

Supplementary Online Content

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eTable 1. 156 XP Disease Associated Missense or Nonsense Mutations in *XPA-G* genes from HGMD database and NIH cohort

eTable 2. 65 XP Mutations Listed in gnomAD Database

This supplementary material has been provided by the authors to give readers additional information about their work.

eTable 1. 156 XP Disease Associated Missense or Nonsense Mutations in *XPA-G* genes from HGMD database and NIH cohort

Complement- ation Group	Gene	Amino Acid Number	Clinical Phenotype*	Mutation Frequency Listed in GnomAD Database?	rs Number**	Protein Change	cDNA
A	<i>XPA</i>	85	XP	NO	n/a***	p.Q85X	c. 253C>T
A	<i>XPA</i>	94	XP	NO	n/a	p.P94L	c.281C>T
A	<i>XPA</i>	95	XP	NO	n/a	p. G95R	c.283G>A
A	<i>XPA</i>	105	XP	NO	n/a	p.C105Y	c. 314G>A
A	<i>XPA</i>	108	XP	YES	104894131	p.C108F	c.323G>T
A	<i>XPA</i>	111	XP	YES	769255883	p.E111X	c.331G>T
A	<i>XPA</i>	116	XP	NO	104894134	p.Y116X	c.348T>A
A	<i>XPA</i>	126	XP	NO	n/a	p.C126W	c.378T>G
A	<i>XPA</i>	126	XP	NO	n/a	p.C126Y	c.377G>A
A	<i>XPA</i>	151	XP	NO	n/a	p.K151X	c.451A>T
A	<i>XPA</i>	185	XP	NO	n/a	p.Q185X	c.553C>T
A	<i>XPA</i>	207	XP	YES	104894133	p.R207X	c.619C>T
A	<i>XPA</i>	208	XP	NO	n/a	p.Q208X	c.622C>T
A	<i>XPA</i>	211	XP	YES	149226993	p.R211X	c.631C>T
A	<i>XPA</i>	228	XP	YES	104894132	p.R228X	c.682C>T
A	<i>XPA</i>	244	XP	YES	104894132	p.H244R	c.731A>G
B	<i>XPB (ERCC3)</i>	425	XP	YES	121913047	p.R425X	c.1273C>T
B	<i>XPB (ERCC3)</i>	545	XP/CS	YES	121913048	p.Q545X	c.1633C>T
C	<i>XPC</i>	1	XP	NO	760324503	p.M1R	c.2T>G
C	<i>XPC</i>	52	XP	NO	n/a	p.S52X	c.155C>G
C	<i>XPC</i>	149	XP	NO	n/a	p.E149X	c.445G>T
C	<i>XPC</i>	155	XP	YES	755825264	p.R155X	c.463C>T
C	<i>XPC</i>	183	XP	NO	n/a	p.K183X	c.547A>T
C	<i>XPC</i>	220	XP	YES	745679643	p.R220X	c.658C>T
C	<i>XPC</i>	247	XP	YES	764321665	p.R247X	c.739C>T
C	<i>XPC</i>	260	XP	NO	n/a	p.W260X	c.780G>A
C	<i>XPC</i>	284	XP	NO	n/a	p.E284X	c. 850G>T
C	<i>XPC</i>	293	XP	YES	373519125	p.R293X	c. 877C>T
C	<i>XPC</i>	320	XP	NO	n/a	p.Q320X	c. 958C>T
C	<i>XPC</i>	334	XP	YES	74737358	p.P334H	c.1001C>A
C	<i>XPC</i>	341	XP	NO	n/a	p.A341S	c. 1021G>T
C	<i>XPC</i>	415	XP	YES	757958943	p.R415X	c.1243C>T
C	<i>XPC</i>	467	XP	NO	n/a	p.Q467X	c.1399C>T
C	<i>XPC</i>	522	XP	YES	746250060	p.K522X	c.1564A>T
C	<i>XPC</i>	554	XP	NO	n/a	p.Q554X	c.1660C>T
C	<i>XPC</i>	559	XP	YES	767569346	p.Y559X	c.1677C>G
C	<i>XPC</i>	568	XP	NO	n/a	p.Y568X	c.1704T>A
C	<i>XPC</i>	579	XP	YES	121965088	p.R579X	c.1735C>T
C	<i>XPC</i>	585	XP	YES	749681216	p.Y585C	c. 1754A>G
C	<i>XPC</i>	603	XP	YES	767209381	p. W603X	c. 1808G>A
C	<i>XPC</i>	690	XP	NO	n/a	p.W690S	c.2069G>C
C	<i>XPC</i>	692	XP	YES	374117852	p.K692X	c.2074A>T
C	<i>XPC</i>	703	XP	NO	n/a	p.P703L	c.2108C>T
C	<i>XPC</i>	718	XP	NO	754775337	p.R718X	c.2152C>T
C	<i>XPC</i>	738	XP	NO	n/a	p.T738A	c.2212A>G
D	<i>XPB (ERCC2)</i>	47	XP	NO	n/a	p. G47R	c.139 G>A
D	<i>XPB (ERCC2)</i>	51	XP	NO	n/a	p.S51F	c.152C>T
D	<i>XPB (ERCC2)</i>	76	XP	NO	n/a	p. T76A	c.226A>G
D	<i>XPB (ERCC2)</i>	234	XP	YES	n/a	p.D234N	c.700G>A
D	<i>XPB (ERCC2)</i>	292	XP	NO	n/a	p.L292P	c.T953C(T23532C)
D	<i>XPB (ERCC2)</i>	368	XP	NO	n/a	p.Q368X	c.1102C>T

