

Table S1: The list of the mutations in patients with GS.

PTCH1 mutation of Gorlin syndrome

Mutation type	Nucleotide change	Amino acid changes	Exon/intron no.	Characterization	Reference no.
Missense	c.317T>G	p.Leu106Arg	Exon2	Germline	[1]
Missense	c.328G>T	p.Gly110Trp	Exon2	Germline	[2]
Missense	c.591G>C	p.Trp197Cys	Exon4	Germline	[3]
Missense	c.688A>C	p.Thr230Pro	Exon5	Germline	[4]
Missense	c.689C>G	p.Thr230Arg	Exon5	.	[3]
Missense	c.709G>A	p.Glu237Lys	Exon5	Germline	[5]
Missense	c.863G>A	p.Gly288Asp	Exon6	Germline	[6]
Missense	c.890T>C	p.Leu297Pro	Exon6	.	[3]
Missense	c.1115T>G	p.Met372Arg	Exon8	.	[7]
Missense	c.1162A>T	p.Asn388Tyr	Exon8	Germline	[8]
Missense	c.1247C>G	p.Thr416Ser	Exon9	Somatic	[6]
Missense	c.1325T>A	p.Val442Glu	Exon9	Germline	[9]
Missense	c.1346T>A	p.Met449Lys	Exon9	Germline	[10]
Missense	c.1415C>A	p.Ala472Asp	Exon10	Germline	[11]
Missense	c.1436T>G	p.Leu479Arg	Exon10	Germline	[12]
Missense	c.1439C>T	p.Ser480Leu	Exon10	.	[3]
Missense	c.1450G>A	p.Gly484Arg	Exon10	.	[3]
Missense	c.1451G>T	p.Gly484Val	Exon10	Germline	[11]
Missense	c.1450G>A	p.Gly484Arg	Exon10	Germline	[13]
Missense	c.1511C>T	p.Pro504Leu	Exon11	Germline	[13]
Missense	c.1517T>G	p.Leu505Arg	Exon11	Germline	[14]
Missense	c.1525G>C	p.Gln509Arg	Exon11	Germline	[15][16]
Missense	c.1526G>T	p.Gly509Val	Exon11	Germline	[12]
Missense	c.1526G>A	p.Gly509Asp	Exon11	Germline	[3][17]
Missense	c.1555G>C	p.Ala519Pro	Exon11	.	[3]
Missense	c.1660A>C	p.Ser554Arg	Exon12	Germline	[2]
Missense	c.1712G>C	p.Arg571Pro	Exon12	.	[3]
Missense	c.1939A>T	p.Ser647Cys	Exon14	Germline	[1]
Missense	c.1941C>A	p.Ser647Arg	Exon14	Germline	[13]
Missense	c.2183C>T	p.Thr728Met	Exon14	.	[3]
Missense	c.2186A>T	p.Lys729Met	Exon14	Germline	[18][19][20]
Missense	c.2250G>C	p.Lys750Asn	Exon14	.	[3]
Missense	c.2414T>G	p.Ile805Arg	Exon15	.	[3]
Missense	c.2447A>G	p.Gln816Arg	Exon15	.	[3]
Missense	c.2465T>C	p.Leu822Pro	Exon15	Germline	[13]
Missense	c.2776T>C	p.Trp926Arg	Exon17	Germline	[13]
Missense	c.2917C>A	p.Gln973Lys	Exon18	.	[3]
Missense	c.3062A>G	p.Tyr1021Cys	Exon18	Germline	[21]

Missense	c.3146A>T	p.Asn1049Ile	Exon18	Germline	[22]
Missense	c.3277G>C	p.Glu1093Arg	Exon19	Germline	[19],[23]
Missense	c.3394T>C	p.Ser1132Pro	Exon20	Germline	[24]
Missense	c.3395C>A	p.Ser1132Tyr	Exon20	Germline	[16]
Missense	c.3398C>T	p.Thr1133Ile	Exon20	Germline	[25]
Missense	c.3440T>G	p.Phe1147Cys	Exon20	Germline	[6]
Missense	c.3499G>A	p.Gly1167Arg	Exon21	Germline	[6],[12],[26]
Missense	c.3956G>A	p.Arg1319His	Exon23	Germline	[11]
Missense	c.4314G>T	p.Glu1438Asp	Exon23	Germline	[27]
Nonsense	c.205A>T	p.Lys69*	Exon2	Germline	[3]
Nonsense	c.233G>A	p.Trp78*	Exon2	Germline	[28]
Nonsense	c.250C>T	p.Gln84*	Exon2	Germline	[4]
Nonsense	c.279C>A	p.Tyr93*	Exon2	Germline	[29]
Nonsense	c.294C>A	p.Cys98*	Exon2	Germline	[3]
