Case No.	Locus	Genotype	Ref	Type	Gene	Location	Exon	Coding	Protein	Variant Effect	ClinVar	COSMIC PREDICTION	Coverage	Allele 1 Type	Allele 1 Coverage	Allele 2 Type	Allele 2 Coverage	% Mutated Allele 2	Allele Ratio	p-value
1	**chr11:108175462**	**G/A**	**G**	**SNV**	**ATM**	**exonic**	**37**	**c.5557G>A**	**p.Asp1853Asn**	**missense**		**Pathogenic**	**465**	**G**	**254**	**A**	**211**	**45,4**	**G=0.5462, A=0.4538**	**0.00001**
1	**chr16:89985940**	**G/A**	**G**	**SNV**	**MC1R**	**exonic**	**1**	**c.274G>A**	**p.Val92Met**	**missense**	**pathogenic**		**324**	**G**	**186**	**A**	**138**	**42,6**	**G=0.5741, A=0.4259**	**0.00001**
1	chr16:89986154	G/A	G	SNV	MC1R	exonic	1	c.488G>A	p.Arg163Gln	missense			271	G	149	A	122	45,0	G=0.5498, A=0.4502	0.00001
3	**chr11:108143456**	**C/G**	**C**	**SNV**	**ATM**	**exonic**	**22**	**c.3161C>G**	**p.Pro1054Arg**	**missense**		**Pathogenic**	**269**	**C**	**141**	**G**	**128**	**47,6**	**C=0.5242, G=0.4758**	**0.00001**
3	**chr16:89985844**	**G/T**	**G**	**SNV**	**MC1R**	**exonic**	**1**	**c.178G>T**	**p.Val60Leu**	**missense**	**pathogenic**		**189**	**G**	**101**	**T**	**88**	**46,6**	**G=0.5344, T=0.4656**	**0.00001**
4	chr16:23635348	A/C	A	SNV	PALB2	exonic	8	c.2816T>G	p.Leu939Trp	missense	probable-non-pathogenic		242	A	127	C	115	47,5	A=0.5248, C=0.4752	0.00001
4	chr16:23646857	A/G	A	SNV	PALB2	exonic	4	c.1010T>C	p.Leu337Ser	missense	probable-non-pathogenic		174	A	92	G	82	47,1	A=0.5287, G=0.4713	0.00001
7	**chr11:108175463**	**A/T**	**A**	**SNV**	**ATM**	**exonic**	**37**	**c.5558A>T**	**p.Asp1853Val**	**missense**		**Pathogenic**	**628**	**A**	**299**	**T**	**329**	**52,4**	**A=0.4761, T=0.5239**	**0.00001**
9	**chr16:89985918**	**C/A**	**C**	**SNV**	**MC1R**	**exonic**	**1**	**c.252C>A**	**p.Asp84Glu**	**missense**	**other,pathogenic**		**292**	**C**	**150**	**A**	**142**	**48,6**	**C=0.5137, A=0.4863**	**0.00001**
9	**chr16:89986144**	**C/T**	**C**	**SNV**	**MC1R**	**exonic**	**1**	**c.478C>T**	**p.Arg160Trp**	**missense**	**pathogenic**		**306**	**C**	**153**	**T**	**153**	**50,0**	**C=0.5, T=0.5**	**0.00001**
9	chr5:33951693	C/G	C	SNV	SLC45A2	exonic	5	c.1122G>C	p.Leu374Phe	missense			147	C	87	G	60	40,8	C=0.5918, G=0.4082	0.00001
10	chr3:70014091	G/A	G	SNV	MITF	exonic	10	c.1255G>A	p.Glu419Lys	missense			478	G	231	A	247	51,7	G=0.4833, A=0.5167	0.00001
10	**chr11:89017961**	**G/A**	**G**	**SNV**	**TYR**	**exonic**	**4**	**c.1205G>A**	**p.Arg402Gln**	**missense**	**other,pathogenic**		**130**	**G**	**65**	**A**	**65**	**50,0**	**G=0.5, A=0.5**	**0.00001**
11	**chr9:21974760**	**C/T**	**C**	**SNV**	**CDKN2A**	**exonic**	**1**	**c.67G>A**	**p.Gly23Ser**	**missense**		**Pathogenic**	**122**	**C**	**64**	**T**	**58**	**47,5**	**C=0.5246, T=0.4754**	**0.00001**
11	**chr16:89985940**	**G/A**	**G**	**SNV**	**MC1R**	**exonic**	**1**	**c.274G>A**	**p.Val92Met**	**missense**	**pathogenic**		**426**	**G**	**251**	**A**	**175**	**41,1**	**G=0.5892, A=0.4108**	**0.00001**
12	**chr11:108175462**	**G/A**	**G**	**SNV**	**ATM**	**exonic**	**37**	**c.5557G>A**	**p.Asp1853Asn**	**missense**		**Pathogenic**	**653**	**G**	**313**	**A**	**340**	**52,1**	**G=0.4793, A=0.5207**	**0.00001**
13	**chr11:108175462**	**G/A**	**G**	**SNV**	**ATM**	**exonic**	**37**	**c.5557G>A**	**p.Asp1853Asn**	**missense**		**Pathogenic**	**411**	**G**	**214**	**A**	**197**	**47,9**	**G=0.5207, A=0.4793**	**0.00001**
13	**chr16:89986117**	**C/T**	**C**	**SNV**	**MC1R**	**exonic**	**1**	**c.451C>T**	**p.Arg151Cys**	**missense**	**other,pathogenic**		**365**	**C**	**173**	**T**	**192**	**52,6**	**C=0.474, T=0.526**	**0.00001**
15	**chr11:108175463**	**A/T**	**A**	**SNV**	**ATM**	**exonic**	**37**	**c.5558A>T**	**p.Asp1853Val**	**missense**		**Pathogenic**	**400**	**A**	**197**	**T**	**203**	**50,8**	**A=0.4925, T=0.5075**	**0.00001**
15	**chr16:89986546**	**G/C**	**G**	**SNV**	**MC1R**	**exonic**	**1**	**c.880G>C**	**p.Asp294His**	**missense**	**pathogenic**		**1860**	**G**	**903**	**C**	**957**	**51,5**	**G=0.4855, C=0.5145**	**0.00001**
16	**chr16:89985844**	**G/T**	**G**	**SNV**	**MC1R**	**exonic**	**1**	**c.178G>T**	**p.Val60Leu**	**missense**	**pathogenic**		**181**	**G**	**87**	**T**	**94**	**51,9**	**G=0.4807, T=0.5193**	**0.00001**
16	**chr11:89017961**	**G/A**	**G**	**SNV**	**TYR**	**exonic**	**4**	**c.1205G>A**	**p.Arg402Gln**	**missense**	**other,pathogenic**		**159**	**G**	**72**	**A**	**87**	**54,7**	**G=0.4528, A=0.5472**	**0.00001**
20	chr16:23646191	T/C	T	SNV	PALB2	exonic	4	c.1676A>G	p.Gln559Arg	missense	probable-non-pathogenic		205	T	100	C	105	51,2	T=0.4878, C=0.5122	0.00001
21	**chr11:108175462**	**G/A**	**G**	**SNV**	**ATM**	**exonic**	**37**	**c.5557G>A**	**p.Asp1853Asn**	**missense**		**Pathogenic**	**444**	**G**	**216**	**A**	**228**	**51,4**	**G=0.4865, A=0.5135**	**0.00001**
21	**chr9:21974760**	**C/T**	**C**	**SNV**	**CDKN2A**	**exonic**	**1**	**c.67G>A**	**p.Gly23Ser**	**missense**		**Pathogenic**	**238**	**C**	**166**	**T**	**72**	**30,3**	**C=0.6975, T=0.3025**	**0.00001**
23	**chr9:21974756**	**C/G**	**C**	**SNV**	**CDKN2A**	**exonic**	**1**	**c.71G>C**	**p.Arg24Pro**	**missense**	**other**	**Pathogenic**	**129**	**C**	**81**	**G**	**48**	**37,2**	**C=0.6279, G=0.3721**	**0.00001**
23	**chr16:89986117**	**C/T**	**C**	**SNV**	**MC1R**	**exonic**	**1**	**c.451C>T**	**p.Arg151Cys**	**missense**	**other,pathogenic**		**297**	**C**	**145**	**T**	**152**	**51,2**	**C=0.4882, T=0.5118**	**0.00001**
23	**chr16:89985940**	**G/A**	**G**	**SNV**	**MC1R**	**exonic**	**1**	**c.274G>A**	**p.Val92Met**	**missense**	**pathogenic**		**252**	**G**	**156**	**A**	**96**	**38,1**	**G=0.619, A=0.381**	**0.00001**