D	XPD (ERCC2)	452	XP	YES	199643821	p.Q452X	c.1354C>T
D	XPD (ERCC2)	485	XP	NO	121913025	p.L485P	c.1454 T>C
D	XPD (ERCC2)	511	XP	NO	772572683	p.R511Q	c.1532G>A
D	XPD (ERCC2)	541	XP	YES	121913019	p.S541R	c.1621A>C
D	XPD (ERCC2)	542	XP	NO	n/a	p.Y542C	c.1625A>G
D	XPD (ERCC2)	595	XP	NO	n/a	p.I595S	c.1784T>G
D	XPD (ERCC2)	601	XP	YES	140522180	p.R601L	c.1802G>T
D	XPD (ERCC2)	601	XP	YES	753641926	p.R601W	c.1801C>T
D	XPD (ERCC2)	602	XP	YES	771824813	p.G602D	c.1805G>A
D	XPD (ERCC2)	611	XP	NO	n/a	p.G611R	n/a
D	XPD (ERCC2)	616	XP/CS	NO	n/a	p.R616G	c.1846C>G
D	XPD (ERCC2)	616	XP	YES	121913024	p.R616W	c.1846C>T
D	XPD (ERCC2)	638	XP	NO	n/a	p.R638Q	n/a
D	XPD (ERCC2)	666	XP	YES	752510317	p.R666W	c.1996C>T
D	XPD (ERCC2)	675	XP	NO	n/a	p.G675R	c.2023G>C
D	XPD (ERCC2)	683	XP	YES	41556519	p.R683W	c.2047C>T
D	XPD (ERCC2)	683	XP	YES	758439420	p.R683Q	c.2048G>A
D	XPD (ERCC2)	726	XP	NO	121913017	p.Q726X	c.2176C>T
D	XPD (ERCC2)	681	TTD or XP	YES	121913023	p.D681N	c.2041G>A
D	XPD (ERCC2)	378	TTD or XP	YES	200043231	p.R378H	c.1133G>A
D	XPD (ERCC2)	616	TTD or XP	YES	376556895	p.R616P	c.1847G>C
D	XPD (ERCC2)	662	TTD or XP	YES	778479250	p.Q662X	c.1984 C>T
D	XPD (ERCC2)	681	TTD, XP or CS	NO	n/a	p.D681H	c.2041G>C
D	XPD (ERCC2)	461	XP or TTD	YES	121913016	p.L461V plus p.V716-R730del	c.1381 C>G
D	XPD (ERCC2)	112	XP or TTD	YES	121913020	p.R112H	c.335G>A
D	XPD (ERCC2)	259	XP or TTD	YES	370454709	p.C259Y	c.776G>A
E	XPE (DDB2)	54	XP	NO	n/a	p.W54*	c.161G>A
E	XPE (DDB2)	192	XP	NO	199822504	p.R192X	c.574C>T
E	XPE (DDB2)	239	XP	NO	n/a	p.R239I	c.716G>T
E	XPE (DDB2)	244	XP	NO	121434639	p.K244E	c.730A>G
E	XPE (DDB2)	273	XP	NO	121434640	p.R273H	c.818G>A
E	XPE (DDB2)	305	XP	NO	886048361	p.T305N	c.914C>A
E	XPE (DDB2)	307	XP	NO	121434642	p.D307Y	c.919G>T
E	XPE (DDB2)	313	XP	NO	121434641	p.R313X	c.937C>T
E	XPE (DDB2)	350	XP	NO	n/a	p.L350P	c.1049T>C
E	XPE (DDB2)	357	XP	NO	780665825	p.P357L	c.1070C>T
F	XPF (ERCC4)	225	XP	YES	764731249	p.I225M	c.675A>G
F	XPF (ERCC4)	379	XP	YES	1799802	p.P379S	c.1135C>T
F	XPF (ERCC4)	454	XP	NO	n/a	p.R454W	c.1360A>T
F	XPF (ERCC4)	490	XP	NO	912480692	p.R490Q	c.1469G>A
F	XPF (ERCC4)	502	XP	NO	n/a	p.E502K	c.1504G>A
F	XPF (ERCC4)	513	XP	YES	769679311	p.G513R	c.1537G>A
F	XPF (ERCC4)	529	XP	NO	n/a	p.I529T	c.1586T>C
F	XPF (ERCC4)	567	XP	NO	n/a	p.T567A	c.1699A>G
F	XPF (ERCC4)	589	XP	YES	147105770	p.R589W	c.1765C>T
F	XPF (ERCC4)	608	XP	NO	n/a	p.L608P	c.1823T>C
F	XPF (ERCC4)	611	XP	YES	749814308	p.G611R	c.1831G>A
F	XPF (ERCC4)	799	XP	YES	121913049	p.R799W	c.2395C>T
G	XPG (ERCC5)	2	XP	NO	n/a	p.G2W	c.4G>T
G	XPG (ERCC5)	11	XP	NO	n/a	p.E11X	c.31G>T
G	XPG (ERCC5)	16	XP	NO	n/a	p.Q16X	c.46C>T
G	XPG (ERCC5)	28	XP	NO	267607281	p.A28D	c.83C>A
G	XPG (ERCC5)	37	XP	NO	761470006	p.Q37X	c.109C>T
G	XPG (ERCC5)	65	XP	NO	n/a	p.L65P	c.194T>C
G	XPG (ERCC5)	72	XP	NO	121434574	p.P72H	c.215C>A
G	XPG (ERCC5)	136	XP	NO	121434577	p.Q136X	c.406C>T
G	XPG (ERCC5)	150	XP	NO	n/a	p.Q150X	c.448C>T
G	XPG (ERCC5)	176	XP	NO	121434573	p.Q176X	c.526C>T
G	XPG (ERCC5)	263	XP	YES	121434572	p.R263X	c.787C>T
G	XPG (ERCC5)	290	XP	NO	n/a	p.I290N	c.869T>A
G	XPG (ERCC5)	585	XP	NO	n/a	p.E585X	c.1753G>T

