try a new search.

Gene	Chromosome	Transcript	Size	Exons	WGL Exome Coverage >20X (%)	HGMD
PTPN11	chr12	NM_080601	1394	11	98.924	●
PTPN11	chr12	NM_002834	1797	16	99.165	●

HGMD isoform

● = Isoform currently used / ● = Isoform not currently used in the Human Gene Mutation Database (HGMD).

Current coverage search website

Whole Exome Coverage Search

Select an area to search: Gene Transcript Enter keyword: Search

Please note: This representation of sequence coverage of specific coding exons was generated from a dataset of samples undergoing exome sequencing, the actual coverage for individual samples may vary.

Gene Name: *PTPN11*

Link to NCBI transcript description.

CHR	GENE	TRANSCRIPT	SIZE	EXONS	WGL Exome Coverage >20X (%)
chr12	PTPN11	NM_002834	1797	16	99.165

EXON NUMBER	EXON START	EXON END	WGL Exome Coverage >20X (%)	COORDS < 20X	Total Bases < 20X
cds_0	112856915	112856929	0	112856915-112856929	14
cds_1	112884079	112884202	100		
cds_2	112888121	112888316	100		
cds_3	112890998	112891191	100		

NCBI transcript description

PTPN11 protein tyrosine phosphatase, non-receptor type 11 [*Homo sapiens* (human)]

Gene ID: 5781, updated on 29-Jun-2014

Summary

Official Symbol PTPN11 provided by [HGNC](#)

Official Full Name protein tyrosine phosphatase, non-receptor type 11 provided by [HGNC](#)

Primary source [HGNC:9644](#)

See related [Ensembl:ENSG00000179295](#); [HPRD:01470](#); [MIM:176876](#); [Vega:OTTHUMG00000134334](#)

Gene type protein coding

RefSeq status REVIEWED

Organism [Homo sapiens](#)

Lineage Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Homo

Also known as CFC; NS1; SHP2; BPTP3; PTP2C; PTP-1D; SH-PTP2; SH-PTP3

Summary The protein encoded by this gene is a member of the protein tyrosine phosphatase (PTP) family. PTPs are known to be signaling molecules that regulate a variety of cellular processes including cell growth, differentiation, mitotic cycle, and oncogenic transformation. This PTP contains two tandem Src homology-2 domains, which function as phospho-tyrosine binding domains and mediate the interaction of this PTP with its substrates. This PTP is widely expressed in most tissues and plays a regulatory role in various cell signaling events that are important for a diversity of cell functions, such as mitogenic activation, metabolic control, transcription regulation, and cell migration. Mutations in this gene are a cause of Noonan syndrome as well as acute myeloid leukemia. Two transcript variants encoding different isoforms have been found for this gene. [provided by RefSeq, May 2012]

Current coverage search website

Whole Exome Coverage Search

Select an area to search: Gene Transcript Enter keyword: Search

Please note: This representation of sequence coverage of specific coding exons was generated from a dataset of samples undergoing exome sequencing, the actual coverage for individual samples may vary.