23	chr16:23641461	C/G	C	SNV	PALB2	exonic	5	c.2014G>C	p.Glu672Gln	missense	probable-non-pathogenic		116	C	56	G	60	51,7	C=0.4828, G=0.5172	0.00001
23	chr7:124469334	G/A	G	SNV	POT1	exonic	16	c.1568C>T	p.Ser523Leu	missense			237	G	114	A	123	51,9	G=0.481, A=0.519	0.00001
25	**chr16:89985844**	**G/T**	**G**	**SNV**	**MC1R**	**exonic**	**1**	**c.178G>T**	**p.Val60Leu**	**missense**	**pathogenic**		**119**	**G**	**59**	**T**	**60**	**50,4**	**G=0.4958, T=0.5042**	**0.00001**
27	**chr11:108175462**	**G/A**	**G**	**SNV**	**ATM**	**exonic**	**37**	**c.5557G>A**	**p.Asp1853Asn**	**missense**		**Pathogenic**	**681**	**G**	**351**	**A**	**330**	**48,5**	**G=0.5154, A=0.4846**	**0.00001**
28	chr7:124475415	G/T	G	SNV	POT1	exonic	15	c.1423C>A	p.Pro475Thr	missense			322	G	154	T	168	52,2	G=0.4783, T=0.5217	0.00001
32	**chr11:108175462**	**G/A**	**G**	**SNV**	**ATM**	**exonic**	**37**	**c.5557G>A**	**p.Asp1853Asn**	**missense**		**Pathogenic**	**184**	**G**	**97**	**A**	**87**	**47,3**	**G=0.5272, A=0.4728**	**0.00001**
32	chr9:21970916	C/T	C	SNV	CDKN2A	exonic	2	c.442G>A	p.Ala148Thr	missense		Neutral	130	C	67	T	63	48,5	C=0.5154, T=0.4846	0.00001
33	**chr16:89985940**	**G/A**	**G**	**SNV**	**MC1R**	**exonic**	**1**	**c.274G>A**	**p.Val92Met**	**missense**	**pathogenic**		**251**	**G**	**155**	**A**	**96**	**38,2**	**G=0.6175, A=0.3825**	**0.00001**
33	chr16:23635370	C/T	C	SNV	PALB2	exonic	8	c.2794G>A	p.Val932Met	missense	probable-non-pathogenic		105	C	55	T	50	47,6	C=0.5238, T=0.4762	0.00001
34	**chr11:108175462**	**G/A**	**G**	**SNV**	**ATM**	**exonic**	**37**	**c.5557G>A**	**p.Asp1853Asn**	**missense**		**Pathogenic**	**544**	**G**	**284**	**A**	**260**	**47,8**	**G=0.5221, A=0.4779**	**0.00001**
34	**chr9:21974756**	**C/G**	**C**	**SNV**	**CDKN2A**	**exonic**	**1**	**c.71G>C**	**p.Arg24Pro**	**missense**	**other**	**Pathogenic**	**133**	**C**	**65**	**G**	**68**	**51,1**	**C=0.4887, G=0.5113**	**0.00001**
34	**chr16:89986117**	**C/T**	**C**	**SNV**	**MC1R**	**exonic**	**1**	**c.451C>T**	**p.Arg151Cys**	**missense**	**other,pathogenic**		**710**	**C**	**354**	**T**	**356**	**50,1**	**C=0.4986, T=0.5014**	**0.00001**
34	chr16:89986154	G/A	G	SNV	MC1R	exonic	1	c.488G>A	p.Arg163Gln	missense			404	G	203	A	201	49,8	G=0.5025, A=0.4975	0.00001
34	chr16:75690279	A/G	A	SNV	TERF2IP	exonic	3	c.970A>G	p.Lys324Glu	missense			185	A	97	G	88	47,6	A=0.5243, G=0.4757	0.00001
35	chr5:33951693	C/G	C	SNV	SLC45A2	exonic	5	c.1122G>C	p.Leu374Phe	missense			134	C	69	G	65	48,5	C=0.5149, G=0.4851	0.00001
36	chr11:108178702	G/C	G	SNV	ATM	exonic	38	c.5753G>C	p.Arg1918Thr	missense			184	G	94	C	90	48,9	G=0.5109, C=0.4891	0.00001
36	**chr11:89017973**	**C/T**	**C**	**SNV**	**TYR**	**exonic**	**4**	**c.1217C>T**	**p.Pro406Leu**	**missense**	**pathogenic**		**126**	**C**	**79**	**T**	**47**	**37,3**	**C=0.627, T=0.373**	**0.00001**
38	**chr16:89985844**	**G/T**	**G**	**SNV**	**MC1R**	**exonic**	**1**	**c.178G>T**	**p.Val60Leu**	**missense**	**pathogenic**		**550**	**G**	**300**	**T**	**250**	**45,5**	**G=0.5455, T=0.4545**	**0.00001**
38	chr7:124532326	C/T	C	SNV	POT1	exonic	6	c.118G>A	p.Gly40Arg	missense			691	C	586	T	105	15,2	C=0.848, T=0.152	0.00059
45	**chr9:21974721**	**C/T**	**C**	**SNV**	**CDKN2A**	**exonic**	**1**	**c.106G>A**	**p.Ala36Thr**	**missense**		**Pathogenic**	**146**	**C**	**74**	**T**	**72**	**49,3**	**C=0.5068, T=0.4932**	**0.00001**
45	**chr16:89986117**	**C/T**	**C**	**SNV**	**MC1R**	**exonic**	**1**	**c.451C>T**	**p.Arg151Cys**	**missense**	**other,pathogenic**		**111**	**C**	**61**	**T**	**50**	**45,0**	**C=0.5495, T=0.4505**	**0.00001**
47	chr7:124482969	A/C	A	SNV	POT1	exonic	13	c.1055T>G	p.Leu352Trp	missense			412	A	325	C	87	21,1	A=0.7888, C=0.2112	0.00001
49	**chr16:89985844**	**T/T**	**G**	**SNV**	**MC1R**	**exonic**	**1**	**c.178G>T**	**p.Val60Leu**	**missense**	**pathogenic**		**964**	**G**	**28**	**T**	**936**	**97,1**	**G=0.029, T=0.971**	**0.00001**
49	**chr11:89017961**	**G/A**	**G**	**SNV**	**TYR**	**exonic**	**4**	**c.1205G>A**	**p.Arg402Gln**	**missense**	**other,pathogenic**		**269**	**G**	**131**	**A**	**138**	**51,3**	**G=0.487, A=0.513**	**0.00001**
52	**chr16:89986546**	**G/C**	**G**	**SNV**	**MC1R**	**exonic**	**1**	**c.880G>C**	**p.Asp294His**	**missense**	**pathogenic**		**726**	**G**	**395**	**C**	**331**	**45,6**	**G=0.5441, C=0.4559**	**0.00001**
52	**chr16:23641041**	**G/A**	**G**	**SNV**	**PALB2**	**exonic**	**5**	**c.2434C>T**	**p.Pro812Ser**	**missense**	**probable-pathogenic**	Neutral	**101**	**G**	**74**	**A**	**27**	**26,7**	**G=0.7327, A=0.2673**	**0.00911**
53	chr16:89986091	G/A	G	SNV	MC1R	exonic	1	c.425G>A	p.Arg142His	missense			263	G	143	A	120	45,6	G=0.5437, A=0.4563	0.00001
53	chr16:23634293	C/T	C	SNV	PALB2	exonic	9	c.2993G>A	p.Gly998Glu	missense	probable-non-pathogenic		111	C	53	T	58	52,3	C=0.4775, T=0.5225	0.00001

Case No.	Locus	Genotype	Ref	Type	Gene	Location	Exon	Coding	Protein	Variant Effect	ClinVar	COSMIC PREDICTION	Coverage	Allele 1 Type	Allele 1 Coverage	Allele 2 Type	Allele 2 Coverage	% Mutated Allele 2	Allele Ratio	p-value
53	chr16:23646191	T/C	T	SNV	PALB2	exonic	4	c.1676A>G	p.Gln559Arg	missense	probable-non-pathogenic		127	T	64	C	63	49,6	T=0.5039, C=0.4961	0.00001
54	**chr16:89985844**	**T/T**	**G**	**SNV**	**MC1R**	**exonic**	**1**	**c.178G>T**	**p.Val60Leu**	**missense**	**pathogenic**		**130**	**G**	**1**	**T**	**64**	**49,2**	**G=0.0154, T=0.9846**	**0.00001**