Nonsense	c.305T>A	p.Leu102*	Exon2	Germline	[4]
Nonsense	c.343G>T	p.Gly115*	.	.	Unpublished
Nonsense	c.379G>T	p.Glu127*	Exon2	.	[30]
Nonsense	c.387G>A	p.Trp129*	Exon2	Germline	[31]
Nonsense	c.403C>T	p.Arg135*	Exon3	Somatic	[32],[33]
Nonsense	c.403C>T	p.Arg135*	Exon3	Germline	[34]
Nonsense	c.466C>T	p.Gln156*	Exon3	Germline	[3]
Nonsense	c.478C>T	p.Gln160*	Exon3	Germline	[35]
Nonsense	c.550C>T	p.Gln184*	Exon3	Germline	[36]
Nonsense	c.661G>T	p.Glu221*	Exon5	.	[30]
Nonsense	c.666T>A	p.Tyr222*	.	.	Unpublished
Nonsense	c.672C>A	p.Tyr224*	Exon5	.	[13]
Nonsense	c.707G>A	p.Trp236*	Exon5	.	[3]
Nonsense	c.722T>G	p.Leu241*	.	.	[37]
Nonsense	c.811del	p.Ile271*	.	.	[38]
Nonsense	c.820C>T	p.Gln274*	Exon6	.	[30]
Nonsense	c.833G>A	p.Trp278*	Exon6	Germline	[11]
Nonsense	c.873C>G	p.Tyr291*	Exon6	Germline	[28]
Nonsense	c.938C>G	p.Ser313*	Exon6	.	[30]
Nonsense	c.1012C>T	p.Gln338*	Exon7	Somatic	[32][33]
Nonsense	c.1018G>T	p.Glu340*	Exon7	Germline	[39]
Nonsense	c.1081C>T	p.Gln361*	Exon8	.	[3]
Nonsense	c.1093C>T	p.Gln365*	Exon8	Germline	[40]
Nonsense	c.1119C>G	p.Tyr373*	.	.	[3]
Nonsense	c.1138G>T	p.Glu380*	Exon8	Germline	[12]
Nonsense	c.1148C>G	p.Ser383*	Exon8	Germline	[41]
Nonsense	c.1160G>A	p.Trp387*	.	.	[42]
Nonsense	c.1196G>A	p.Trp399*	Exon8	Germline	[43]

Nonsense	c.1198C>T	p.Gln400*	Exon8	.	[3]
Nonsense	c.1237C>T	p.Gln413*	Exon9	Germline	[19]
Nonsense	c.1380G>A	p.Trp460*	Exon10	.	[3]
Nonsense	c.1585A>T	p.Lys529*	Exon11	Germline	[38]
Nonsense	c.1691T>G	p.Leu564*	Exon12	.	[3]
Nonsense	c.1712G>C	p.Arg571Pro	Exon12	.	[3]
Nonsense	c.1804C>T	p.Arg602*	Exon13	.	[3]
Nonsense	c.1810G>T	p.Glu604*	Exon13	.	[44]
Nonsense	c.1975C>T	p.Gln659*	Exon14	.	[3]
Nonsense	c.1987C>T	p.Gln663*	Exon14	Germline	[19][38]
Nonsense	c.2062C>T	p.Gln688*	Exon14	Germline	[19][34]
Nonsense	c.2098C>T	p.Gln700*	Exon14	.	[3]
Nonsense	c.2170G>T	p.Glu724*	Exon14	.	[3]
Nonsense	c.2308C>T	p.Arg770*	Exon15	.	[3]
Nonsense	c.2359G>T	p.Glu787*	Exon15	.	[3]
Nonsense	c.2391C>A	p.Tyr787*	Exon15	Germline	[45]
Nonsense	c.2446C>T	p.Gln816*	Exon15	.	[3]
Nonsense	c.2557C>T	p.Gln853*	Exon15	.	[3]
Nonsense	c.2619C>A	p.Tyr873*	Exon16	Germline	[1][6][13][46]
Nonsense	c.2760C>A	p.Tyr920*	Exon17	Germline	[26][47]
Nonsense	c.2833C>T	p.Arg945*	Exon17	Germline	[48]
Nonsense	c.2908G>T	p.Glu970*	Exon18	Germline	[2]
Nonsense	c.3027C>A	p.Tyr1009*	Exon18	.	[3]
Nonsense	c.3027C>G	p.Tyr1009*	Exon18	.	[3]
Nonsense	c.3053G>A	p.Trp1018*	Exon18	.	[49]
Nonsense	c.3058C>T	p.Gln1020*	Exon18	Germline	[3][47]
Nonsense	c.3062A>G	p.Tyr1021Cys	Exon18	.	[21]
Nonsense	c.3265T>C	p.Ser1089Pro	.	.	[35]
Nonsense	c.3277G>T	p.Gly1093*	Exon19	.	[30]
Nonsense	c.3422C>T	p.Ala1141Val	.	.	[50]
Small out-of-frame deletions	c.220delA	p.Ala75Argfs*5	Exon2	Germline	[4]
Small out-of-frame deletions	c.239_240delGA	p.Arg80Serfs*9	Exon2	Germline	[43]