G	<i>XPG (ERCC5)</i>	727	XP	NO	n/a	p.E727X	c.2179G>T
G	<i>XPG (ERCC5)</i>	778	XP	YES	929424117	p.L778P	c.2333T>C
G	<i>XPG (ERCC5)</i>	792	XP	YES	121434571	p.A792V	c.2375C>T
G	<i>XPG (ERCC5)</i>	795	XP	YES	751772171	p.A795T	c.2383G>A
G	<i>XPG (ERCC5)</i>	818	XP	YES	774078839	p.A818V	c.2453C>T
G	<i>XPG (ERCC5)</i>	858	XP	NO	121434575	p.L858P	c.2573T>C
G	<i>XPG (ERCC5)</i>	874	XP	YES	121434576	p.A874T	c.2620G>A
G	<i>XPG (ERCC5)</i>	960	XP	YES	121434570	p.E960X	c.2878G>T
G	<i>XPG (ERCC5)</i>	968	XP	NO	267607280	p.W968C	c.2904G>C
G	<i>XPG (ERCC5)</i>	138	XP/CS	YES	188898060	p.R138X	c.412C>T
G	<i>XPG (ERCC5)</i>	798	XP/CS	NO	n/a	p.D798Y	c.2392G>T
G	<i>XPG (ERCC5)</i>	805	XP/CS	YES	899478069	p.G805R	c.2413G>A
G	<i>XPG (ERCC5)</i>	814	XP/CS	NO	n/a	p.W814S	c.2441G>C
variant	<i>POLH</i>	9	XP	YES	781288440	p.V9F	c.25G>T
variant	<i>POLH</i>	23	XP	NO	n/a	p.Q23X	c.67C>T
variant	<i>POLH</i>	111	XP	YES	758423288	p.R111H	c.332G>A
variant	<i>POLH</i>	117	XP	NO	n/a	p.A117P	c.349G>C
variant	<i>POLH</i>	122	XP	NO	n/a	p.T122P	c.364A>C
variant	<i>POLH</i>	126	XP	YES	121908563	p.Q126X	c.376C>T
variant	<i>POLH</i>	152	XP	NO	n/a	p.Q152X	c.454C>T
variant	<i>POLH</i>	167	XP	NO	n/a	p.R167X	c.499C>T
variant	<i>POLH</i>	174	XP	NO	n/a	p.W174C	c.522G>T
variant	<i>POLH</i>	174	XP	NO	n/a	p.W174X	c.521G>A
variant	<i>POLH</i>	220	XP	YES	n/a	p.K220E	c.658A>G
variant	<i>POLH</i>	228	XP	NO	n/a	p.G228X	c.682G>T
variant	<i>POLH</i>	242	XP	YES	745778317	p.S242X	c.725C>G
variant	<i>POLH</i>	263	XP	YES	n/a	p.G263V	c.788G>T
variant	<i>POLH</i>	264	XP	YES	778739941	p.A264P	c.790G>C
variant	<i>POLH</i>	266	XP	NO	n/a	p.V266D	c.797T>A
variant	<i>POLH</i>	295	XP	NO	n/a	p.G295R	c.883G>A
variant	<i>POLH</i>	297	XP	NO	n/a	p.W297X	c.890G>A
variant	<i>POLH</i>	299	XP	YES	190423114	p.Y299X	c.897T>G
variant	<i>POLH</i>	303	XP	YES	759607901	p.R303X	c.907C>T
variant	<i>POLH</i>	306	XP	NO	121908562	p.E306X	c.916G>T
variant	<i>POLH</i>	356	XP	YES	559497462	p.R356X	c.1066C>T
variant	<i>POLH</i>	361	XP	NO	n/a	p.R361S	c.1083G>T
variant	<i>POLH</i>	373	XP	YES	121908564	p.Q373X	c.1117C>T
variant	<i>POLH</i>	482	XP	NO	n/a	p.S482X	c.1445C>A
variant	<i>POLH</i>	521	XP	NO	n/a	p.Q521X	c.1561C>T
variant	<i>POLH</i>	535	XP	YES	56307355	p.K535E	c.1603A>G
variant	<i>POLH</i>	548	XP	NO	n/a	p.S548X	c.1643C>A
variant	<i>POLH</i>	589	XP	YES	121908565	p.K589T	c.1766A>C
variant	<i>POLH</i>	692	XP	YES	199562456	p.T692A	c.2074A>G
variant	<i>POLH</i>	714	XP	NO	n/a	p.X714W	c.2141A>G