56	**chr9:21971179**	**G/A**	**G**	**SNV**	**CDKN2A**	**exonic**	**2**	**c.179C>T**	**p.Ala60Val**	**missense**		**Pathogenic**	**193**	**G**	**97**	**A**	**96**	**49,7**	**G=0.5026, A=0.4974**	**0.00001**
56	**chr16:89986117**	**C/T**	**C**	**SNV**	**MC1R**	**exonic**	**1**	**c.451C>T**	**p.Arg151Cys**	**missense**	**other,pathogenic**		**216**	**C**	**103**	**T**	**113**	**52,3**	**C=0.4769, T=0.5231**	**0.00001**
56	chr16:89986091	G/A	G	SNV	MC1R	exonic	1	c.425G>A	p.Arg142His	missense			184	G	100	A	84	45,7	G=0.5435, A=0.4565	0.00001
60	**chr16:89986144**	**C/T**	**C**	**SNV**	**MC1R**	**exonic**	**1**	**c.478C>T**	**p.Arg160Trp**	**missense**	**pathogenic**		**661**	**C**	**345**	**T**	**316**	**47,8**	**C=0.5219, T=0.4781**	**0.00001**
60	**chr11:89017961**	**G/A**	**G**	**SNV**	**TYR**	**exonic**	**4**	**c.1205G>A**	**p.Arg402Gln**	**missense**	**other,pathogenic**		**107**	**G**	**60**	**A**	**47**	**43,9**	**G=0.5607, A=0.4393**	**0.00001**
61	chr11:108141872	T/TA	T	INDEL	ATM	exonic	19	c.2920_2921insA	p.Ser974fs	frameshiftInsertion			353	T	277	TA	76	21,5	T=0.7847, TA=0.2153	0.00001
61	**chr16:89986122**	**C/A**	**C**	**SNV**	**MC1R**	**exonic**	**1**	**c.456C>A**	**p.Tyr152Ter**	**nonsense**		**Pathogenic**	**341**	**C**	**178**	**A**	**163**	**47,8**	**C=0.522, A=0.478**	**0.00001**
61	chr7:124482969	A/C	A	SNV	POT1	exonic	13	c.1055T>G	p.Leu352Trp	missense			1222	A	977	C	245	20,0	A=0.7995, C=0.2005	0.00001
62	chr16:89986154	G/A	G	SNV	MC1R	exonic	1	c.488G>A	p.Arg163Gln	missense			230	G	110	A	120	52,2	G=0.4783, A=0.5217	0.00001
65	chr11:108218009	G/GT	G	INDEL	ATM	exonic	59	c.8588_8589insT	p.Tyr2864fs	frameshiftInsertion			212	G	138	GT	74	34,9	G=0.6509, GT=0.3491	0.00001
65	chr11:108141872	T/TA	T	INDEL	ATM	exonic	19	c.2920_2921insA	p.Ser974fs	frameshiftInsertion			643	T	529	TA	114	17,7	T=0.8227, TA=0.1773	0.00001
65	**chr16:89985844**	**G/T**	**G**	**SNV**	**MC1R**	**exonic**	**1**	**c.178G>T**	**p.Val60Leu**	**missense**	**pathogenic**		**316**	**G**	**180**	**T**	**136**	**43,0**	**G=0.5696, T=0.4304**	**0.00001**
65	chr16:89986091	G/A	G	SNV	MC1R	exonic	1	c.425G>A	p.Arg142His	missense			629	G	296	A	333	52,9	G=0.4706, A=0.5294	0.00001
66	chr11:108141872	T/TA	T	INDEL	ATM	exonic	19	c.2920_2921insA	p.Ser974fs	frameshiftInsertion			242	T	186	TA	56	23,1	T=0.7686, TA=0.2314	0.00001
66	chr9:21970916	C/T	C	SNV	CDKN2A	exonic	2	c.442G>A	p.Ala148Thr	missense		Neutral	187	C	97	T	90	48,1	C=0.5187, T=0.4813	0.00001
66	chr16:23646191	T/C	T	SNV	PALB2	exonic	4	c.1676A>G	p.Gln559Arg	missense	probable-non-pathogenic		154	T	83	C	71	46,1	T=0.539, C=0.461	0.00001
67	chr11:108141872	T/TA	T	INDEL	ATM	exonic	19	c.2920_2921insA	p.Ser974fs	frameshiftInsertion			298	T	254	TA	44	14,8	T=0.8523, TA=0.1477	0.04654
67	chr9:21970916	C/T	C	SNV	CDKN2A	exonic	2	c.442G>A	p.Ala148Thr	missense		Neutral	117	C	52	T	65	55,6	C=0.4444, T=0.5556	0.00001
67	**chr16:89986117**	**C/T**	**C**	**SNV**	**MC1R**	**exonic**	**1**	**c.451C>T**	**p.Arg151Cys**	**missense**	**other,pathogenic**		**394**	**C**	**189**	**T**	**205**	**52,0**	**C=0.4797, T=0.5203**	**0.00001**
68	**chr11:108175462**	**G/A**	**G**	**SNV**	**ATM**	**exonic**	**37**	**c.5557G>A**	**p.Asp1853Asn**	**missense**		**Pathogenic**	**457**	**G**	**250**	**A**	**207**	**45,3**	**G=0.547, A=0.453**	**0.00001**
68	**chr16:89986117**	**C/T**	**C**	**SNV**	**MC1R**	**exonic**	**1**	**c.451C>T**	**p.Arg151Cys**	**missense**	**other,pathogenic**		**1294**	**C**	**615**	**T**	**679**	**52,5**	**C=0.4753, T=0.5247**	**0.00001**
68	chr16:89985722	C/T	C	SNV	MC1R	exonic	1	c.56C>T	p.Thr19Ile	missense			283	C	136	T	147	51,9	C=0.4806, T=0.5194	0.00001
68	chr16:23646191	T/C	T	SNV	PALB2	exonic	4	c.1676A>G	p.Gln559Arg	missense	probable-non-pathogenic		295	T	136	C	159	53,9	T=0.461, C=0.539	0.00001
69	chr11:108141872	T/TA	T	INDEL	ATM	exonic	19	c.2920_2921insA	p.Ser974fs	frameshiftInsertion			439	T	376	TA	63	14,4	T=0.8565, TA=0.1435	0.0223
69	**chr16:89985844**	**G/T**	**G**	**SNV**	**MC1R**	**exonic**	**1**	**c.178G>T**	**p.Val60Leu**	**missense**	**pathogenic**		**139**	**G**	**73**	**T**	**66**	**47,5**	**G=0.5252, T=0.4748**	**0.00001**
73	**chr16:89985940**	**G/A**	**G**	**SNV**	**MC1R**	**exonic**	**1**	**c.274G>A**	**p.Val92Met**	**missense**	**pathogenic**		**853**	**G**	**518**	**A**	**335**	**39,3**	**G=0.6073, A=0.3927**	**0.00001**
73	chr7:124481185	C/A	C	SNV	POT1	exonic	14	c.1211G>T	p.Gly404Val	missense			863	C	422	A	441	51,1	C=0.489, A=0.511	0.00001
78	**chr16:89985940**	**G/A**	**G**	**SNV**	**MC1R**	**exonic**	**1**	**c.274G>A**	**p.Val92Met**	**missense**	**pathogenic**		**512**	**G**	**310**	**A**	**202**	**39,5**	**G=0.6055, A=0.3945**	**0.00001**
78	chr16:23646191	T/C	T	SNV	PALB2	exonic	4	c.1676A>G	p.Gln559Arg	missense	probable-non-pathogenic		251	T	145	C	106	42,2	T=0.5777, C=0.4223	0.00001
79	**chr16:89985844**	**G/T**	**G**	**SNV**	**MC1R**	**exonic**	**1**	**c.178G>T**	**p.Val60Leu**	**missense**	**pathogenic**		**309**	**G**	**155**	**T**	**154**	**49,8**	**G=0.5016, T=0.4984**	**0.00001**
79	**chr16:89986144**	**C/T**	**C**	**SNV**	**MC1R**	**exonic**	**1**	**c.478C>T**	**p.Arg160Trp**	**missense**	**pathogenic**		**625**	**C**	**339**	**T**	**286**	**45,8**	**C=0.5424, T=0.4576**	**0.00001**
80	chr9:21970916	C/T	C	SNV	CDKN2A	exonic	2	c.442G>A	p.Ala148Thr	missense		Neutral	1047	C	501	T	546	52,1	C=0.4785, T=0.5215	0.00001
80	**chr16:89985844**	**G/T**	**G**	**SNV**	**MC1R**	**exonic**	**1**	**c.178G>T**	**p.Val60Leu**	**missense**	**pathogenic**		**404**	**G**	**209**	**T**	**195**	**48,3**	**G=0.5173, T=0.4827**	**0.00001**
