

Supplementary Table 7: Putative pathogenic missense variants identified in each gene based on *in silico* software programs SIFT, Polyphen-2 and Provean

cDNA position	Protein	Damaging*	Controls	Cases	Total
APEX1					
c.56C>G	p.T19R	No	0	1	1
c.62C>T	p.P21L	No	3	1	4
c.103A>C	p.K35Q	No	4	1	5
c.128C>T	p.A43V	No	1	0	1
c.190A>G	p.I64V	No	1	1	2
c.286G>A	p.E96K	Yes	1	0	1
c.319G>A	p.E107K	Yes	1	0	1
c.385A>G	p.S129G	No	0	1	1
c.407G>A	p.R136H	No	1	0	1
c.413G>A	p.C138Y	No	1	1	2
c.442G>A	p.D148N	No	1	0	1
c.455A>C	p.D152A	Yes	1	0	1
c.467G>A	p.R156Q	Yes	0	1	1
c.542G>A	p.R181Q	No	0	1	1
c.560G>A	p.R187H	Yes	3	4	7
c.577C>T	p.R193C	No	0	1	1
c.578G>C	p.R193P	Yes	0	1	1
c.578G>A	p.R193H	No	1	1	2
c.709C>T	p.R237C	Yes	1	0	1
c.743C>T	p.P248L	No	1	1	2
c.756C>A	p.S252R	Yes	0	1	1
c.808A>G	p.M270V	No	0	1	1
c.827A>G	p.K276R	No	0	1	1
c.916G>A	p.G306S	Yes	0	1	1
c.920G>A	p.S307N	Yes	1	0	1
c.931C>A	p.P311T	Yes	0	1	1
APLF					
c.28C>T	p.R10W	No	2	1	3
c.31G>A	p.D11N	No	1	0	1
c.41C>T	p.P14L	Yes	0	1	1
c.56C>T	p.A19V	No	0	1	1
c.58C>T	p.P20S	No	0	1	1
c.86C>T	p.P29L	Yes	0	1	1
c.88C>A	p.L30M	Yes	1	0	1
c.125A>T	p.H42L	Yes	18	28	46
c.245A>G	p.Y82C	No	39	58	97
c.326T>C	p.M109T	No	0	1	1
c.413A>G	p.H138R	No	1	0	1
c.428C>T	p.A143V	No	1	0	1

c.505A>G	p.N169D	No	2	4	6
c.512A>G	p.D171G	No	1	0	1
c.518A>G	p.N173S	No	1	0	1
c.541G>A	p.E181K	Yes	0	1	1
c.545G>A	p.R182K	Yes	2	3	5
c.555C>G	p.I185M	Yes	0	1	1
c.575C>T	p.A192V	No	0	2	2
c.631A>G	p.I211V	No	0	1	1
c.666T>A	p.D222E	No	0	2	2
c.793A>G	p.I265V	No	0	1	1
c.808A>G	p.M270V	No	0	1	1
c.847G>A	p.E283K	No	0	1	1
c.890C>A	p.P297Q	Yes	1	0	1
c.937A>G	p.R313G	No	0	1	1
c.949A>G	p.K317E	No	0	1	1
c.974C>G	p.S325C	Yes	1	0	1
c.1016A>T	p.E339V	Yes	1	0	1
c.1073A>T	p.H358L	No	1	0	1
c.1130C>T	p.T377I	No	0	3	3
c.1142A>G	p.Y381C	Yes	2	3	5
c.1276T>C	p.S426P	Yes	4	0	4
c.1427A>G	p.Y476C	No	1	0	1
c.1439A>G	p.D480G	Yes	0	1	1
c.1480A>G	p.K494E	No	0	1	1
c.1509A>T	p.E503D	No	1	0	1
c.1517G>A	p.R506K	No	1	1	2
c.1519T>C	p.F507L	Yes	0	1	1
APTX					
c.971A>T	p.Q324L	No	23	19	42
c.946C>T	p.P316S	Yes	0	1	1
c.926T>C	p.M309T	No	0	1	1
c.742T>A	p.L248M	No	10	23	33
c.740G>A	p.R247Q	No	2	1	3
c.671T>C	p.L224P	Yes	1	0	1
c.617C>T	p.P206L	Yes	0	1	1
c.587C>T	p.P196L	Yes	1	1	2
c.553G>A	p.D185N	Yes	0	1	1
c.457A>G	p.K153E	No	1	0	1
c.431C>A	p.S144Y	No	55	78	133
c.430T>C	p.S144P	No	0	1	1
c.426C>G	p.S142R	No	0	2	2
c.425G>A	p.S142N	No	0	1	1

