Supplementary Materials

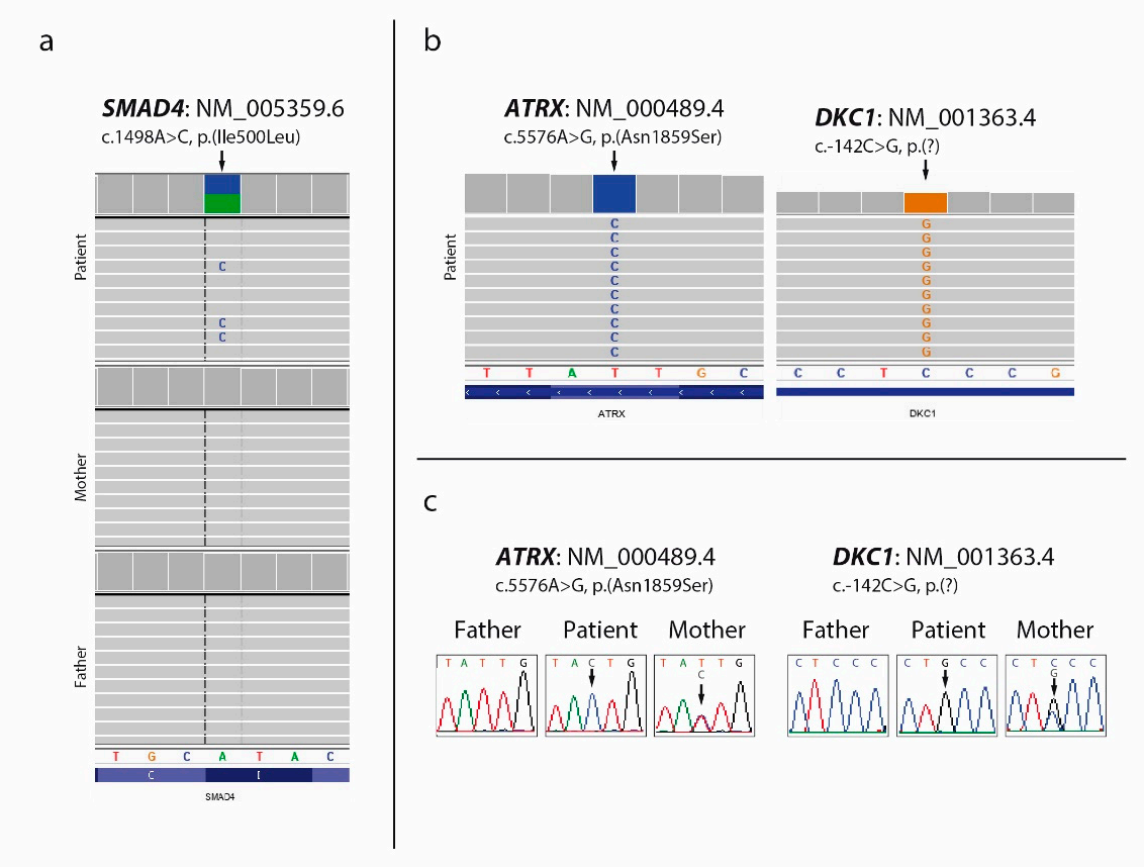


**Figure S1.** The Integrative Genomics Viewer (IGV) from Amplicon Deep Sequencing (**a**), IGV from Whole Exome Sequencing (**b**) an Sanger confirmation (c) of additional WES molecular findings. IGV of c.1498A>C, p.(Ile500Leu) variant found in *SMAD4* gene of case 2 (**a**). IGV and Sanger sequencing plots of p.5576A>G, p.(ASN1859Ser) variant found in *ATRX* gene of case 3, and c.-142C>G (p.?) variant found in *DKC1* gene of case 3 (**b**,**c**).