80	chr16:89986154	G/A	G	SNV	MC1R	exonic	1	c.488G>A	p.Arg163Gln	missense			1471	G	747	A	724	49,2	G=0.5078, A=0.4922	0.00001
80	chr16:75690279	A/G	A	SNV	TERF2IP	exonic	3	c.970A>G	p.Lys324Glu	missense			446	A	229	G	217	48,7	A=0.5135, G=0.4865	0.00001
80	**chr11:89017961**	**G/A**	**G**	**SNV**	**TYR**	**exonic**	**4**	**c.1205G>A**	**p.Arg402Gln**	**missense**	**other,pathogenic**		**150**	**G**	**68**	**A**	**82**	**54,7**	**G=0.4533, A=0.5467**	**0.00001**
81	chr11:108141872	T/TA	T	INDEL	ATM	exonic	19	c.2920_2921insA	p.Ser974fs	frameshiftInsertion			3884	T	3059	TA	825	21,2	T=0.7876, TA=0.2124	0.00001
81	**chr16:89985844**	**T/T**	**G**	**SNV**	**MC1R**	**exonic**	**1**	**c.178G>T**	**p.Val60Leu**	**missense**	**pathogenic**		**1917**	**G**	**59**	**T**	**1858**	**96,9**	**G=0.0308, T=0.9692**	**0.00001**
86	chr11:108124761	T/C	T	SNV	ATM	exonic	13	c.2119T>C	p.Ser707Pro	missense		Neutral	122	T	100	C	22	18,0	T=0.8197, C=0.1803	0.00636
86	chr11:108196152	AT/A	AT	INDEL	ATM	exonic	46	c.6689delT	p.Leu2231fs	frameshiftDeletion		None	367	AT	283	A	84	22,9	AT=0.7711, A=0.2289	0.00001
86	**chr16:89986144**	**C/T**	**C**	**SNV**	**MC1R**	**exonic**	**1**	**c.478C>T**	**p.Arg160Trp**	**missense**	**pathogenic**		**1405**	**C**	**870**	**T**	**535**	**38,1**	**C=0.6192, T=0.3808**	**0.00001**
86	chr16:23614812	CT/C	CT	INDEL	PALB2	exonic	13	c.3528delA	p.Asp1177fs	frameshiftDeletion			537	CT	172	C	365	68,0	CT=0.3203, C=0.6797	0.00001
86	chr16:23634303	CAA/CA	CAA	INDEL	PALB2	exonic	9	c.2982delT	p.Phe994fs	frameshiftDeletion			144	CA	99	CAA	40	27,8	CAA=0.2778, CA=0.6875, C=0.0347	0.00001
86	chr16:23614857	CA/C	CA	INDEL	PALB2	exonic	13	c.3483delT	p.Phe1161fs	frameshiftDeletion			298	CA	251	C	47	15,8	CA=0.8423, C=0.1577	0.02025
87	chr11:108150294	GA/G	GA	INDEL	ATM	exonic	23	c.3362delA	p.Asn1122fs	frameshiftDeletion			236	GA	178	G	58	24,6	GA=0.7542, G=0.2458	0.00003

Case No.	Locus	Genotype	Ref	Type	Gene	Location	Exon	Coding	Protein	Variant Effect	ClinVar	COSMIC PREDICTION	Coverage	Allele 1 Type	Allele 1 Coverage	Allele 2 Type	Allele 2 Coverage	% Mutated Allele 2	Allele Ratio	p-value
89	chr11:108196152	AT/A	AT	INDEL	ATM	exonic	46	c.6689delT	p.Leu2231fs	frameshiftDeletion		None	817	AT	654	A	163	20,0	AT=0.8005, A=0.1995	0.00001
89	**chr3:52436850**	**A/G**	**A**	**SNV**	**BAP1**	**exonic**	**15**	**c.1928T>C**	**p.Ile643Thr**	**missense**		**Pathogenic**	**1026**	**A**	**252**	**G**	**774**	**75,4**	**A=0.2456, G=0.7544**	**0.00001**
89	**chr16:89985918**	**C/A**	**C**	**SNV**	**MC1R**	**exonic**	**1**	**c.252C>A**	**p.Asp84Glu**	**missense**	**other,pathogenic**		**2624**	**C**	**1359**	**A**	**1265**	**48,2**	**C=0.5179, A=0.4821**	**0.00001**
91	**chr3:52436850**	**A/G**	**A**	**SNV**	**BAP1**	**exonic**	**15**	**c.1928T>C**	**p.Ile643Thr**	**missense**		**Pathogenic**	**726**	**A**	**142**	**G**	**584**	**80,4**	**A=0.1956, G=0.8044**	**0.00001**
92	**chr11:108216476**	**CA/C**	**CA**	**INDEL**	**ATM**	**exonic**	**58**	**c.8426delA**	**p.Lys2811fs**	**frameshiftDeletion**	**probable-pathogenic**		**101**	**CA**	**71**	**C**	**30**	**29,7**	**CA=0.703, C=0.297**	**0.00097**
92	**chr3:52436850**	**A/G**	**A**	**SNV**	**BAP1**	**exonic**	**15**	**c.1928T>C**	**p.Ile643Thr**	**missense**		**Pathogenic**	**1074**	**A**	**231**	**G**	**843**	**78,5**	**A=0.2151, G=0.7849**	**0.00001**
92	**chr16:89986117**	**C/T**	**C**	**SNV**	**MC1R**	**exonic**	**1**	**c.451C>T**	**p.Arg151Cys**	**missense**	**other,pathogenic**		**3457**	**C**	**1706**	**T**	**1751**	**50,7**	**C=0.4935, T=0.5065**	**0.00001**
92	**chr16:89986144**	**C/T**	**C**	**SNV**	**MC1R**	**exonic**	**1**	**c.478C>T**	**p.Arg160Trp**	**missense**	**pathogenic**		**3346**	**C**	**1986**	**T**	**1360**	**40,6**	**C=0.5935, T=0.4065**	**0.00001**
92	chr16:23647173	CT/C	CT	INDEL	PALB2	exonic	4	c.693delA	p.Gly232fs	frameshiftDeletion			172	CT	128	C	44	25,6	CT=0.7442, C=0.2558	0.00017
92	chr7:124493112	TA/T	TA	INDEL	POT1	exonic	10	c.782delT	p.Leu261Ter	nonsense			3802	TA	2632	T	1170	30,8	TA=0.6923, T=0.3077	0.00001
94	chr11:108121521	AC/A	AC	INDEL	ATM	exonic	10	c.1330delC	p.Gln445fs	frameshiftDeletion			197	AC	114	A	83	42,1	AC=0.5787, A=0.4213	0.00001
94	chr11:108150294	GA/G	GA	INDEL	ATM	exonic	23	c.3362delA	p.Asn1122fs	frameshiftDeletion			252	GA	193	G	59	23,4	GA=0.7659, G=0.2341	0.00002
94	**chr3:52436850**	**A/G**	**A**	**SNV**	**BAP1**	**exonic**	**15**	**c.1928T>C**	**p.Ile643Thr**	**missense**		**Pathogenic**	**1199**	**A**	**264**	**G**	**935**	**78,0**	**A=0.2202, G=0.7798**	**0.00001**
94	**chr16:89985844**	**G/T**	**G**	**SNV**	**MC1R**	**exonic**	**1**	**c.178G>T**	**p.Val60Leu**	**missense**	**pathogenic**		**192**	**G**	**137**	**T**	**55**	**28,6**	**G=0.7135, T=0.2865**	**0.00001**
94	chr16:23647173	CT/C	CT	INDEL	PALB2	exonic	4	c.693delA	p.Gly232fs	frameshiftDeletion			129	CT	103	C	26	20,2	CT=0.7984, C=0.2016	0.00951
96	**chr11:108115627**	**C/T**	**C**	**SNV**	**ATM**	**exonic**	**7**	**c.775C>T**	**p.Leu259Phe**	**missense**		**Pathogenic**	**109**	**C**	**54**	**T**	**55**	**50,5**	**C=0.4954, T=0.5046**	**0.00001**
96	**chr3:52436850**	**A/G**	**A**	**SNV**	**BAP1**	**exonic**	**15**	**c.1928T>C**	**p.Ile643Thr**	**missense**		**Pathogenic**	**925**	**A**	**208**	**G**	**717**	**77,5**	**A=0.2249, G=0.7751**	**0.00001**
96	**chr16:89986546**	**G/C**	**G**	**SNV**	**MC1R**	**exonic**	**1**	**c.880G>C**	**p.Asp294His**	**missense**	**pathogenic**		**3993**	**G**	**1918**	**C**	**2075**	**52,0**	**G=0.4803, C=0.5197**	**0.00001**