*XP- xeroderma pigmentosum; TTD - trichothiodystrophy; CS - Cockayne syndrome; XP/CS xeroderma pigmentosum - Cockayne syndrome complex

**from dbSNP

***none reported

eTable 2. 65 XP Mutations Listed in gnomAD Database

Complementati on Group	Gene	Amino Acid Num- ber	Clinical Pheno- type*	Protein Change	cDNA	rs Number**	Allele Count gnomAD	Number of Homozygotes gnomAD	Total Alleles Sequenced gnomAD	gnomAD Allele Frequency (q)	Sum (q) for Each Gene	Estimated Frequency of Homozygous Affected Individ- uals in gnomAD (q ²)	Sum (q ²) of Each Gene
A	XPA	108	XP	p.C108F	c.323G>T	n/a***	1	0	245202	4.08E-06		1.66E-11	
A	XPA	111	XP	p.E111X	c.331G>T	n/a	5	0	276416	1.81E-05		3.27E-10	
A	XPA	207	XP	p.R207X	c.619C>T	104894133	6	0	245910	2.44E-05		5.95E-10	
A	XPA	211	XP	p.R211X	c.631C>T	149226993	1	0	245906	4.07E-06		1.65E-11	
A	XPA	228	XP	p.R228X	c.682C>T	104894132	95	0	276474	3.44E-04		1.18E-07	
A	XPA	244	XP	p.H244R	c.731A>G	104894132	38	0	276984	1.38E-04	0.053%	1.91E-08	1.38E-07
B	XPB (ERCC3)	425	XP	p.R425X	c.1273C>T	121913047	2	0	246178	8.12E-06		6.60E-11	
B	XPB (ERCC3)	545	XP/CS	p.Q545X	c.1633C>T	121913048	1	0	246244	4.06E-06	0.001%	1.65E-11	8.25E-11
C	XPC	155	XP	p.R155X	c.463C>T	755825264	4	0	246142	1.63E-05		2.64E-10	
C	XPC	220	XP	p.R220X	c.658C>T	745679643	6	0	275118	2.18E-05		4.76E-10	
C	XPC	247	XP	p.R247X	c.739C>T	764321665	1	0	30964	3.23E-05		1.04E-09	
C	XPC	293	XP	p.R293X	c.877C>T	373519125	4	0	245672	1.63E-05		2.65E-10	
C	XPC	334	XP	p.P334H	c.1001C>A	74737358	838	7	274914	3.05E-03		9.29E-06	
C	XPC	415	XP	p.R415X	c.1243C>T	757958943	3	0	246190	1.22E-05		1.49E-10	
C	XPC	522	XP	p.K522X	c.1564A>T	746250060	1	0	246248	4.06E-06		1.65E-11	
C	XPC	559	XP	p.Y559X	c.1677C>G	767569346	1	0	246260	4.06E-06		1.65E-11	
C	XPC	579	XP	p.R579X	c.1735C>T	121965088	3	0	246248	1.22E-05		1.48E-10	
C	XPC	585	XP	p.Y585X	c.1754A>G	749681216	1	0	246248	4.06E-06		1.65E-11	
C	XPC	603	XP	p.W603X	c.1808G>A	767209381	2	0	246090	8.13E-06		6.60E-11	