c.370A>G	p.I124V	No	1	0	1
c.315G>T	p.K105N	No	0	1	1
c.280T>A	p.Y94N	No	0	1	1
c.262C>T	p.H88Y	No	1	0	1
c.199A>G	p.S67G	No	0	1	1
c.191A>G	p.N64S	No	0	1	1
c.170A>G	p.K57R	No	1	0	1
c.85C>A	p.R29S	Yes	0	1	1
c.80T>C	p.I27T	No	0	4	4
c.38G>A	p.R13Q	No	1	3	4
c.18G>T	p.W6C	Yes	9	13	22
c.8G>A	p.R3Q	No	1	0	1
EME1					
c.15G>T	p.K5N	Yes	2	2	4
c.19T>C	p.S7P	No	1	0	1
c.38G>C	p.G13A	No	3	1	4
c.48C>A	p.D16E	Yes	0	1	1
c.65C>T	p.T22I	Yes	0	2	2
c.119G>A	p.R40K	No	1	1	2
c.124G>A	p.E42K	No	1	0	1
c.124G>C	p.E42Q	No	0	1	1
c.145A>G	p.I49V	No	6	3	9
c.210A>G	p.I70M	No	0	1	1
c.267T>G	p.D89E	No	0	1	1
c.275A>C	p.E92A	No	0	1	1
c.340G>A	p.D114N	No	5	12	17
c.389G>A	p.G130D	No	1	0	1
c.481C>G	p.L161V	No	2	1	3
c.543C>A	p.S181R	No	1	0	1
c.567T>A	p.N189K	No	8	11	19
c.604A>G	p.S202G	No	1	0	1
c.612G>T	p.K204N	Yes	1	0	1
c.782T>C	p.L261S	Yes	1	0	1
c.787A>T	p.M263L	No	0	1	1
c.824C>T	p.T275I	Yes	0	1	1
c.836G>A	p.R279H	No	0	2	2
c.845T>C	p.I282T	Yes	1	0	1
c.950G>A	p.R317Q	No	0	1	1
c.976G>A	p.D326N	No	1	0	1
c.996C>G	p.S332R	No	0	1	1
c.1039G>A	p.V347I	No	1	0	1
c.1061C>T	p.T354I	No	0	2	2

c.1103A>T	p.K368I	Yes	0	1	1
c.1243A>G	p.M415V	No	1	0	1
c.1276G>A	p.V426M	No	0	1	1
c.1297G>C	p.E433Q	No	0	1	1
c.1306G>A	p.A436T	No	8	6	14
c.1341C>G	p.D447E	No	1	0	1
c.1394G>A	p.R465Q	No	2	0	2
c.1435G>C	p.A479P	Yes	9	5	14
c.1474G>A	p.A492T	No	2	0	2
c.1541T>C	p.V514A	No	1	0	1
c.1558C>T	p.P520S	Yes	1	0	1
c.1609C>G	p.Q537E	No	1	0	1
c.1636C>T	p.R546C	Yes	6	2	8
c.1637G>A	p.R546H	No	0	1	1
c.1640G>C	p.R547P	Yes	2	0	2
c.1687T>A	p.S563T	Yes	0	1	1
c.1693C>T	p.R565C	Yes	3	0	3
c.1727A>G	p.H576R	No	0	1	1
FANCL					
c.1091G>A	p.C364Y	Yes	1	0	1
c.1087G>A	p.E363K	No	1	0	1
c.1067G>A	p.S356N	No	0	2	2
c.1066A>G	p.S356G	Yes	1	0	1
c.980A>G	p.Q327R	No	1	0	1
c.978T>A	p.D326E	Yes	1	0	1
c.877T>G	p.L293V	No	1	0	1
c.834G>T	p.L278F	No	2	1	3
c.825C>A	p.N275K	No	2	1	3
c.824A>C	p.N275T	Yes	1	0	1
c.754A>G	p.M252V	No	1	0	1
c.733G>A	p.V245I	No	1	0	1
c.728T>C	p.I243T	Yes	1	0	1
c.722T>A	p.I241K	Yes	1	0	1
c.719C>A	p.S240Y	Yes	1	1	2
c.710A>G	p.N237S	No	0	1	1
c.692G>A	p.R231H	Yes	1	0	1
c.685A>G	p.T229A	No	5	0	5
c.664G>C	p.E222Q	No	1	1	2
c.655C>G	p.L219V	No	0	1	1
c.558C>A	p.S186R	Yes	1	0	1
c.523G>A	p.A175T	No	0	1	1
c.493T>G	p.Y165D	No	0	1	1