Small out-of-frame deletions	c.256_257delCT	p.Leu87Ilefs*2	Exon2	Germline	[34]
Small out-of-frame deletions	c.265_287del23	p.Gly91Argfs*41	Exon2	Germline	[16]
Small out-of-frame deletions	c.278delA	p.Tyr93Serfs*24	Exon2	Germline	[51]
Small out-of-frame deletions	c.331delG	p.Ala111Profs*6	Exon2	Germline	[1]
Small out-of-frame deletions	c.479_482delAGAC	p.Gln160Profs*10	Exon3	Germline	[52]
Small out-of-frame deletions	c.747_759del	p.Gly250Cysfs*15	.	.	Unpublished
Small out-of-frame deletions	c.768_777del10	p.Asn258Leufs*8	Exon6	Germline	[35]
Small out-of-frame deletions	c.808delA	p.Ile271*	Exon6	Germline	[38]
Small out-of-frame deletions	c.865delC	p.His289Metfs*35	Exon6	Germline	[2]
Small out-of-frame deletions	c.912delC	p.Ala306Profs*18	Exon6	Germline	[17]

Small out-of-frame deletions	c.941delC	p.Lys315Asnfs*9	Exon6	Germline	[34]
Small out-of-frame deletions	c.980delG	p.Cys327Phefs*5	Exon7	Germline	[47]
Small out-of-frame deletions	c.1042_1043delAA	p.Lys348Glufs*88	.	.	[50]
Small out-of-frame deletions	c.1208_1209delAT	p.Tyr403Cysfs*33	Exon8	Germline	[13]
Small out-of-frame deletions	c.1291delC	p.Leu431*	Exon9	Germline	[13]
Small out-of-frame deletions	c.1341_1344delACTC	p.Leu446fs*	Exon9	.	[14]
Small out-of-frame deletions	c.1383delC	p.Asp460fs*	Exon10	.	[14]
Small out-of-frame deletions	c.1472delT	p.Ile491Thrfs*51	Exon10	Germline	[53]
Small out-of-frame deletions	c.1615delG	p.Glu539Serfs*3	Exon12	Germline	[4]
Small out-of-frame deletions	c.1670_1671delCA	p.Thr557Serfs*69	Exon12	Germline	[43]
Small out-of-frame deletions	c.2029delA	p.Thr667Profs*16	Exon14	Germline	[43]
Small out-of-frame deletions	c.2011delC	p.His671Thrfs*22	Exon14	Germline	[17]
Small out-of-frame deletions	c.2178delC	p.Cys727Valfs*19	Exon14	Germline	[54]
Small out-of-frame deletions	c.2179delT	p.Cys727Valfs*19	Exon14	Germline	[32][33]
Small out-of-frame deletions	c.2195_2196delTC	p.Ser733Ilefs*4	Exon14	Germline	[34]
Small out-of-frame deletions	c.2196_2197delCT	p.Ser733Ilefs*4	Exon14	Germline	[6]
Small out-of-frame deletions	c.2197_2198delTC	p.Ser733Ilefs*4	Exon14	Germline	[54]
Small out-of-frame deletions	c.2287delG	p.Val763Serfs*9	Exon15	Germline	[4]
Small out-of-frame deletions	c.2395delT	p.Ser799Leufs*7	Exon15	Germline	[29]
Small out-of-frame deletions	c.2442_2452del11	p.Ile815Thrfs*10	Exon15	Germline	[55]
Small out-of-frame deletions	c.2577_2583del7	p.Phe859Leufs*42	Exon16	Germline	[12]
Small out-of-frame deletions	c.2576_2582del7	p.Phe859Leufs*42	Exon16	Germline	[4]
Small out-of-frame deletions	c.2582delG	p.Ser861Metfs*14	Exon16	Germline	[56]
Small out-of-frame deletions	c.2595delC	p.Ile868Serfs*35	Exon16	Germline	[34]
Small out-of-frame deletions	c.2719delC	p.Leu907Trpfs*17	Exon17	Germline	[34]
Small out-of-frame deletions	c.2798delC	p.Ala933Glyfs*29	Exon17	Germline	[11]