C	XPC	692	XP	p.K692X	c.2074A>T	374117852	3	0	276036	1.09E-05	0.319%	1.18E-10	9.29E-06
D	XPD (ERCC2)	234	XP	p.D234N	c.700G>A	n/a	1	0	245512	4.07E-06		1.66E-11	
D	XPD (ERCC2)	541	XP	p.S541R	c.1621A>C	121913019	1	0	246242	4.06E-06		1.65E-11	
D	XPD (ERCC2)	601	XP	p.R601L	c.1802G>T	140522180	6	0	276602	2.17E-05		4.70E-10	
D	XPD (ERCC2)	601	XP	p.R601W	c.1801C>T	753641926	12	0	276668	4.34E-05		1.88E-09	
D	XPD (ERCC2)	602	XP	p.G602D	c.1805G>A	771824813	1	0	245708	4.07E-06		1.66E-11	
D	XPD (ERCC2)	616	XP	p.R616W	c.1846C>T	121913024	16	0	276718	5.78E-05		3.34E-09	
D	XPD (ERCC2)	666	XP	p.R666W	c.1996C>T	752510317	3	0	246154	1.22E-05		1.49E-10	
D	XPD (ERCC2)	683	XP	p.R683W	c.2047C>T	41556519	16	0	276886	5.78E-05		3.34E-09	
D	XPD (ERCC2)	683	XP	p.R683Q	c.2048G>A	758439420	4	0	246000	1.59E-05		2.52E-10	
D	XPD (ERCC2)	681	XP or TTD	p.D681N	c.2041G>A	121913023	5	0	246130	2.03E-05		4.12E-10	
D	XPD (ERCC2)	378	XP or TTD	p.R378H	c.1133G>A	200043231	3	0	246128	1.22E-05		1.49E-10	
D	XPD (ERCC2)	616	XP or TTD	p.R616P	c.1847G>C	376556895	38	0	276686	1.37E-04		1.89E-08	
D	XPD (ERCC2)	662	XP or TTD	p.Q662X	c.1984 C>T	778479250	2	0	277088	7.22E-06		5.21E-11	
D	XPD (ERCC2)	461	XP or TTD	p.L461V plus p.V716-R730del	c.1381 C>G	121913016	314	2	276906	1.13E-03		1.29E-06	
D	XPD (ERCC2)	112	XP or TTD	p.R112H	c.335G>A	121913020	6	0	277006	2.17E-05		4.69E-10	
D	XPD (ERCC2)	259	XP or TTD	p.C259Y	c.776G>A	370454709	18	0	276676	6.51E-05	0.162%	4.23E-09	1.32E-06
F	XPF (ERCC4)	225	XP	p.I225M	c.675A>G	764731249	1	0	246128	4.06E-06		1.65E-11	
F	XPF (ERCC4)	379	XP	p.P379S	c.1135C>T	1799802	1122	4	276560	4.05E-03		1.64E-05	
F	XPF (ERCC4)	513	XP	p.G513R	c.1537G>A	769679311	1	0	246060	4.06E-06		1.65E-11	
F	XPF (ERCC4)	589	XP	p.R589W	c.1765C>T	147105770	16	0	274824	5.82E-05		3.39E-09	
F	XPF (ERCC4)	611	XP	p.G611R	c.1831G>A	749814308	3	0	277130	1.08E-05		1.17E-10	
F	XPF (ERCC4)	799	XP	p.R799W	c.2395C>T	121913049	124	0	277034	4.48E-04	0.457%	2.00E-07	1.66E-05
G	XPG (ERCC5)	263	XP	p.R263X	c.787C>T	121434572	1	0	246138	4.06E-06		1.65E-11	