c.452C>A	p.T151N	Yes	1	0	1
c.354A>G	p.I118M	No	1	0	1
c.332A>G	p.Y111C	Yes	1	0	1
c.317C>T	p.P106L	Yes	1	0	1
c.252G>A	p.M84I	No	0	1	1
c.203G>C	p.R68P	No	5	3	8
c.191G>T	p.S64I	No	0	1	1
c.112C>T	p.L38F	Yes	26	22	48
c.108C>G	p.F36L	Yes	2	0	2
c.38C>A	p.P13H	Yes	0	1	1
c.19A>G	p.S7G	No	1	0	1
c.2T>C	p.M1T	No	0	1	1
FANCM					
c.43A>G	p.S15G	No	1	0	1
c.53G>A	p.R18Q	No	1	1	2
c.59C>G	p.S20C	No	1	0	1
c.68C>A	p.P23Q	No	0	1	1
c.163G>A	p.D55N	Yes	3	7	10
c.171G>C	p.L57F	No	29	34	63
c.269C>T	p.P90L	Yes	3	4	7
c.356T>A	p.F119Y	Yes	1	0	1
c.488C>T	p.S163F	Yes	0	1	1
c.497C>A	p.A166D	Yes	1	0	1
c.504G>C	p.M168I	Yes	1	0	1
c.527C>T	p.T176I	No	34	32	66
c.538A>G	p.I180V	No	0	3	3
c.545G>C	p.C182S	No	0	1	1
c.547A>C	p.S183R	Yes	4	3	7
c.641A>G	p.D214G	Yes	0	1	1
c.650A>G	p.H217R	Yes	1	0	1
c.710A>G	p.N237S	No	1	0	1
c.752A>G	p.D251G	Yes	0	1	1
c.763G>A	p.V255M	Yes	0	1	1
c.775A>G	p.I259V	No	1	2	3
c.808C>T	p.R270C	Yes	0	2	2
c.874C>T	p.P292S	Yes	0	1	1
c.926A>C	p.E309A	Yes	1	0	1
c.938G>A	p.R313H	No	1	0	1
c.946A>G	p.I316V	No	0	1	1
c.950A>G	p.Q317R	No	1	0	1
c.1017T>A	p.D339E	Yes	0	1	1
c.1072G>A	p.E358K	Yes	0	1	1

c.1087A>G	p.I363V	No	0	3	3
c.1115A>G	p.E372G	Yes	0	1	1
c.1127A>G	p.Q376R	Yes	0	1	1
c.1192C>T	p.R398W	Yes	0	1	1
c.1222G>C	p.D408H	Yes	1	1	2
c.1237T>C	p.Y413H	Yes	8	19	27
c.1264C>T	p.R422C	No	1	1	2
c.1366G>A	p.V456I	No	1	0	1
c.1425_1427delTGA	p.475_476del	No	0	1	1
c.1438A>T	p.M480L	Yes	0	1	1
c.1439T>G	p.M480R	Yes	0	1	1
c.1440G>A	p.M480I	Yes	0	1	1
c.1457G>A	p.R486Q	Yes	0	1	1
c.1462A>G	p.S488G	Yes	1	0	1
c.1576C>G	p.L526V	No	13	25	38
c.1597C>T	p.R533C	Yes	2	4	6
c.1598G>A	p.R533H	Yes	0	1	1
c.1603G>A	p.G535S	Yes	1	0	1
c.1604G>A	p.G535D	Yes	2	0	2
c.1627A>G	p.T543A	Yes	1	0	1
c.1636G>A	p.G546S	Yes	2	0	2
c.1651G>T	p.D551Y	Yes	0	1	1
c.1660G>A	p.E554K	Yes	0	1	1
c.1667A>G	p.D556G	Yes	1	1	2
c.1703T>C	p.I568T	No	0	1	1
c.1729A>T	p.T577S	Yes	0	1	1
c.1741C>T	p.R581C	Yes	1	1	2
c.1742G>A	p.R581H	Yes	0	1	1
c.1756G>C	p.V586L	Yes	0	1	1
c.1760T>C	p.I587T	Yes	2	0	2
c.1775G>A	p.G592E	Yes	1	0	1
c.1814A>G	p.K605R	No	0	1	1
c.1846A>G	p.R616G	No	0	1	1
c.1849C>G	p.Q617E	No	0	3	3
c.1861T>C	p.F621L	No	0	1	1
c.1880G>A	p.R627Q	Yes	1	0	1
c.1880G>C	p.R627P	Yes	0	1	1
c.1918A>G	p.M640V	Yes	0	1	1
c.2084G>C	p.R695T	No	0	1	1
c.2182A>T	p.I728F	No	0	1	1
c.2190A>T	p.Q730H	Yes	1	0	1
c.2215T>C	p.W739R	Yes	1	0	1