**Table S1.** Summary of the published mutations in the *WDR62* gene.

| No | Mutation (DNA level) | Mutation (amino acid level) | Exon/Intron | Homo-/heterozygoticty | Type mutation | Population | References |
|---|---|---|---|---|---|---|---|
| 1 | c.28G>T | p.Ala10Ser | 1 | ht | M | chinese | 7 |
| 2 | c.189G>T | p.Glu63Asp | 2 | ht | M | chinese | 7 |
| 3 | c.193G>A | p.Val65Met | 2 | h | M | saudi | 22,29 |
| 4 | c.194T>A | p.Val65Glu | 2 | ht | M | polish | case 2 |
| 5 | c.332G>C | p.Arg111Thr | 3 | h | M | pakistani | 13 |
| 6 | c.363delT | p.Asp112Metfs*5 | 4 | h | fs | mexican | 29 |
| 7 | c.390G>A | p.Glu130Glu | 4 | h | M | sudan | 8 |
| 8 | c.535_536insA | p.Met179fs*21 | 5 | h | fs | indian | 9 |
| 9 | c.617G>C | p.Trp224Ser | 6 | h | M | turkish | 10 |
| 10 | c.668T>C | p. Phe223Ser | 6 | h | M | romani | 27 |
| 11 | c.731C>T | p.Ser244Leu | 7 | ht | M | japanese | 17 |
| 12 | c.797C>T | p.Ala266Val | 7 | ht | M | saudi | 20 |
| 13 | c.900C>A | p.Cys300* | 8 | h | N | indian | 9 |
| 14 | c.883-1273_850del | del ex 8-9 | 8-9 | h | del | pakistani /korean | 25 |
| 15 | c.883-4_890del | del intron 7 +exon8 | 8 | ht | del | korean | 24 |
| 16 | c.1027C>T | p. Gln343* | 8 | h | N | morrocan | 30 |
| 17 | c.1043+1G>A | p.Ser348Argfs*63 | 8 | h | ss | turkish | 29 |
| 18 | c.1102G>A | p.Asp368Asn | 9 | ht | M | saudi | 20 |
| 19 | c.1143delA | p.His381Profs*48 | 9 | h | fs | pakistani | 16 |
| 20 | c.1194G>A | p.Trp398 | 9 | h | M | pakistani | 12,13 |
| 21 | c.1198G>A | p.Glu400Lys | 9 | h | M | spanish | 6 |
| 22 | c.1313G>A | p.Arg438His | 10 | h | M | pakistani, german | 11,12,13,14 |
| 23 | c.1408C>T | p.Gln470* | 11 | h | N | turkish | 10 |
| 24 | c.1531G>A | p. Asp511Asn | 11 | h | M | pakistani | 14,22 |
| 25 | C1535G>A | p.Arg512Gln | 11 | ht | M | chinese | 26 |
| 26 | c.1576G>A | p.Glu526Lys | 12 | h | M | turkish | 10 |
| 27 | c.1576G>T | p.Glu526* | 12 | h | N | turkish | 10 |
| 28 | c.1605_1606insT | p. Glu536* | 12 | h | N | turkish | 23 |
| 29 | c.1642+2T>g | p.? | 12 | ht | ss | polish | Case 1 |
| 30 | c.1684C>G | p.His562Asp | 13 | ht | fs | korean | 24 |
| 31 | c.1711_1712insTA | p.Asn571Ilefs*27 | 13 | ht | N | polish | Case 3 |
| 32 | c.1777_1778delGA | p.Asp593Hisfs*9 | 13 | ht | N | polish | Case 1 |
| 33 | c.1821dupT | p.Arg608Serfs*26 | 14 | h | fs | french canadian | 15 |
| 34 | c.1942C>T | p.Gln648* | 15 | h | M | pakistani | 14 |
| 35 | c.2083delA | p.Ser696Alafs*4 | 17 | ht | fs | ? | 18 |
| 36 | c.2195C>T | p.Thr732Ile | 20 | h | M | pakistani | 12 |
| 37 | c.2413G>T | p.Glu805* | 20 | ht | N | japanese | 17 |
| 38 | c.2520+5G>T | p.Asp823Alafs*5 | 21 | h | ss | pakistani | 25 |
| 39 | c.2527dup G | p.asp843glyfs*3 | 22 | h | fs | pakistani | 24 |
| 40 | c. 2588G>A | p.Arg863His | 22 | h | M | tunisian | 23 |
| 41 | c.2618dupT | p.Lys874Glnfs*40 | 22 | ht | fs | chinese, korean | 26,28 |
| 42 | c.2667_2668GA>TT | p.[Met8891Ile;Lys890*] | 22 | h | | pakistani | 25 |
| 43 | c.2472_2473delAG | p.Gln918 Glyfs*18 | 23 | ht | fs | north european | 18 |
| 44 | ? | p. Asp955Alafs*111 | 23 | h | fs | ? | 23 |
| 45 | c.2864_2867delACAG | p.Asp955Alafs*112 | 23 | ht | N | polish, german | Case 3, 11 |

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|---|---|---|---|---|---|---|---|
| 46 | c.2867+4_2867+7delGGTG | Ser956Cysfs*38 | 23 in | h | ss | turkish | 29 |
| 47 | c.3232G>A | p.Ala1078Thr | 27 | h | mis | pakistani | 22 |
| 48 | c.3335+1G>C | ? | intr | h | ss | italian | 19 |
| 49 | c.3361delG | p.Ala1121Glnfs*6 | 28 | h | fs | pakistani | 12,13 |
| 50 | c.3503G>A | p.Trp1168 | 29 | h | M | pakistani | 13 |
| 51 | c.3839_3855delCAG… | p.Gly1280Alafs*21 | 30 | h | fs | turkish / pakistani | 10,29 |
| 52 | c.3878C>A | p.Ala1293Asp | 30 | h | M | saudi | 21 |
| 53 | c.3936_3937insC | p.Val1314Glyfs*17 | 30 | h | fs | turkish | 14,29 |
| 54 | c.3936dupC | p.Val1314Argfs*18 | 30 | h | N | caucasian | 12,22 |
| 55 | c.4205delTGCC | p.Val1402Glyfs*12 | 31 | h | N | turkish | 10 |
| 56 | c.4241dupT | p.Leu1414Leufs*41 | 31 | h | fs | pakistani, german | 12,22 |
| 57 | c.4258C>T | p.Gln1420* | 31 | ht | N | polish | Case 2 |

h: homozygote, ht: heterozygote, N: nonsense, fs: frameshift, M: missense, del: deletion, ss: splice site mutation.