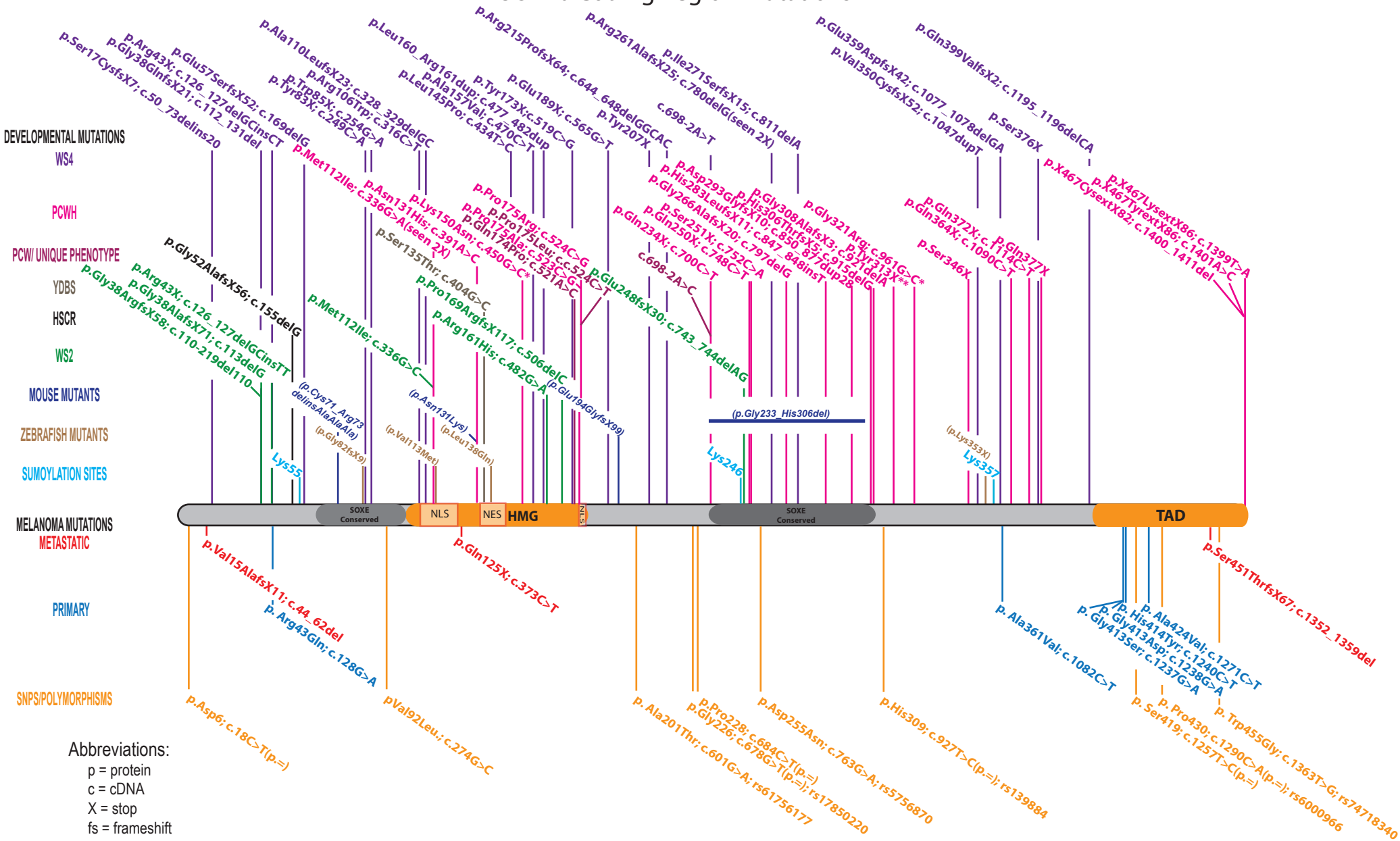
SOX10 Coding Region Mutations



Abbreviations:

- p = protein
- c = cDNA
- X = stop
- fs = frameshift
- dup = duplication
- ins = insertion
- ext = extension
- WS4 = Waardenburg syndrome 4
- PCWH = peripheral demyelinating neuropathy, central dysmyelination, Waardenburg syndrome, and Hirschsprung disease
- HSCR = Hirschsprung disease
- YDBS = Yemenite deaf-blind hypopigmentation syndrome
- WS2 = Waardenburg syndrome 2

*Patient harbored two SOX10 mutations, p.Lys150Asn and p.Gly321Arg

**Mutation seen 4 independent times, with cDNA changes c.938dupA, c.939C>A (2X), c.939C>G

Adapted from Harris et al., 2010 *Pigment Cell Melanoma Res*

Pigment Cell Gene Resource, NHGRI, NIH

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http://research.nhgri.nih.gov/pigment_cell/