96	**chr16:89986144**	**C/T**	**C**	**SNV**	**MC1R**	**exonic**	**1**	**c.478C>T**	**p.Arg160Trp**	**missense**	**pathogenic**		**3343**	**C**	**1998**	**T**	**1345**	**40,2**	**C=0.5977, T=0.4023**	**0.00001**
96	chr16:23647173	CT/C	CT	INDEL	PALB2	exonic	4	c.693delA	p.Gly232fs	frameshiftDeletion			141	CT	90	CC	18	12,8	CT=0.6383, CC=0.1277, C=0.234	0.00001
98	**chr11:108175462**	**G/A**	**G**	**SNV**	**ATM**	**exonic**	**37**	**c.5557G>A**	**p.Asp1853Asn**	**missense**		**Pathogenic**	**3287**	**G**	**1665**	**A**	**1622**	**49,3**	**G=0.5065, A=0.4935**	**0.00001**
101	**chr11:108160480**	**T/G**	**T**	**SNV**	**ATM**	**exonic**	**29**	**c.4388T>G**	**p.Phe1463Cys**	**missense**		**Pathogenic**	**138**	**T**	**63**	**G**	**75**	**54,3**	**T=0.4565, G=0.5435**	**0.00001**
101	**chr11:108123551**	**C/T**	**C**	**SNV**	**ATM**	**exonic**	**12**	**c.1810C>T**	**p.Pro604Ser**	**missense**		**Pathogenic**	**177**	**C**	**91**	**T**	**86**	**48,6**	**C=0.5141, T=0.4859**	**0.00001**
101	**chr3:52436850**	**A/G**	**A**	**SNV**	**BAP1**	**exonic**	**15**	**c.1928T>C**	**p.Ile643Thr**	**missense**		**Pathogenic**	**1366**	**A**	**292**	**G**	**1074**	**78,6**	**A=0.2138, G=0.7862**	**0.00001**
102	**chr11:108175462**	**G/A**	**G**	**SNV**	**ATM**	**exonic**	**37**	**c.5557G>A**	**p.Asp1853Asn**	**missense**		**Pathogenic**	**3976**	**G**	**2091**	**A**	**1745**	**43,9**	**G=0.5451, A=0.4549**	**0.00001**
102	chr11:108124551	CA/C	CA	INDEL	ATM	exonic	13	c.1910delA	p.Asp639fs	frameshiftDeletion			271	CA	231	C	40	14,8	CA=0.8524, C=0.1476	0.02675
102	**chr3:52436850**	**A/G**	**A**	**SNV**	**BAP1**	**exonic**	**15**	**c.1928T>C**	**p.Ile643Thr**	**missense**		**Pathogenic**	**1197**	**A**	**344**	**G**	**853**	**71,3**	**A=0.2874, G=0.7126**	**0.00001**
102	chr16:89986154	G/A	G	SNV	MC1R	exonic	1	c.488G>A	p.Arg163Gln	missense			2196	G	1101	A	1095	49,9	G=0.5014, A=0.4986	0.00001
102	**chr16:23632778**	**CA/C**	**CA**	**INDEL**	**PALB2**	**exonic**	**10**	**c.3017delT**	**p.Leu1006Ter**	**nonsense**		**Stop Codon**	**316**	**CA**	**151**	**C**	**165**	**52,2**	**CA=0.4778, C=0.5222**	**0.00001**
102	chr16:23646687	GT/G	GT	INDEL	PALB2	exonic	4	c.1179delA	p.Lys393fs	frameshiftDeletion			132	GT	100	G	32	24,2	GT=0.7576, G=0.2424	0.00057
102	chr16:23614857	CA/C	CA	INDEL	PALB2	exonic	13	c.3483delT	p.Phe1161fs	frameshiftDeletion			476	CA	403	C	73	15,3	CA=0.8466, C=0.1534	0.01177
105	**chr11:108175463**	**A/T**	**A**	**SNV**	**ATM**	**exonic**	**37**	**c.5558A>T**	**p.Asp1853Val**	**missense**		**Pathogenic**	**1728**	**A**	**904**	**T**	**824**	**47,7**	**A=0.5231, T=0.4769**	**0.00001**
105	chr11:108196111	AT/A	AT	INDEL	ATM	exonic	46	c.6648delT	p.Phe2217fs	frameshiftDeletion			232	AT	151	A	81	34,9	AT=0.6509, A=0.3491	0.00001
105	**chr3:52436850**	**A/G**	**A**	**SNV**	**BAP1**	**exonic**	**15**	**c.1928T>C**	**p.Ile643Thr**	**missense**		**Pathogenic**	**270**	**A**	**47**	**G**	**223**	**82,6**	**A=0.1741, G=0.8259**	**0.00001**
106	**chr3:52436850**	**A/G**	**A**	**SNV**	**BAP1**	**exonic**	**15**	**c.1928T>C**	**p.Ile643Thr**	**missense**		**Pathogenic**	**518**	**A**	**84**	**G**	**434**	**83,8**	**A=0.1622, G=0.8378**	**0.00001**
106	chr16:75682028	A/C	A	SNV	KARS, TERF2IP	upstream, exonic	1	c.248A>C	p.Tyr83Ser	missense			221	A	186	C	35	15,8	A=0.8416, C=0.1584	0.00904
106	chr16:23635370	C/T	C	SNV	PALB2	exonic	8	c.2794G>A	p.Val932Met	missense	probable-non-pathogenic		480	C	286	T	194	40,4	C=0.5958, T=0.4042	0.00001
107	**chr11:108175463**	**A/T**	**A**	**SNV**	**ATM**	**exonic**	**37**	**c.5558A>T**	**p.Asp1853Val**	**missense**		**Pathogenic**	**3987**	**A**	**2054**	**T**	**1933**	**48,5**	**A=0.5152, T=0.4848**	**0.00001**
107	chr11:108143522	AAT/AA	AAT	INDEL	ATM	exonic	22	c.3229delT	p.Leu1078fs	frameshiftDeletion			120	AAT	89	AA	28	23,3	AAT=0.7417, AA=0.2333, A=0.025	0.00106
107	chr11:108124551	CA/C	CA	INDEL	ATM	exonic	13	c.1910delA	p.Asp639fs	frameshiftDeletion			156	CA	130	C	26	16,7	CA=0.8333, C=0.1667	0.0142
107	chr16:89986531	T/C	T	SNV	MC1R	exonic	1	c.865T>C	p.Cys289Arg	missense			3997	T	2062	C	1935	48,4	T=0.5159, C=0.4841	0.00001
107	chr16:89986154	G/A	G	SNV	MC1R	exonic	1	c.488G>A	p.Arg163Gln	missense			1953	G	1020	A	933	47,8	G=0.5223, A=0.4777	0.00001
107	chr16:23634303	CA/C	CA	INDEL	PALB2	exonic	9	c.2982delT	p.Phe994fs	frameshiftDeletion			136	CA	52	C	84	61,8	CA=0.3824, C=0.6176	0.00001
109	chr11:108196152	AT/A	AT	INDEL	ATM	exonic	46	c.6689delT	p.Leu2231fs	frameshiftDeletion		None	708	AT	545	A	163	23,0	AT=0.7698, A=0.2302	0.00001

Case No.	Locus	Genotype	Ref	Type	Gene	Location	Exon	Coding	Protein	Variant Effect	ClinVar	COSMIC PREDICTION	Coverage	Allele 1 Type	Allele 1 Coverage	Allele 2 Type	Allele 2 Coverage	% Mutated Allele 2	Allele Ratio	p-value
109	chr11:108126978	CT/C	CT	INDEL	ATM	exonic	14	c.2162delT	p.Leu722fs	frameshiftDeletion			564	CT	402	C	162	28,7	CT=0.7128, C=0.2872	0.00001
109	chr11:108124551	CA/C	CA	INDEL	ATM	exonic	13	c.1910delA	p.Asp639fs	frameshiftDeletion			332	CA	270	C	62	18,7	CA=0.8133, C=0.1867	0.00055
109	**chr3:52436850**	**A/G**	**A**	**SNV**	**BAP1**	**exonic**	**15**	**c.1928T>C**	**p.Ile643Thr**	**missense**		**Pathogenic**	**986**	**A**	**278**	**G**	**708**	**71,8**	**A=0.2819, G=0.7181**	**0.00001**
109	**chr16:89986117**	**C/T**	**C**	**SNV**	**MC1R**	**exonic**	**1**	**c.451C>T**	**p.Arg151Cys**	**missense**	**other,pathogenic**		**2749**	**C**	**1519**	**T**	**1230**	**44,7**	**C=0.5526, T=0.4474**	**0.00001**
109	**chr16:23632778**	**CA/C**	**CA**	**INDEL**	**PALB2**	**exonic**	**10**	**c.3017delT**	**p.Leu1006Ter**	**nonsense**		**Stop Codon**	**305**	**CA**	**122**	**C**	**183**	**60,0**	**CA=0.4, C=0.6**	**0.00001**