c.2236A>G	p.T746A	Yes	1	0	1
c.2261G>A	p.R754Q	Yes	0	1	1
c.2267G>A	p.R756H	No	0	3	3
c.2302A>G	p.M768V	Yes	0	1	1
c.2330A>G	p.Y777C	Yes	0	1	1
c.2372C>A	p.T791N	No	0	1	1
c.2458A>G	p.N820D	No	0	1	1
c.2483C>T	p.S828L	No	0	1	1
c.2497G>A	p.D833N	No	0	1	1
c.2501A>G	p.E834G	Yes	0	1	1
c.2575A>G	p.K859E	No	0	1	1
c.2629T>C	p.S877P	No	1	0	1
c.2638A>G	p.N880D	No	0	1	1
c.2704G>A	p.D902N	No	0	1	1
c.2716A>T	p.I906F	No	1	1	2
c.2741A>G	p.E914G	No	0	1	1
c.2749A>G	p.I917V	No	3	1	4
c.2752A>G	p.K918E	No	1	0	1
c.2759C>T	p.P920L	No	0	2	2
c.2764G>A	p.V922M	No	0	1	1
c.2813C>T	p.A938V	No	0	1	1
c.2831C>T	p.S944F	Yes	0	1	1
c.2859A>C	p.K953N	No	15	24	39
c.2888T>C	p.F963S	No	0	1	1
c.2986T>C	p.Y996H	No	0	1	1
c.3016G>A	p.D1006N	No	1	0	1
c.3049C>T	p.L1017F	No	0	1	1
c.3062T>C	p.L1021P	No	1	0	1
c.3064T>C	p.F1022L	No	0	1	1
c.3113T>C	p.L1038P	No	1	0	1
c.3147A>C	p.L1049F	No	1	0	1
c.3175T>C	p.Y1059H	No	1	0	1
c.3293A>G	p.N1098S	No	1	0	1
c.3296G>A	p.R1099H	No	5	2	7
c.3350A>T	p.D1117V	Yes	1	0	1
c.3371C>A	p.P1124Q	No	1	0	1
c.3414A>T	p.E1138D	No	0	1	1
c.3436G>A	p.D1146N	No	0	2	2
c.3469G>A	p.E1157K	No	0	1	1
c.3557A>G	p.N1186S	No	1	0	1
c.3613G>A	p.D1205N	No	0	1	1
c.3658A>T	p.I1220F	No	1	0	1

c.3704G>T	p.G1235V	Yes	2	1	3
c.3731A>G	p.E1244G	No	1	1	2
c.3785G>A	p.R1262K	No	1	0	1
c.3827C>T	p.S1276L	No	0	2	2
c.3845C>A	p.P1282Q	No	1	0	1
c.3863A>G	p.N1288S	No	2	3	5
c.3869C>T	p.T1290I	No	0	1	1
c.3911A>G	p.N1304S	No	0	1	1
c.3935T>C	p.L1312P	No	0	1	1
c.3956A>G	p.E1319G	No	0	1	1
c.3959T>C	p.L1320S	No	1	0	1
c.3991C>T	p.P1331S	Yes	0	1	1
c.3992C>T	p.P1331L	No	0	3	3
c.3998A>C	p.Q1333P	No	2	1	3
c.4022T>C	p.L1341P	No	0	1	1
c.4084G>A	p.D1362N	No	0	1	1
c.4101A>C	p.K1367N	No	0	1	1
c.4147T>G	p.Y1383D	No	1	0	1
c.4216G>A	p.V1406I	No	1	0	1
c.4306A>G	p.N1436D	No	0	1	1
c.4366C>T	p.R1456C	No	3	3	6
c.4367G>A	p.R1456H	No	0	2	2
c.4465G>A	p.G1489R	No	1	1	2
c.4480G>T	p.G1494C	Yes	1	0	1
c.4489G>A	p.V1497I	No	0	1	1
c.4523C>G	p.A1508G	No	2	0	2
c.4556C>A	p.S1519Y	Yes	0	1	1
c.4606A>G	p.N1536D	No	1	0	1
c.4607A>G	p.N1536S	No	0	1	1
c.4627C>G	p.L1543V	No	1	0	1
c.4627C>T	p.L1543F	No	1	2	3
c.4709G>A	p.R1570H	No	1	0	1
c.4859A>G	p.E1620G	Yes	0	1	1
c.4872T>G	p.C1624W	Yes	1	0	1
c.4912A>G	p.S1638G	No	1	1	2
c.4933C>T	p.R1645C	Yes	1	0	1
c.4934G>A	p.R1645H	No	0	1	1
c.4946T>G	p.L1649R	Yes	1	0	1
c.5107C>G	p.H1703D	No	1	2	3
c.5108A>G	p.H1703R	No	1	3	4
c.5117A>C	p.N1706T	No	0	1	1
c.5144A>C	p.Q1715P	No	0	1	1