Small out-of-frame deletions	c.2824delC	p.Arg942Glyfs*20	Exon17	Germline	[32][33]
Small out-of-frame deletions	c.2833_2843del	p.Arg945Glyfs*10	Exon17	.	[3]
Small out-of-frame deletions	c.2869delC	p.Pro957Leufs*5	Exon17	Germline	[57]
Small out-of-frame deletions	c.3000_3007del8	p.Asn1000Lysfs*142	Exon18	Germline	[27]
Small out-of-frame deletions	c.3016delC	p.Leu1006Cysfs*43	Exon18	Germline	[2]
Small out-of-frame deletions	c.3042delC	p.Phe1015Serfs*34	Exon18	Germline	[13]
Small out-of-frame deletions	c.3375delC	p.Val1126Serfs*13	Exon20	Germline	[4]
Small out-of-frame deletions	c.3475delC	p.Leu1159Serfs*32	Exon21	Germline	[58]
Small out-of-frame deletions	c.3570delG	p.Leu1190*	Exon22	Germline	[27]
Small in-frame deletions	c.114del	p.Leu39Cysfs*41	Exon1	Germline	[3]
Small in-frame deletions	c.254_255del	p.Arg85Thrfs*4	Exon2	Germline	[3]
Small in-frame deletions	c.258_259del	p.Leu87Ilefs*2	Exon2	.	[3]
Small in-frame deletions	c.262_265del	p.Phe88Asnfs*28	Exon2	Germline	[30]
Small in-frame deletions	c.262_266del	p.Phe88Thrfs*50	Exon2	Germline	[3]
Small in-frame deletions	c.290del	p.Asn97Thrfs*20	.	Germline	[51]
Small in-frame deletions	c.331del	p.Ala111Profs*6	.	Germline	[30]

Small in-frame deletions	c.459del	p.Phe153Leufs*6	.	Germline	[59]
Small in-frame deletions	c.479_482del	p.Gln160Profs*10	Exon3	.	[3]
Small in-frame deletions	c.724del	p.Gln242Serfs*8	Exon5	.	[3]
Small in-frame deletions	c.771_780del	p.Asn258Leufs*8	.	.	[35]
Small in-frame deletions	c.842del	p.Met281Serfs*2	Exon6	.	[3]
Small in-frame deletions	c.865del	p.His289Metfs*35	Exon6	Germline	[2]
Small in-frame deletions	c.874del	p.Met292Trpfs*32	.	.	[30]
Small in-frame deletions	c.903del	p.Pro302Glnfs*22	.	.	[60]
Small in-frame deletions	c.922del	p.Ala308Profs*16	.	.	[49]
Small in-frame deletions	c.942del	p.Lys315Asnfs*9	.	.	[34]
Small in-frame deletions	c.1026del	p.Ile342Metfs*25	Exon7	.	[30]
Small in-frame deletions	c.1040_1049del	p.Val347Alafs*17	Exon7	.	[3]
Small in-frame deletions	c.1114del	p.Met372Cysfs*60	Exon8	.	[3]
Small in-frame deletions	c.1208_1209del	p.Tyr403Cysfs*33	Exon8	.	[13]
Small in-frame deletions	c.1279del	p.Leu427Trpfs*5	Exon9	.	[3]
Small in-frame deletions	c.1309_1329del	p.Val437_Ala443del	Exon9	.	[30]
Small in-frame deletions	c.1340_1343del	p.Leu447Serfs*8	Exon9	.	[30]
Small in-frame deletions	c.1342_1345del	p.Leu448Cysfs*7	Exon9	Germline	[28]
Small in-frame deletions	c.1348_1350del	p.Leu450del	Exon10	.	[3]
Small in-frame deletions	c.1415_1429del	p.Ala472_Leu476del	Exon10	.	[3]
Small in-frame deletions	c.1537_1539delGAT or c.1540_1542delGAT	p.Asp513del or p.Asp514del	Exon11	Germline	[61]
Small in-frame deletions	c.1767_1769del	p.Leu590del	Exon13	.	[3]
Small in-frame deletions	c.1852del	p.Cys618Alafs*5	Exon14	.	[3]
