Mutations in _EZH2_ Cause Weaver Syndrome

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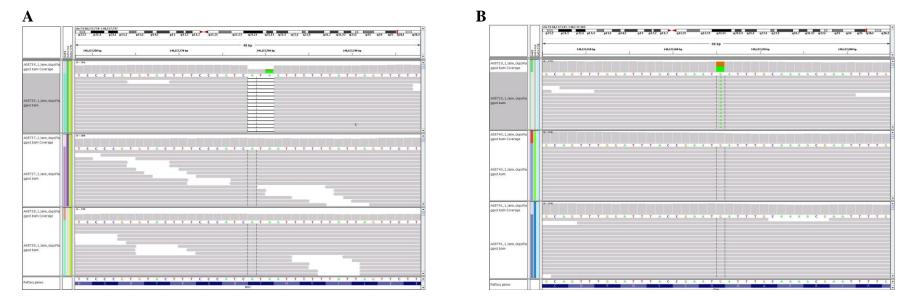
A

B

Figure S1. Integrative Genomics Viewer[1]-Generated Images of BAM File Read Alignments to hg18 for Probands 1 and 2 and Their Parents

(A) *EZH2* mutation c.457_459del (p.Tyr153del) in Proband 1 (top), his mother (middle) and father (bottom).

(B) *EZH2* mutation c.2080C>T (p.His694Tyr) in Proband 2 (top), her mother (middle) and father (bottom).

Table S1. Summary Statistics of Exome Resequencing

Subject	Proband 1 (A08736)	Mother 1 (A08737)	Father 1 (A08738)	Proband 2 (A08739)	Mother 2 (A08740)	Father 2 (A08741)
Total sequence yield (Gb) [a]	1.86	1.97	2.02	2.08	1.90	1.98
Total reads	429,667,422	424,496,474	463,366,406	506,490,420	463,258,182	409,296,116
Chastity-passed reads	348,012,586	386,608,086	414,972,084	441,242,928	415,420,736	372,566,906
Reads aligned to hg18	343,664,632	382,044,390	409,662,802	435,193,786	410,538,142	368,077,609
Reads aligned with mapping quality >=10	314,597,518	350,236,460	374,396,369	400,054,154	376,045,604	336,881,973
Reads used for variant calling [b]	143,593,996	141,579,235	187,165,231	202,315,301	176,426,879	182,790,043
Mean exome coverage [c]	213	236	253	274	260	228
Total exonic variants [d]	24,287	24,396	25,207	25,127	24,914	24,688
Exonic synonymous SNVs	11,355	11,420	11,777	11,782	11,704	11,676
Exonic non-synonymous (ns) variants [e]	12,932	12,976	13,430	13,345	13,210	13,012
nsSNVs	11,091	11,079	11,396	11,306	11,207	11,081
Splice-site SNVs [also nsSNVs]	889 [273]	905 [265]	964 [274]	985 [288]	935 [276]	912 [267]
Insertions/deletions [f]	585/640	583/674	636/708	637/705	618/726	602/684
ns variants not in dbSNP129/130	2,885	2,839	3,173	3,093	3,046	2,977
Novel ns variants [g]	2 (17)	117	196	2 (16)	148	113
Rare variants in compound heterozygous state	None	Not Analyzed	Not Analyzed	None	Not Analyzed	Not Analyzed
de novo candidate variants	c.457_459del (p.Tyr153del)	None	None	c.2080C>T (p.His694Tyr)	None	None

[a] – span of the human genome (hg18) covered by ≥ 1 read aligned with Phred-scaled mapping quality of ≥ 10

[b] – reads having Phred-scaled mapping quality of ≥ 10 and after duplicates are removed

[c] – average read depth of exons annotated in Ensembl 54; (sum of the number of reads aligned per site for all exonic sites) / (total number of exonic sites)

[d] – variants as output from [samtools.pl varFilter –D 1000 | awk '$6>=20'], excluding those in 5'UTR, 3'UTR, introns, and intergenic regions

[e] – variants include nsSNVs, splice-site SNVs within 2bp of exon boundaries, and small indels

[f] – coding region insertions/deletions supported by ≥ 6 aligned reads

[g] – not previously reported in dbSNP129/130, 1000 Genomes Project, or other non-cancer genomes collected in the Genome Sciences Centre local database. For probands, the total of "novel" variants excludes those seen in their unaffected parents and represents *de novo* variants. Numbers in parentheses include apparently *de novo* variants seen at low coverage (i.e. fewer than 100 reads per variant).

Supplementary References

1. Robinson, J.T., Thorvaldsdóttir, H., Winckler, W., Guttman, M., Lander, E.S., Getz, G., and Mesirov, J.P. (2011).
   Integrative Genomics Viewer. Nat Biotechnology *29*, 24–26. URL: http://www.broadinstitute.org/igv/