109	chr16:23634303	CA/C	CA	INDEL	PALB2	exonic	9	c.2982delT	p.Phe994fs	frameshiftDeletion			145	CA	62	C	83	57,2	CA=0.4276, C=0.5724	0.00001
109	chr7:124469337	GT/G	GT	INDEL	POT1	exonic	16	c.1564delA	p.Thr522fs	frameshiftDeletion			135	GT	86	G	49	36,3	GT=0.637, G=0.363	0.00001
111	**chr11:108175462**	**G/A**	**G**	**SNV**	**ATM**	**exonic**	**37**	**c.5557G>A**	**p.Asp1853Asn**	**missense**		**Pathogenic**	**2974**	**G**	**1555**	**A**	**1339**	**45,0**	**G=0.5373, A=0.4627**	**0.00001**
111	chr11:108124551	CA/C	CA	INDEL	ATM	exonic	13	c.1910delA	p.Asp639fs	frameshiftDeletion			150	CA	102	C	48	32,0	CA=0.68, C=0.32	0.00001
111	**chr3:52436850**	**A/G**	**A**	**SNV**	**BAP1**	**exonic**	**15**	**c.1928T>C**	**p.Ile643Thr**	**missense**		**Pathogenic**	**507**	**A**	**132**	**G**	**375**	**74,0**	**A=0.2604, G=0.7396**	**0.00001**
111	chr16:23646191	T/C	T	SNV	PALB2	exonic	4	c.1676A>G	p.Gln559Arg	missense	probable-non-pathogenic		144	T	74	C	70	48,6	T=0.5139, C=0.4861	0.00001
111	chr16:23641461	C/G	C	SNV	PALB2	exonic	5	c.2014G>C	p.Glu672Gln	missense	probable-non-pathogenic		129	C	71	G	58	45,0	C=0.5504, G=0.4496	0.00001
111	chr16:23634293	C/T	C	SNV	PALB2	exonic	9	c.2993G>A	p.Gly998Glu	missense	probable-non-pathogenic		108	C	65	T	43	39,8	C=0.6019, T=0.3981	0.00001
111	chr16:23634303	CA/C	CA	INDEL	PALB2	exonic	9	c.2982delT	p.Phe994fs	frameshiftDeletion			117	CA	42	C	75	64,1	CA=0.359, C=0.641	0.00001
112	chr11:108124551	CA/C	CA	INDEL	ATM	exonic	13	c.1910delA	p.Asp639fs	frameshiftDeletion			153	CA	113	C	40	26,1	CA=0.7386, C=0.2614	0.00076
112	**chr16:89986144**	**C/T**	**C**	**SNV**	**MC1R**	**exonic**	**1**	**c.478C>T**	**p.Arg160Trp**	**missense**	**pathogenic**		**1255**	**C**	**688**	**T**	**567**	**45,2**	**C=0.5482, T=0.4518**	**0.00001**
112	**chr16:23632778**	**CA/C**	**CA**	**INDEL**	**PALB2**	**exonic**	**10**	**c.3017delT**	**p.Leu1006Ter**	**nonsense**		**Stop Codon**	**186**	**CA**	**59**	**C**	**127**	**68,3**	**CA=0.3172, C=0.6828**	**0.00001**
112	chr16:23646191	T/C	T	SNV	PALB2	exonic	4	c.1676A>G	p.Gln559Arg	missense	probable-non-pathogenic		143	T	68	C	75	52,4	T=0.4755, C=0.5245	0.00001
112	chr16:23641461	C/G	C	SNV	PALB2	exonic	5	c.2014G>C	p.Glu672Gln	missense	probable-non-pathogenic		144	C	82	G	62	43,1	C=0.5694, G=0.4306	0.00001
112	chr16:23614812	CT/C	CT	INDEL	PALB2	exonic	13	c.3528delA	p.Asp1177fs	frameshiftDeletion			381	CT	107	C	274	71,9	CT=0.2808, C=0.7192	0.00001
112	chr16:23614857	CA/C	CA	INDEL	PALB2	exonic	13	c.3483delT	p.Phe1161fs	frameshiftDeletion			240	CA	197	C	43	17,9	CA=0.8208, C=0.1792	0.00358
112	chr7:124503493	AT/A	AT	INDEL	POT1	exonic	8	c.456delA	p.Lys152fs	frameshiftDeletion			154	AT	116	A	38	24,7	AT=0.7532, A=0.2468	0.00472
116	**chr11:108202221**	**AATG/ATG**	**ATTG**	**SNV,INDEL**	**ATM**	**exonic, exonic**	**51**	**c.7567T>A, c.7567delT**	**p.Leu2523Met, p.Leu2523fs**	**missense, frameshiftDeletion**	**probable-pathogenic**		**318**	**AATG**	**98**	**ATG**	**220**	**69,2**	**ATTG=0.0, AATG=0.3082, AT=0.0, ATG=0.6918**	**0.00001**
116	**chr16:89985940**	**G/A**	**G**	**SNV**	**MC1R**	**exonic**	**1**	**c.274G>A**	**p.Val92Met**	**missense**	**pathogenic**		**908**	**G**	**472**	**A**	**436**	**48,0**	**G=0.5198, A=0.4802**	**0.00001**
117	chr11:108196152	AT/A	AT	INDEL	ATM	exonic	46	c.6689delT	p.Leu2231fs	frameshiftDeletion		None	369	AT	286	A	83	22,5	AT=0.7751, A=0.2249	0.00001
117	**chr16:89985940**	**G/A**	**G**	**SNV**	**MC1R**	**exonic**	**1**	**c.274G>A**	**p.Val92Met**	**missense**	**pathogenic**		**1459**	**G**	**820**	**A**	**639**	**43,8**	**G=0.562, A=0.438**	**0.00001**
117	**chr16:23632778**	**CAA/CA**	**CAA**	**INDEL**	**PALB2**	**exonic**	**10**	**c.3017delT**	**p.Leu1006Ter**	**nonsense**		**Stop Codon**	**173**	**CAA**	**61**	**CA**	**103**	**59,5**	**CAA=0.3526, CA=0.5954, C=0.052**	**0.00001**
118	**chr11:108216476**	**CA/C**	**CA**	**INDEL**	**ATM**	**exonic**	**58**	**c.8426delA**	**p.Lys2811fs**	**frameshiftDeletion**	**probable-pathogenic**		**101**	**CA**	**68**	**C**	**33**	**32,7**	**CA=0.6733, C=0.3267**	**0.00003**
118	chr11:108122700	T/C	T	SNV	ATM	exonic	11	c.1744T>C	p.Phe582Leu	missense			373	T	182	C	191	51,2	T=0.4879, C=0.5121	0.00001
118	chr11:108142090	A/G	A	SNV	ATM	exonic	20	c.3034A>G	p.Arg1012Gly	missense			107	A	87	G	20	18,7	A=0.8131, G=0.1869	0.00591
118	**chr16:89986117**	**C/T**	**C**	**SNV**	**MC1R**	**exonic**	**1**	**c.451C>T**	**p.Arg151Cys**	**missense**	**other,pathogenic**		**1826**	**C**	**980**	**T**	**846**	**46,3**	**C=0.5367, T=0.4633**	**0.00001**
119	**chr3:52436850**	**A/G**	**A**	**SNV**	**BAP1**	**exonic**	**15**	**c.1928T>C**	**p.Ile643Thr**	**missense**		**Pathogenic**	**1002**	**A**	**272**	**G**	**730**	**72,9**	**A=0.2715, G=0.7285**	**0.00001**
119	**chr16:23632778**	**CA/C**	**CA**	**INDEL**	**PALB2**	**exonic**	**10**	**c.3017delT**	**p.Leu1006Ter**	**nonsense**		**Stop Codon**	**309**	**CA**	**170**	**C**	**139**	**45,0**	**CA=0.5502, C=0.4498**	**0.00001**
119	chr16:23634303	CA/C	CA	INDEL	PALB2	exonic	9	c.2982delT	p.Phe994fs	frameshiftDeletion			232	CA	117	C	115	49,6	CA=0.5043, C=0.4957	0.00001
120	**chr11:108202221**	**AATG/ATG**	**ATTG**	**SNV, INDEL**	**ATM**	**exonic, exonic**	**51**	**c.7567T>A, c.7567delT**	**p.Leu2523Met, p.Leu2523fs**	**missense, frameshiftDeletion**	**probable-pathogenic**		**307**	**ATG**	**212**	**AATG**	**94**	**30,6**	**ATTG=0.0033, AATG=0.3062, ATG=0.6906**	**0.00001**
