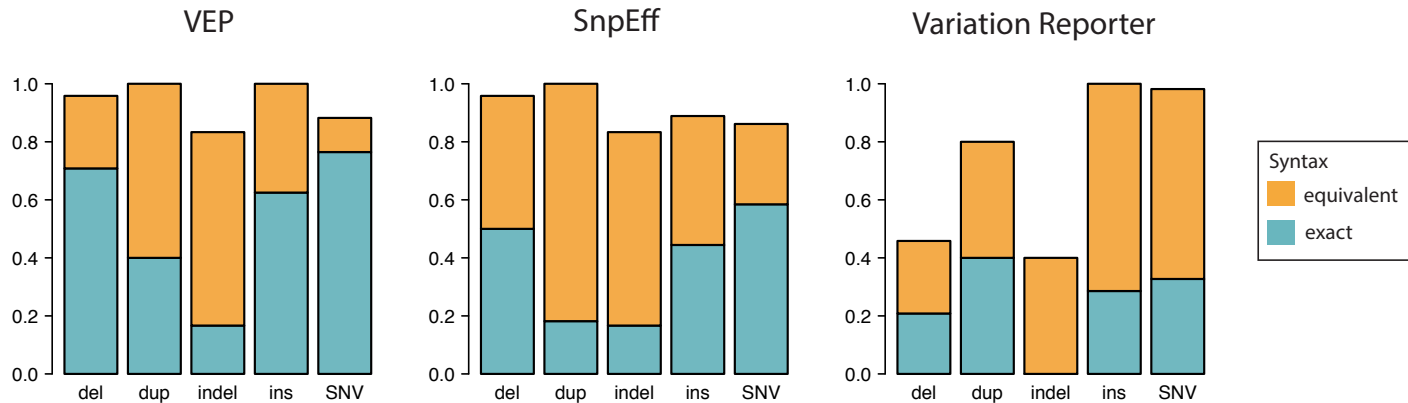


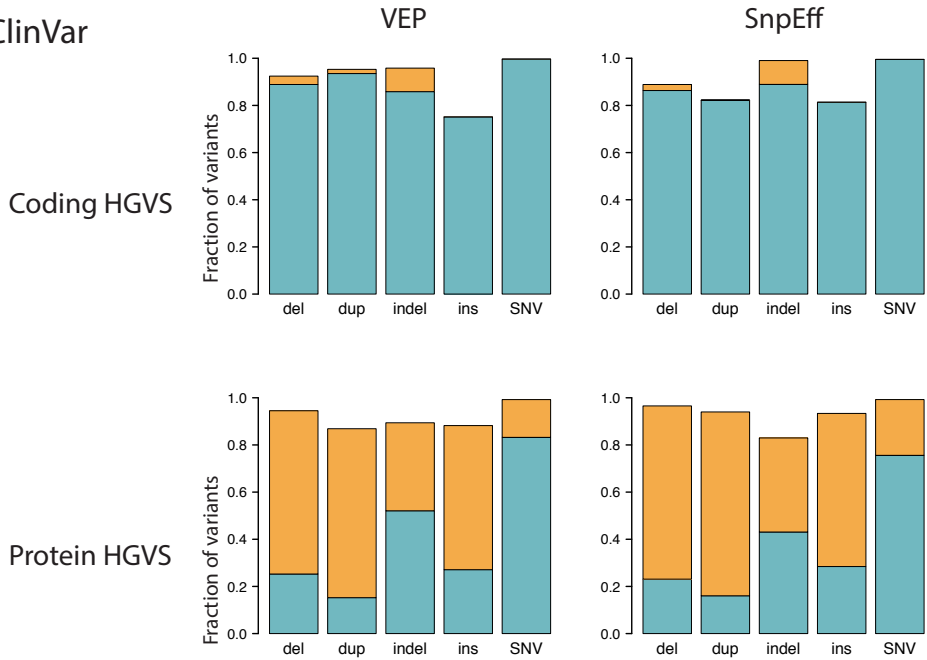
S1

## Effect Impact

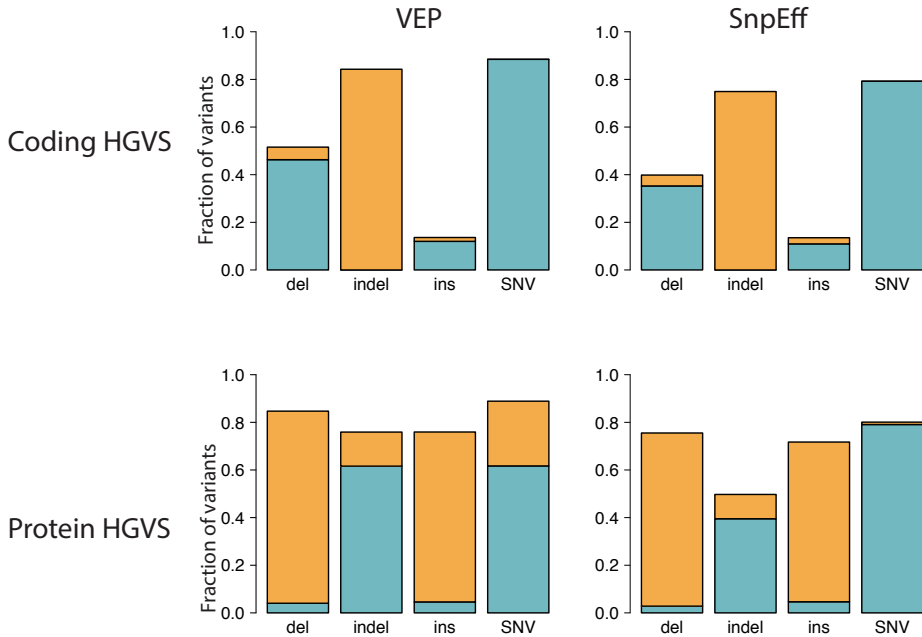


## S2.

### a. ClinVar



### b. COSMIC



## a. Possible transcripts and nomenclature

Transcripts	Coding HGVS	Protein HGVS
NM_001126128.1	c.297dupT	p.Gly100Trpfs*22
NM_021935.3	c.234dupT	p.Gly79Trpfs*22

## b. HGVS syntax search matrix

Search	Result - PubMed	Result - Google
PROK2 297dup	(nothing)	LOVD entry from ESP
PROK2 Gly100Trpfs OR Gly100fs OR G100Wfs OR G100fs	(nothing)	LOVD entry from ESP; Dissertation in Portuguese
PROK2 234dup	(nothing)	(nothing)
PROK2 Gly79Trpfs OR Gly79fs OR G79Wfs OR G79fs	(nothing)	Link to company primer assay webpage
<b>PROK2 G100fsX121</b>	<b>Abreu et al., 2008</b>	<b>Abreu et al., 2008; other review papers</b>

## c. Spider-webbing to evidence and classification