G	<i>XPG (ERCC5)</i>	778	XP	p.L778P	c.2333T>C	n/a	1	0	245310	4.08E-06	1.66E-11			
G	<i>XPG (ERCC5)</i>	792	XP	p.A792V	c.2375C>T	121434571	6	0	246078	2.44E-05	5.94E-10			
G	<i>XPG (ERCC5)</i>	795	XP	p.A795T	c.2383G>A	751772171	8	0	277096	2.89E-05	8.33E-10			
G	<i>XPG (ERCC5)</i>	818	XP	p.A818V	c.2453C>T	774078839	11	0	246172	4.47E-05	2.00E-09			
G	<i>XPG (ERCC5)</i>	874	XP	p.A874T	c.2620G>A	121434576	5	0	246258	2.03E-05	4.12E-10			
G	<i>XPG (ERCC5)</i>	960	XP	p.E960X	c.2878G>T	121434570	2	0	246116	8.13E-06	6.60E-11			
G	<i>XPG (ERCC5)</i>	138	XP/CS	p.R138X	c.412C>T	188898060	2	0	246240	8.12E-06	6.60E-11			
G	<i>XPG (ERCC5)</i>	805	XP/CS	p.G805R	c.2413G>A	n/a	3	0	246196	1.22E-05	0.015%	1.49E-10	4.15E-09	
variant	<i>POLH</i>	9	XP	p.V9F	c.25G>T	781288440	17	0	277246	6.13E-05		3.76E-09		
variant	<i>POLH</i>	111	XP	p.R111H	c.332G>A	758423288	11	0	277250	3.97E-05		1.57E-09		
variant	<i>POLH</i>	126	XP	p.Q126X	c.376C>T	121908563	1	0	246262	4.06E-06		1.65E-11		
variant	<i>POLH</i>	220	XP	p.K220E	c.658A>G	n/a	1	0	246218	4.06E-06		1.65E-11		
variant	<i>POLH</i>	242	XP	p.S242X	c.725C>G	745778317	2	0	246246	8.12E-06		6.60E-11		
variant	<i>POLH</i>	263	XP	p.G263V	c.788G>T	n/a	1	0	246184	4.06E-06		1.65E-11		
variant	<i>POLH</i>	264	XP	p.A264P	c.790G>C	778739941	4	0	246176	1.63E-05		2.64E-10		
variant	<i>POLH</i>	299	XP	p.Y299X	c.897T>G	190423114	1	0	246218	4.06E-06		1.65E-11		
variant	<i>POLH</i>	303	XP	p.R303X	c.907C>T	759607901	3	0	246202	1.22E-05		1.49E-10		
variant	<i>POLH</i>	356	XP	p.R356X	c.1066C>T	559497462	6	0	243794	2.46E-05		6.06E-10		
variant	<i>POLH</i>	373	XP	p.Q373X	c.1117C>T	121908564	1	0	246148	4.06E-06		1.65E-11		
variant	<i>POLH</i>	535	XP	p.K535E	c.1603A>G	56307355	234	3	276598	8.42E-04		7.10E-07		
variant	<i>POLH</i>	589	XP	p.K589T	c.1766A>C	121908565	6	0	246216	2.44E-05		5.94E-10		
variant	<i>POLH</i>	692	XP	p.T692A	c.2074A>G	199562456	42	0	276998	1.52E-04	0.120%	2.30E-08	7.40E-07	
TOTALS		65	65	65		65	58	3,098	16	253,995	1.13%	1.13%	2.81E-05	2.81E-05

*XP- xeroderma pigmentosum; TTD - trichothiodystrophy; CS - Cockayne syndrome; XP/CS xeroderma pigmentosum - Cockayne syndrome complex

**from dbSNP

***none reported