**Impact of genetic variation on three dimensional structure and function of proteins**

improve understanding of the relationship between point mutations and experimentally observed consequences in 3D

detailed overview about the observed effects of SNVs on the structure, function, stability, and binding properties of proteins

results document that the range of possible SNV effects at the protein level are significantly greater than currently assumed by existing software prediction methods, and that correct prediction of consequences remains a significant challenge. Qualitative, not quantitative assessment.

**Protein of Interest:**

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HSOD (Gene: SOD1) Ala→Val (A4V) (rs121912442) leading to Aggregation (familial ALS)

PDB: 1N19

Searching for HSOD on PDB, we find 13 entries overall.

12 of those had their structure characterized using X-Ray Diffraction, one with Solution NMR.

The 1N19 (HSOD A4V mt) structure was determined using X-Ray Diffraction.

The UniProt ID of HSOD is P00441

1N19 has one unique protein chain and as it’s a homo dimer, it has two chains. Its sequence counts 308 residues. It has 3 unique Ligands: Zinc Ion, Sulfate Ion, Copper (1) Ion.