120	**chr9:21971179**	**G/A**	**G**	**SNV**	**CDKN2A**	**exonic**	**2**	**c.179C>T**	**p.Ala60Val**	**missense**		**Pathogenic**	**242**	**G**	**147**	**A**	**95**	**39,3**	**G=0.6074, A=0.3926**	**0.00001**
120	**chr16:89985940**	**G/A**	**G**	**SNV**	**MC1R**	**exonic**	**1**	**c.274G>A**	**p.Val92Met**	**missense**	**pathogenic**		**612**	**G**	**338**	**A**	**274**	**44,8**	**G=0.5523, A=0.4477**	**0.00001**
120	chr16:23646191	T/C	T	SNV	PALB2	exonic	4	c.1676A>G	p.Gln559Arg	missense	probable-non-pathogenic		120	T	76	C	44	36,7	T=0.6333, C=0.3667	0.00001
122	**chr16:89986144**	**C/T**	**C**	**SNV**	**MC1R**	**exonic**	**1**	**c.478C>T**	**p.Arg160Trp**	**missense**	**pathogenic**		**3623**	**C**	**2112**	**T**	**1511**	**41,7**	**C=0.5829, T=0.4171**	**0.00001**

Case No.	Locus	Genotype	Ref	Type	Gene	Location	Exon	Coding	Protein	Variant Effect	ClinVar	COSMIC PREDICTION	Coverage	Allele 1 Type	Allele 1 Coverage	Allele 2 Type	Allele 2 Coverage	% Mutated Allele 2	Allele Ratio	p-value
122	**chr16:89986546**	**G/C**	**G**	**SNV**	**MC1R**	**exonic**	**1**	**c.880G>C**	**p.Asp294His**	**missense**	**pathogenic**		**3987**	**G**	**1955**	**C**	**2032**	**51,0**	**G=0.4903, C=0.5097**	**0.00001**
122	chr16:23646191	T/C	T	SNV	PALB2	exonic	4	c.1676A>G	p.Gln559Arg	missense	probable-non-pathogenic		327	T	175	C	152	46,5	T=0.5352, C=0.4648	0.00001
123	**chr16:89986144**	**C/T**	**C**	**SNV**	**MC1R**	**exonic**	**1**	**c.478C>T**	**p.Arg160Trp**	**missense**	**pathogenic**		**1781**	**C**	**1038**	**T**	**743**	**41,7**	**C=0.5828, T=0.4172**	**0.00001**
123	chr16:23646191	T/C	T	SNV	PALB2	exonic	4	c.1676A>G	p.Gln559Arg	missense	probable-non-pathogenic		288	T	137	C	151	52,4	T=0.4757, C=0.5243	0.00001
123	chr16:23641461	C/G	C	SNV	PALB2	exonic	5	c.2014G>C	p.Glu672Gln	missense	probable-non-pathogenic		767	C	385	G	382	49,8	C=0.502, G=0.498	0.00001
126	chr11:108196152	AT/A	AT	INDEL	ATM	exonic	46	c.6689delT	p.Leu2231fs	frameshiftDeletion		None	308	AT	255	A	53	17,2	AT=0.8279, A=0.1721	0.0004
126	**chr11:108175462**	**G/A**	**G**	**SNV**	**ATM**	**exonic**	**37**	**c.5557G>A**	**p.Asp1853Asn**	**missense**		**Pathogenic**	**2470**	**G**	**1327**	**A**	**1006**	**40,7**	**G=0.5688, A=0.4312**	**0.00001**
126	**chr16:89986117**	**C/T**	**C**	**SNV**	**MC1R**	**exonic**	**1**	**c.451C>T**	**p.Arg151Cys**	**missense**	**other,pathogenic**		**1123**	**C**	**604**	**T**	**519**	**46,2**	**C=0.5378, T=0.4622**	**0.00001**
126	chr16:23634384	CT/C	CT	INDEL	PALB2	exonic	9	c.2901delA	p.Ala968fs	frameshiftDeletion			151	CT	113	C	38	25,2	CT=0.7483, C=0.2517	0.00059
127	**chr11:108175462**	**G/A**	**G**	**SNV**	**ATM**	**exonic**	**37**	**c.5557G>A**	**p.Asp1853Asn**	**missense**		**Pathogenic**	**2330**	**G**	**1232**	**A**	**1098**	**47,1**	**G=0.5288, A=0.4712**	**0.00001**
127	**chr3:52436850**	**A/G**	**A**	**SNV**	**BAP1**	**exonic**	**15**	**c.1928T>C**	**p.Ile643Thr**	**missense**		**Pathogenic**	**422**	**A**	**121**	**G**	**301**	**71,3**	**A=0.2867, G=0.7133**	**0.00001**
128	**chr16:89986117**	**C/T**	**C**	**SNV**	**MC1R**	**exonic**	**1**	**c.451C>T**	**p.Arg151Cys**	**missense**	**other,pathogenic**		**1547**	**C**	**821**	**T**	**726**	**46,9**	**C=0.5307, T=0.4693**	**0.00001**
128	chr16:89986091	G/A	G	SNV	MC1R	exonic	1	c.425G>A	p.Arg142His	missense			629	G	338	A	291	46,3	G=0.5374, A=0.4626	0.00001
129	chr16:23646191	T/C	T	SNV	PALB2	exonic	4	c.1676A>G	p.Gln559Arg	missense	probable-non-pathogenic		174	T	91	C	83	47,7	T=0.523, C=0.477	0.00001
130	**chr11:108138003**	**T/C**	**T**	**SNV**	**ATM**	**exonic**	**17**	**c.2572T>C**	**p.Phe858Leu**	**missense**		**Pathogenic**	**150**	**T**	**87**	**C**	**63**	**42,0**	**T=0.58, C=0.42**	**0.00001**
132	chr11:108196152	AT/A	AT	INDEL	ATM	exonic	46	c.6689delT	p.Leu2231fs	frameshiftDeletion		None	694	AT	596	A	98	14,1	AT=0.8588, A=0.1412	0.00255
132	**chr11:108175462**	**G/A**	**G**	**SNV**	**ATM**	**exonic**	**37**	**c.5557G>A**	**p.Asp1853Asn**	**missense**		**Pathogenic**	**3958**	**G**	**2112**	**A**	**1644**	**41,5**	**G=0.5623, A=0.4377**	**0.00001**
132	chr11:108150294	GA/G	GA	INDEL	ATM	exonic	23	c.3362delA	p.Asn1122fs	frameshiftDeletion			126	GA	91	G	35	27,8	GA=0.7222, G=0.2778	0.00006
132	chr11:108170508	TA/T	TA	INDEL	ATM	exonic	34	c.5074delA	p.Cys1726fs	frameshiftDeletion			125	TA	99	T	26	20,8	TA=0.792, T=0.208	0.0106
132	**chr3:52436850**	**A/G**	**A**	**SNV**	**BAP1**	**exonic**	**15**	**c.1928T>C**	**p.Ile643Thr**	**missense**		**Pathogenic**	**1046**	**A**	**349**	**G**	**697**	**66,6**	**A=0.3337, G=0.6663**	**0.00001**
132	**chr16:89986546**	**G/C**	**G**	**SNV**	**MC1R**	**exonic**	**1**	**c.880G>C**	**p.Asp294His**	**missense**	**pathogenic**		**3984**	**G**	**1961**	**C**	**2023**	**50,8**	**G=0.4922, C=0.5078**	**0.00001**
132	chr16:23634303	CA/C	CA	INDEL	PALB2	exonic	9	c.2982delT	p.Phe994fs	frameshiftDeletion			171	CA	92	C	79	46,2	CA=0.538, C=0.462	0.00001
132	chr16:23641767	CT/C	CT	INDEL	PALB2	exonic	5	c.1707delA	p.Glu570fs	frameshiftDeletion			188	CT	142	C	46	24,5	CT=0.7553, C=0.2447	0.00018
132	chr7:124503677	CT/C	CT	INDEL	POT1	exonic	8	c.272delA	p.Lys91fs	frameshiftDeletion			511	CT	428	C	83	16,2	CT=0.8376, C=0.1624	0.00048
135	chr11:108196152	AT/A	AT	INDEL	ATM	exonic	46	c.6689delT	p.Leu2231fs	frameshiftDeletion		None	568	AT	451	A	117	20,6	AT=0.794, A=0.206	0.00001
135	**chr3:52436850**	**A/G**	**A**	**SNV**	**BAP1**	**exonic**	**15**	**c.1928T>C**	**p.Ile643Thr**	**missense**		**Pathogenic**	**1075**	**A**	**283**	**G**	**792**	**73,7**	**A=0.2633, G=0.7367**	**0.00001**