Small in-frame deletions	c.(2250+1_2251- 1)_(2703+1_2704-1)del	p.Val751_Gln901del	.	.	[3]
Small in-frame deletions	c.2444_2461del18	p.Ile815_Asp821delinsAsn	Exon15	Germline	[62]
Small in-frame deletions	c.2446_2448delCAG	p.Gln816del	Exon15	Germline	[16]
Small in-frame deletions	c.2612_2615del	p.Asn871Ilefs*31	Exon16	.	[3]
Small in-frame deletions	c.2748del	p.Ser917Alafs*7	Exon17	.	[3]
Small in-frame deletions	c.2793del	p.Val932Serfs*30	Exon17	.	[3]
Small in-frame deletions	c.2833_2843del	p.Arg945Glyfs*10	Exon17	.	[3]
Small in-frame deletions	c.2924del	p.Pro975Leufs*20	Exon17	.	[30]
Small in-frame deletions	c.2993del	p.Cys998Serfs*51	.	.	[50]
Small in-frame deletions	c.3000_3005del6	p.Asn1000_Thr1002delinsLy s	Exon18	Germline	[12]
Small in-frame deletions	c.3042del	p.Phe1015Serfs*34	.	.	[13]
Small in-frame deletions	c.3050del	p.Phe1017Serfs*32	Exon18	.	[3]
Small in-frame deletions	c.3051_3052del	p.Phe1017Leufs*127	.	.	[12]
Small in-frame deletions	c.3055del	p.Glu1019Serfs*30	Exon18	.	[63]
Small in-frame deletions	c.3056_3059del	p.Glu1019Glyfs*29	Exon18	.	[3]
Small in-frame deletions	c.3107_3108del	p.Leu1036Cysfs*108	Exon18	.	[3]

Small in-frame deletions	c.3135del	p.Phe1046Serfs*3	Exon18	.	[3]
Small in-frame deletions	c.3139del	p.Leu1047Phefs*2	Exon18	.	[3]
Small in-frame deletions	c.3139_3142del	p.Leu1047*	Exon18	.	[3]
Small in-frame deletions	c.3146A>T	p.Asn1049Ile	.	.	[3]
Small in-frame deletions	c.3150del	p.Trp1051Glyfs*7	Exon18	.	[3]
Small in-frame deletions	c.3181_3186del	p.Ala1061_Leu1062del	Exon19	Germline	[28]
Small in-frame deletions	c.3233_3239del	p.Leu1078Profs*7	Exon19	.	[3]
Small in-frame deletions	c.3244_3249del6	p.Pro1082_Val1083del	Exon19	Germline	[16]
Small in-frame deletions	c.3251_3272del	p.Val1084Alafs*2	Exon19	.	[3]
Small in-frame deletions	c.3280del	p.Val1094Trpfs*12	.	.	[30]
Small in-frame deletions	c.3364_3365del	p.Met1122Valfs*22	Exon20	.	[3]
Small in-frame deletions	c.3371del	p.Ala1124Aspfs*15	.	.	[30]
Small in-frame deletions	c.3375del	p.Val1126Serfs*13	Exon20	.	[3]
Small in-frame deletions	c.3394T>C	p.Ser1132Pro	.	.	[24]
Small in-frame deletions	c.3475del	p.Leu1159Serfs*32	Exon21	.	[58]
Small in-frame deletions	c.3525_3526del	p.Leu1175Phefs*8	Exon21	.	[3]
Small in-frame deletions	c.3531_3534del	p.Phe1177Leufs*13	.	.	[64]
Small in-frame deletions	c.4048C>T	p.Arg1350Trp	Exon23	.	[3]
Small out-of-frame insertions	c.264_265insAATA	p.Lys89Asnfs*52	Exon2	Somatic	[53]
Small out-of-frame insertions	c.282_283insA	p.Gln95Thrfs*45	Exon2	Germline	[34]
Small out-of-frame insertions	c.323_324insGA	p.Ile108Metfs*10	Exon2	Germline	[12]
Small out-of-frame insertions	c.361_362insGAGC	p.Leu121Argfs*20	Exon2	Somatic	[1]
Small in-frame insertions	c.1338_1339insGCG	p.Tyr446_Leu447insAla	Exon9	Germline	[1][46]