c.5155C>T	p.R1719C	No	0	1	1
c.5177C>T	p.P1726L	No	0	2	2
c.5183C>G	p.A1728G	No	0	1	1
c.5249C>T	p.P1750L	No	1	0	1
c.5263G>A	p.E1755K	No	0	1	1
c.5291C>T	p.T1764I	No	1	0	1
c.5309A>G	p.K1770R	No	0	1	1
c.5337G>T	p.Q1779H	No	0	1	1
c.5413A>G	p.T1805A	No	0	1	1
c.5440G>A	p.E1814K	No	1	0	1
c.5474A>G	p.H1825R	No	0	1	1
c.5545C>A	p.P1849T	Yes	1	0	1
c.5569G>A	p.V1857M	Yes	9	7	16
c.5578C>T	p.R1860C	Yes	0	1	1
c.5579G>A	p.R1860H	Yes	0	1	1
c.5656C>T	p.H1886Y	No	3	3	6
c.5679A>T	p.R1893S	Yes	0	1	1
c.5759G>C	p.S1920T	No	0	1	1
c.5770A>T	p.T1924S	No	0	2	2
c.5777T>C	p.I1926T	Yes	1	0	1
c.5782G>A	p.A1928T	No	0	1	1
c.5792G>A	p.R1931Q	No	1	0	1
c.5832G>T	p.L1944F	Yes	2	0	2
c.5866A>G	p.N1956D	Yes	1	0	1
c.5951A>T	p.Y1984F	Yes	0	2	2
c.6097C>A	p.Q2033K	No	0	1	1
c.6128G>A	p.R2043K	No	0	1	1
c.6143T>C	p.I2048T	No	1	2	3
PARP2					
c.26C>A	p.T9N	No	1	1	2
c.41C>G	p.A14G	No	1	0	1
c.43A>G	p.R15G	No	5	9	14
c.67G>A	p.V23I	No	0	1	1
c.148A>G	p.M50V	No	0	1	1
c.197A>G	p.Q66R	No	2	0	2
c.440T>G	p.V147G	Yes	1	0	1
c.449G>A	p.R150K	Yes	0	1	1
c.454G>A	p.G152S	Yes	0	1	1
c.464G>A	p.G155E	Yes	1	0	1
c.475C>A	p.Q159K	Yes	1	0	1
c.482G>A	p.S161N	No	4	1	5
c.487G>A	p.V163M	No	0	1	1

c.503A>G	p.N168S	No	0	1	1
c.509A>G	p.N170S	No	0	1	1
c.540C>G	p.F180L	Yes	0	2	2
c.551C>T	p.T184M	Yes	1	0	1
c.610C>G	p.L204V	No	2	0	2
c.652A>G	p.T218A	No	1	0	1
c.785C>A	p.T262N	Yes	0	1	1
c.799C>T	p.L267F	Yes	1	0	1
c.859G>A	p.D287N	No	0	1	1
c.868C>T	p.R290W	Yes	1	1	2
c.880C>T	p.H294Y	No	1	1	2
c.895A>C	p.M299L	No	0	1	1
c.947G>A	p.R316H	Yes	1	1	2
c.950C>T	p.T317I	No	1	0	1
c.965G>A	p.R322Q	No	4	6	10
c.1031T>C	p.I344T	Yes	0	1	1
c.1043A>G	p.K348R	No	0	1	1
c.1069C>G	p.P357A	Yes	0	1	1
c.1072T>A	p.L358M	No	0	1	1
c.1081C>T	p.H361Y	Yes	0	2	2
c.1084T>G	p.Y362D	Yes	0	1	1
c.1108C>T	p.R370C	Yes	1	1	2
c.1109G>A	p.R370H	No	0	1	1
c.1229A>G	p.D410G	No	1	0	1
c.1234G>A	p.E412K	Yes	3	2	5
c.1331G>A	p.R444Q	Yes	1	1	2
c.1398G>T	p.M466I	Yes	0	1	1
c.1399T>C	p.S467P	Yes	1	0	1
c.1442A>G	p.N481S	No	0	2	2
c.1445C>T	p.T482I	No	1	0	1
c.1468G>A	p.V490I	Yes	0	1	1
c.1600A>G	p.S534G	Yes	0	1	1
c.1638T>G	p.I546M	No	1	0	1
c.1655A>G	p.Y552C	No	0	1	1
c.1670A>G	p.N557S	Yes	1	0	1
c.1691C>G	p.P564R	Yes	1	0	1
c.1708C>T	p.R570W	Yes	0	1	1
PARP3					
c.51G>T	p.Q17H	No	0	1	1
c.53C>A	p.T18N	No	0	3	3
c.56A>G	p.E19G	No	0	8	8
c.57G>C	p.E19D	No	0	4	4