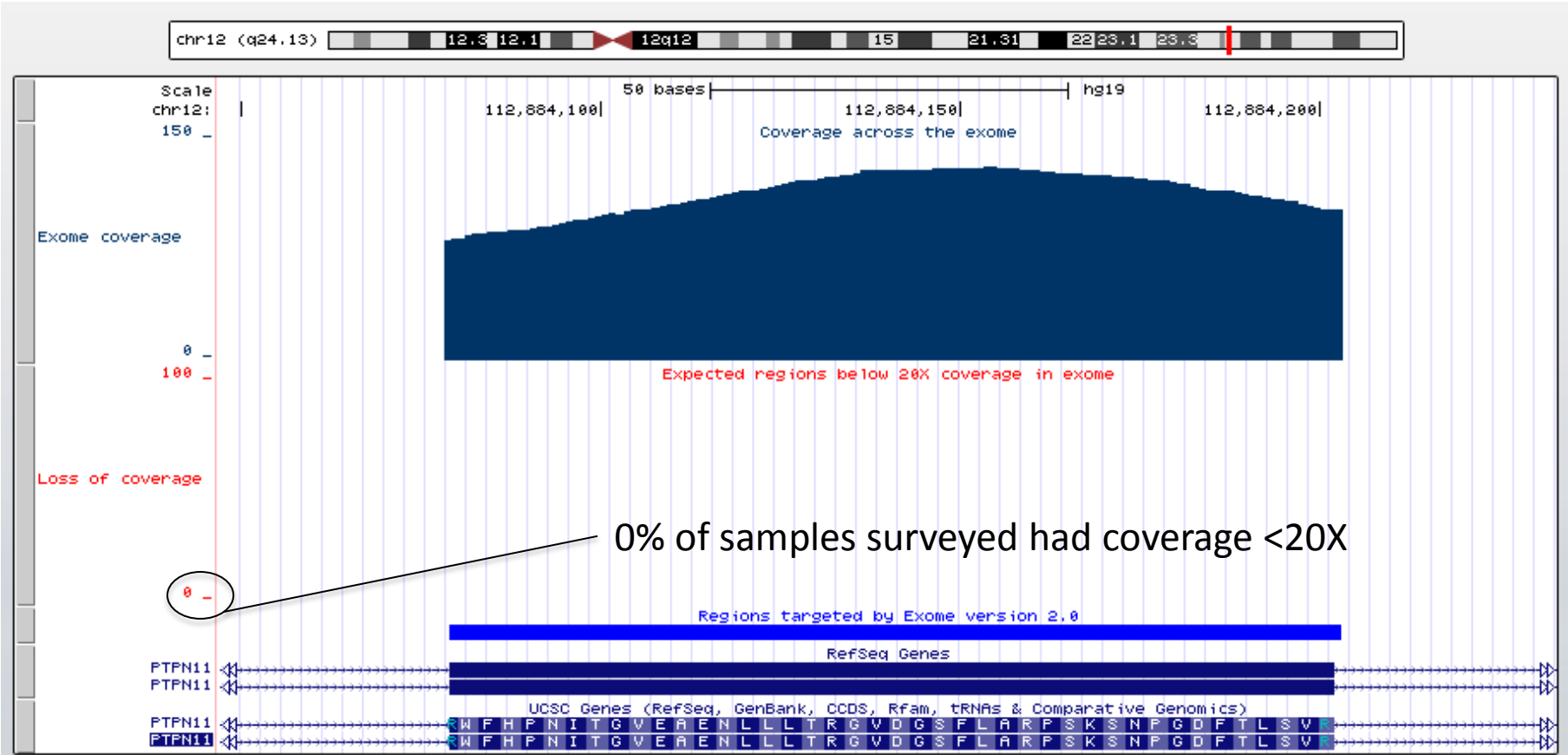
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cds_1	112884079	112884202	<u>100</u>		
cds_2	112888121	112888316	<u>100</u>		
cds_3	112890998	112891191	<u>100</u>		

Percent sequence coverage $\geq 20X$ in exon. Link to exon view in UCSC browser.

UCSC browser view of exon >20X



Current coverage search website

Whole Exome Coverage Search

Select an area to search: Gene Transcript Enter keyword: Search

Please note: This representation of sequence coverage of specific coding exons was generated from a dataset of samples undergoing exome sequencing, the actual coverage for individual samples may vary.

Gene Name: *PTPN11*

CHR	GENE	TRANSCRIPT	SIZE	EXONS	WGL Exome Coverage >20X (%)
chr12	PTPN11	NM_002834	1797	16	99.165

EXON NUMBER	EXON START	EXON END	WGL Exome Coverage >20X (%)	COORDS < 20X	Total Bases < 20X
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cds_1	112884079	112884202	100		
cds_2	112888121	112888316	100		
cds_3	112890998	112891191	100		

Info about regions below 20X sequence coverage. Link to exon view in UCSC browser.

UCSC browser view of exon <20X