Small in-frame insertions	c.2178_2179insA	p.Cys727Metfs*11	Exon14	.	[3]
Small duplications	c.280dup	p.Ile94Asnfs*46	Exon2	Germline	[13]
Small duplications	c.290dupA	p.Asn97Lysfs*43	Exon2	Germline	[65]
Small duplications	c.385_386dup	p.Trp129Cysfs*9	Exon2	Germline	[3]
Small duplications	c.572_575dup	p.Met192Ilefs*61	Exon3	Germline	[3]
Small duplications	c.705dupC	p.Trp236Leufs*16	Exon5	Germline	[27]
Small duplications	c.919dup	p.Thr307Asnfs*12	.	.	[30]
Small duplications	c.931dupA	p.Asn312Lysfs*7	Exon6	Germline	[18][19]
Small duplications	c.935dup	p.Asn312Lysfs*7	.	.	[12]
Small duplications	c.955dup	p.Met319Asnfs*118	.	.	[30]
Small duplications	c.1027_1028dupGT	p.Gly344Thrfs*24	Exon7	Germline	[4]
Small duplications	c.1028_1029dup	p.Gly344Trpfs*24	.	.	[4]
Small duplications	c.1043_1046dupAGAA	p.Asn349Lysfs*88	Exon7	Germline	[66]
Small duplications	c.1100_1108dup	p.Met367_Pro369dup	.	.	[30]
Small duplications	c.1138_1195dup58	p.Trp399*	Exon8	Germline	[43]

Small duplications	c.1259dupT	p.Ser421Phefs*16	Exon9	Germline	[67]
Small duplications	c.1261dupT	p.Ser421Phefs*16	Exon9	Germline	[17]
Small duplications	c.1349dupT	p.Ala451Argfs*46	Exon10	Germline	[68]
Small duplications	c.1366dup	p.Thr456Asnfs*41	Exon10	.	[3]
Small duplications	c.1370dupT	p.Met457Ilefs*40	Exon10	Germline	[69]
Small duplications	c.1353_1377dup	p.Trp460Leufs*45	.	.	[70]
Small duplications	c.1467dupA	p.Leu490Ilefs*7	Exon10	Germline	[71]
Small duplications	c.1508dup	p.Leu503Phefs*12	Exon11	.	[3]
Small duplications	c.1651dupA	p.Thr551Asnfs*76	Exon12	Germline	[34]
Small duplications	c.1723dupC	p.Leu575Profs*52	Exon12	Germline	[34]
Small duplications	c.1925dup	p.Pro643Thrfs*11	Exon14	.	[3]
Small duplications	c.2011dup	p.His671Profs*10	Exon14	.	[3]
Small duplications	c.2011_2012dupCA	p.His673Argfs*21	Exon14	Germline	[12]
Small duplications	c.2178dupC	p.Cys727Leufs*17	Exon14	Germline	[13]
Small duplications	c.2256_2262dup7	p.Phe755Glyfs*37	Exon15	Germline	[72]
Small duplications	c.2392dupC	p.Phe798Leufs*31	Exon15	Germline	[71]
Small duplications	c.2454dupA	p.Leu819Thrfs*10	Exon15	Germline	[2]
Small duplications	c.2535dupG	p.His846Argfs*15	Exon15	Germline	[73]
Small duplications	c.2712dupA	p.Gln905Thrfs*11	Exon17	Germline	[47]
Small duplications	c.2795_2799dup5	p.Tyr934Serfs*30	Exon17	Germline	[16]
Small duplications	c.2918_2925dup8	p.Phe976Serfs*22	Exon18	Germline	[11]
Small duplications	c.2960_2961dup	p.Val988Leufs*8	.	.	[49]
Small duplications	c.3025dup	p.Tyr1009Leufs*136	.	.	[74]
Small duplications	c.3026dupA	p.Tyr1009*	Exon18	Germline	[27]
Small duplications	c.3244_3246dupCCC	p.Pro1082dup	Exon19	Germline	[6]
Small duplications	c.3325_3328dupGGCG	p.Asp1110Glyfs*36	Exon20	Germline	[75]
Small duplications	c.3497dup	p.Asn1166Lysfs*18	Exon21	.	[3]

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