c.58G>C	p.G20R	No	2	6	8
c.59G>C	p.G20A	No	3	10	13
c.69G>C	p.K23N	No	2	0	2
c.79C>T	p.R27W	No	1	2	3
c.83A>C	p.Q28P	No	0	1	1
c.92G>C	p.R31T	No	1	0	1
c.110G>A	p.R37H	No	1	0	1
c.119C>G	p.A40G	No	1	0	1
c.142G>T	p.A48S	No	2	1	3
c.147G>T	p.E49D	No	1	0	1
c.151C>A	p.R51S	No	1	0	1
c.161G>T	p.R54L	No	1	0	1
c.227C>T	p.T76I	No	1	0	1
c.244A>T	p.I82F	Yes	0	1	1
c.260A>G	p.N87S	Yes	0	1	1
c.293G>A	p.S98N	No	1	1	2
c.319C>T	p.H107Y	No	1	0	1
c.322T>C	p.W108R	Yes	0	1	1
c.325G>A	p.G109S	Yes	1	0	1
c.379G>C	p.D127H	No	0	1	1
c.401A>T	p.K134M	Yes	0	1	1
c.409C>T	p.R137W	No	1	3	4
c.410G>A	p.R137Q	No	0	1	1
c.439C>T	p.R147W	Yes	4	13	17
c.517G>A	p.V173M	No	0	1	1
c.556C>T	p.R186W	No	2	0	2
c.637A>G	p.M213V	Yes	0	1	1
c.652C>G	p.L218V	No	1	0	1
c.682C>A	p.L228M	No	0	1	1
c.689A>G	p.K230R	No	1	0	1
c.745G>A	p.G249S	No	1	0	1
c.752C>T	p.T251M	No	0	1	1
c.757G>A	p.G253S	No	1	0	1
c.789C>G	p.H263Q	No	0	1	1
c.799G>A	p.V267I	No	2	0	2
c.827A>G	p.Q276R	No	5	2	7
c.898G>C	p.E300Q	No	0	1	1
c.941C>T	p.T314M	No	0	1	1
c.946G>C	p.E316Q	No	0	1	1
c.1103T>C	p.V368A	No	2	2	4
c.1138C>T	p.H380Y	Yes	0	1	1
c.1174G>A	p.G392S	Yes	4	0	4

c.1189G>A	p.V397M	No	1	0	1
c.1198G>A	p.A400T	Yes	0	1	1
c.1204C>T	p.L402F	Yes	1	0	1
c.1219C>T	p.R407C	Yes	1	0	1
c.1220G>A	p.R407H	Yes	1	0	1
c.1243C>T	p.R415C	Yes	1	0	1
c.1244G>A	p.R415H	Yes	0	1	1
c.1316G>T	p.G439V	No	1	0	1
c.1324C>G	p.H442D	No	3	2	5
c.1430T>C	p.I477T	No	1	0	1
c.1436G>A	p.R479Q	No	1	0	1
c.1447G>A	p.E483K	Yes	1	0	1
c.1505C>A	p.P502H	Yes	1	0	1
c.1561G>A	p.E521K	Yes	1	0	1
c.1592G>A	p.R531H	No	1	0	1
c.1597C>T	p.R533C	Yes	1	0	1
POLN					
c.2684C>T	p.S895L	No	1	0	1
c.2672C>T	p.P891L	No	2	0	2
c.2671C>T	p.P891S	No	1	0	1
c.2648C>A	p.S883Y	No	0	1	1
c.2626A>T	p.N876Y	No	0	3	3
c.2609G>A	p.R870H	No	0	1	1
c.2609G>T	p.R870L	No	1	1	2
c.2590C>T	p.P864S	No	1	0	1
c.2548C>T	p.R850C	Yes	0	2	2
c.2435C>A	p.P812Q	No	0	1	1
c.2435C>T	p.P812L	No	0	1	1
c.2425G>A	p.V809M	No	1	0	1
c.2410G>A	p.E804K	Yes	0	1	1
c.2381C>T	p.T794M	Yes	0	1	1
c.2231C>A	p.P744H	Yes	1	0	1
c.2230C>A	p.P744T	No	1	1	2
c.2219G>C	p.G740A	Yes	1	0	1
c.2218G>A	p.G740S	Yes	0	1	1
c.2201G>A	p.C734Y	No	1	0	1
c.2198G>T	p.G733V	Yes	1	1	2
c.2194A>G	p.T732A	No	0	2	2
c.2168G>A	p.R723Q	No	7	2	9
c.2153T>C	p.I718T	Yes	0	1	1
c.2133T>G	p.F711L	Yes	2	0	2
c.2075G>A	p.R692Q	No	1	2	3

c.2057A>G	p.Y686C	Yes	0	1	1
c.2048C>T	p.A683V	Yes	0	1	1
c.2041G>A	p.V681M	No	3	2	5
c.2026C>A	p.Q676K	No	0	1	1
c.1996G>A	p.V666M	No	1	0	1
c.1939G>A	p.E647K	No	1	0	1
c.1916C>A	p.P639Q	Yes	1	0	1
c.1886T>C	p.L629S	Yes	0	1	1
c.1862C>T	p.A621V	Yes	1	0	1
c.1852A>G	p.T618A	No	6	5	11
c.1803G>C	p.K601N	No	0	1	1
c.1754A>G	p.H585R	Yes	1	0	1
c.1730C>T	p.P577L	Yes	0	1	1
c.1729C>A	p.P577T	Yes	0	1	1
c.1615C>T	p.H539Y	No	1	0	1
c.1547A>G	p.E516G	No	0	1	1
c.1520G>A	p.G507E	No	1	0	1
c.1520G>C	p.G507A	No	0	1	1
c.1517C>T	p.T506M	Yes	1	1	2
c.1454G>T	p.R485L	Yes	0	1	1
c.1436C>T	p.T479M	No	8	2	10
c.1403C>T	p.A468V	No	0	1	1
c.1399G>A	p.E467K	Yes	1	0	1
c.1394A>G	p.E465G	Yes	0	1	1
c.1379G>A	p.R460H	No	0	1	1
c.1378C>T	p.R460C	Yes	0	2	2
c.1348A>G	p.M450V	No	1	0	1
c.1264C>A	p.Q422K	No	0	1	1
c.1184A>G	p.Q395R	No	1	1	2
c.1159A>C	p.N387H	No	1	0	1
c.1075A>G	p.S359G	No	1	0	1
c.1057G>C	p.A353P	Yes	1	0	1
c.937A>G	p.K313E	No	0	1	1
c.928A>G	p.M310V	No	0	1	1
c.928A>C	p.M310L	No	51	39	90
c.913G>A	p.V305M	No	1	1	2
c.907C>T	p.R303W	No	0	1	1
c.826T>C	p.S276P	No	0	1	1
c.793C>T	p.P265S	No	22	24	46
c.734G>A	p.R245K	No	0	1	1
c.677T>C	p.M226T	No	0	1	1
c.656C>T	p.A219V	No	0	1	1

c.575C>T	p.A192V	No	1	0	1
c.572G>A	p.G191E	No	1	0	1
c.548G>T	p.G183V	No	4	0	4
c.488A>C	p.N163T	No	1	0	1
c.470A>G	p.K157R	No	1	0	1
c.452G>A	p.S151N	No	2	0	2
c.370C>G	p.Q124E	No	1	1	2
c.353T>C	p.L118S	No	1	1	2
c.319A>G	p.K107E	No	0	2	2
c.290C>T	p.T97I	No	1	0	1
c.254C>T	p.A85V	No	0	1	1
c.227T>C	p.L76S	No	0	1	1
c.167A>G	p.Y56C	No	0	1	1
c.151A>G	p.K51E	No	0	1	1
c.149A>C	p.N50T	No	0	1	1
c.83T>C	p.M28T	No	1	0	1
c.47C>T	p.P16L	Yes	1	1	2
RAD54L					
c.65A>T	p.D22V	No	1	0	1
c.95C>G	p.P32R	No	0	1	1
c.145T>C	p.S49P	No	0	1	1
c.160C>T	p.P54S	No	1	0	1
c.223C>G	p.R75G	No	0	1	1
c.227G>C	p.S76T	No	1	0	1
c.259C>G	p.P87A	No	1	0	1
c.367C>T	p.P123S	Yes	0	1	1
c.374C>T	p.P125L	No	3	0	3
c.473G>C	p.R158T	Yes	0	3	3
c.515G>A	p.R172H	No	0	1	1
c.569C>T	p.T190M	Yes	1	1	2
c.604C>T	p.R202C	Yes	24	17	41
c.620G>A	p.C207Y	Yes	0	1	1
c.785G>T	p.R262L	No	2	1	3
c.796G>C	p.V266L	No	0	1	1
c.883T>C	p.C295R	Yes	1	0	1
c.901C>T	p.L301F	Yes	0	1	1
c.955C>T	p.R319W	Yes	0	1	1
c.956G>A	p.R319Q	Yes	0	1	1
c.1021T>C	p.F341L	Yes	0	1	1
c.1094G>A	p.R365Q	No	0	1	1
c.1138C>T	p.R380W	Yes	1	1	2
c.1298C>T	p.P433L	No	3	0	3

c.1316A>G	p.E439G	No	0	1	1
c.1351A>G	p.T451A	Yes	0	1	1
c.1389C>G	p.I463M	Yes	1	1	2
c.1492A>G	p.M498V	No	1	0	1
c.1517C>T	p.A506V	Yes	0	1	1
c.1532G>A	p.R511H	No	0	1	1
c.1542C>A	p.D514E	Yes	0	1	1
c.1592A>C	p.K531T	Yes	1	0	1
c.1598G>A	p.C533Y	Yes	1	0	1
c.1600C>T	p.R534C	Yes	0	1	1
c.1601G>C	p.R534P	Yes	0	1	1
c.1637C>T	p.T546M	Yes	2	0	2
c.1655G>A	p.R552Q	Yes	2	2	4
c.1679A>G	p.N560S	Yes	0	1	1
c.1748T>C	p.I583T	Yes	1	0	1
c.1759C>T	p.R587W	Yes	4	8	12
c.1849T>A	p.Y617N	Yes	0	1	1
c.1850A>G	p.Y617C	Yes	1	0	1
c.1858C>T	p.R620C	Yes	1	0	1
c.1934T>G	p.V645G	Yes	1	0	1
c.1970C>G	p.S657C	Yes	3	2	5
c.1996A>G	p.I666V	No	1	0	1
c.2063G>A	p.R688H	No	1	0	1
c.2072G>A	p.R691Q	No	4	1	5
c.2080C>G	p.P694A	No	1	0	1
c.2080C>T	p.P694S	No	1	0	1
c.2126G>A	p.C709Y	No	1	0	1
c.2129C>G	p.T710S	No	1	0	1
c.2138G>T	p.W713L	No	1	0	1
c.2213G>A	p.R738H	No	33	32	65
SMUG1					
c.770A>G	p.E257G	No	1	0	1
c.700G>A	p.V234M	Yes	0	1	1
c.698A>G	p.Q233R	No	0	1	1
c.667C>T	p.R223W	Yes	0	1	1
c.659G>A	p.R220Q	Yes	1	0	1
c.622C>T	p.R208W	Yes	1	0	1
c.605T>G	p.V202G	Yes	0	1	1
c.547C>T	p.P183S	Yes	0	1	1
c.527A>G	p.N176S	Yes	0	1	1
c.523C>T	p.R175C	Yes	1	0	1
c.466T>C	p.F156L	Yes	1	0	1

c.313C>T	p.R105W	Yes	5	15	20
c.256C>G	p.P86A	Yes	1	0	1
c.218G>A	p.C73Y	Yes	0	1	1
c.212G>A	p.R71H	Yes	0	1	1
c.169C>A	p.P57T	Yes	1	1	2
c.143C>T	p.S48L	Yes	3	5	8
c.106C>T	p.L36F	No	1	0	1
c.67T>A	p.C23S	No	0	1	1
c.44G>T	p.G15V	No	3	1	4
c.2T>C	p.M1T	No	0	1	1

*Predicted damaging by 2 out of three *in silico* software programs: SIFT, Polyphen-2 and Provean