135	**chr9:21971120**	**G/A**	**G**	**SNV**	**CDKN2A**	**exonic**	**2**	**c.238C>T**	**p.Arg80Ter**	**nonsense**	**pathogenic**		**816**	**G**	**550**	**A**	**266**	**32,6**	**G=0.6740, AG=0.3260**	**0.00001**
135	**chr16:89986546**	**G/C**	**G**	**SNV**	**MC1R**	**exonic**	**1**	**c.880G>C**	**p.Asp294His**	**missense**	**pathogenic**		**3990**	**G**	**1865**	**C**	**2125**	**53,3**	**G=0.4674, C=0.5326**	**0.00001**
135	chr16:23614812	CT/C	CT	INDEL	PALB2	exonic	13	c.3528delA	p.Asp1177fs	frameshiftDeletion			572	CT	196	C	376	65,7	CT=0.3427, C=0.6573	0.00001
135	chr16:23646687	GT/G	GT	INDEL	PALB2	exonic	4	c.1179delA	p.Lys393fs	frameshiftDeletion			108	GT	69	G	39	36,1	GT=0.6389, G=0.3611	0.00001
135	chr16:23634384	CT/C	CT	INDEL	PALB2	exonic	9	c.2901delA	p.Ala968fs	frameshiftDeletion			336	CT	275	C	61	18,2	CT=0.8185, C=0.1815	0.00214
135	chr7:124503677	CT/C	CT	INDEL	POT1	exonic	8	c.272delA	p.Lys91fs	frameshiftDeletion			398	CT	338	C	60	15,1	CT=0.8492, C=0.1508	0.01461
136	**chr11:108175462**	**G/A**	**G**	**SNV**	**ATM**	**exonic**	**37**	**c.5557G>A**	**p.Asp1853Asn**	**missense**		**Pathogenic**	**2476**	**G**	**1263**	**A**	**1098**	**44,3**	**G=0.5349, A=0.4651**	**0.00001**
136	chr11:108126978	CT/C	CT	INDEL	ATM	exonic	14	c.2162delT	p.Leu722fs	frameshiftDeletion			203	CT	148	C	55	27,1	CT=0.7291, C=0.2709	0.00004
136	chr11:108122627	G/T	G	SNV	ATM	exonic	11	c.1671G>T	p.Met557Ile	missense			373	G	313	T	60	16,1	G=0.8391, T=0.1609	0.00573
136	**chr16:89985940**	**G/A**	**G**	**SNV**	**MC1R**	**exonic**	**1**	**c.274G>A**	**p.Val92Met**	**missense**	**pathogenic**		**1205**	**G**	**670**	**A**	**535**	**44,4**	**G=0.556, A=0.444**	**0.00001**
136	chr16:23634303	CA/C	CA	INDEL	PALB2	exonic	9	c.2982delT	p.Phe994fs	frameshiftDeletion			107	CA	43	C	64	59,8	CA=0.4019, C=0.5981	0.00001
138	chr11:108124551	CA/C	CA	INDEL	ATM	exonic	13	c.1910delA	p.Asp639fs	frameshiftDeletion			133	CA	96	C	37	27,8	CA=0.7218, C=0.2782	0.00049
138	**chr3:52436850**	**A/G**	**A**	**SNV**	**BAP1**	**exonic**	**15**	**c.1928T>C**	**p.Ile643Thr**	**missense**		**Pathogenic**	**757**	**A**	**230**	**G**	**527**	**69,6**	**A=0.3038, G=0.6962**	**0.00001**
138	**chr16:89986546**	**G/C**	**G**	**SNV**	**MC1R**	**exonic**	**1**	**c.880G>C**	**p.Asp294His**	**missense**	**pathogenic**		**3993**	**G**	**1974**	**C**	**2019**	**50,6**	**G=0.4944, C=0.5056**	**0.00001**
138	chr16:23641767	CT/C	CT	INDEL	PALB2	exonic	5	c.1707delA	p.Glu570fs	frameshiftDeletion			110	CT	72	C	38	34,5	CT=0.6545, C=0.3455	0.00001

Case No.	Locus	Genotype	Ref	Type	Gene	Location	Exon	Coding	Protein	Variant Effect	ClinVar	COSMIC PREDICTION	Coverage	Allele 1 Type	Allele 1 Coverage	Allele 2 Type	Allele 2 Coverage	% Mutated Allele 2	Allele Ratio	p-value
138	chr7:124503677	CT/C	CT	INDEL	POT1	exonic	8	c.272delA	p.Lys91fs	frameshiftDeletion			421	CT	319	C	102	24,2	CT=0.7577, C=0.2423	0.00001
141	**chr11:108216476**	**CA/C**	**CA**	**INDEL**	**ATM**	**exonic**	**58**	**c.8426delA**	**p.Lys2811fs**	**frameshiftDeletion**	**probable-pathogenic**		**103**	**CA**	**73**	**C**	**30**	**29,1**	**CA=0.7087, C=0.2913**	**0.00073**
141	**chr3:52436850**	**A/G**	**A**	**SNV**	**BAP1**	**exonic**	**15**	**c.1928T>C**	**p.Ile643Thr**	**missense**		**Pathogenic**	**1159**	**A**	**253**	**G**	**906**	**78,2**	**A=0.2183, G=0.7817**	**0.00001**
141	**chr16:89986117**	**C/T**	**C**	**SNV**	**MC1R**	**exonic**	**1**	**c.451C>T**	**p.Arg151Cys**	**missense**	**other,pathogenic**		**3986**	**C**	**2181**	**T**	**1805**	**45,3**	**C=0.5472, T=0.4528**	**0.00001**
141	chr16:23646191	T/C	T	SNV	PALB2	exonic	4	c.1676A>G	p.Gln559Arg	missense	probable-non-pathogenic		346	T	177	C	169	48,8	T=0.5116, C=0.4884	0.00001
142	chr11:108124551	CA/C	CA	INDEL	ATM	exonic	13	c.1910delA	p.Asp639fs	frameshiftDeletion			316	CA	252	C	64	20,3	CA=0.7975, C=0.2025	0.00085
142	**chr3:52436850**	**A/G**	**A**	**SNV**	**BAP1**	**exonic**	**15**	**c.1928T>C**	**p.Ile643Thr**	**missense**		**Pathogenic**	**1045**	**A**	**253**	**G**	**792**	**75,8**	**A=0.2421, G=0.7579**	**0.00001**
142	**chr16:89986546**	**G/C**	**G**	**SNV**	**MC1R**	**exonic**	**1**	**c.880G>C**	**p.Asp294His**	**missense**	**pathogenic**		**3992**	**G**	**2026**	**C**	**1966**	**49,2**	**G=0.5075, C=0.4925**	**0.00001**
142	chr16:23641767	CT/C	CT	INDEL	PALB2	exonic	5	c.1707delA	p.Glu570fs	frameshiftDeletion			145	CT	100	C	45	31,0	CT=0.6897, C=0.3103	0.00002
143	chr11:108202222	T/A	T	SNV	ATM	exonic	51	c.7567T>A	p.Leu2523Met	missense			217	T	179	A	38	17,5	T=0.8249, A=0.1751	0.00477
143	chr16:89986154	G/A	G	SNV	MC1R	exonic	1	c.488G>A	p.Arg163Gln	missense			489	G	244	A	245	50,1	G=0.499, A=0.501	0.00001
145	chr11:108196152	AT/A	AT	INDEL	ATM	exonic	46	c.6689delT	p.Leu2231fs	frameshiftDeletion		None	536	AT	413	A	123	22,9	AT=0.7705, A=0.2295	0.00001
145	chr11:108124551	CA/C	CA	INDEL	ATM	exonic	13	c.1910delA	p.Asp639fs	frameshiftDeletion			263	CA	209	C	54	20,5	CA=0.7947, C=0.2053	0.00036
145	**chr11:108143456**	**C/G**	**C**	**SNV**	**ATM**	**exonic**	**22**	**c.3161C>G**	**p.Pro1054Arg**	**missense**		**Pathogenic**	**102**	**C**	**73**	**G**	**29**	**28,4**	**C=0.7157, G=0.2843**	**0.00001**
145	chr16:23637613	A/C	A	SNV	PALB2	exonic	7	c.2692T>G	p.Trp898Gly	missense			189	A	156	C	33	17,5	A=0.8254, C=0.1746	0.00349
146	chr11:108150294	GA/G	GA	INDEL	ATM	exonic	23	c.3362delA	p.Asn1122fs	frameshiftDeletion			266	GA	219	G	47	17,7	GA=0.8233, G=0.1767	0.00361
146	chr16:23646191	T/C	T	SNV	PALB2	exonic	4	c.1676A>G	p.Gln559Arg	missense	probable-non-pathogenic		249	T	119	C	130	52,2	T=0.4779, C=0.5221	0.00001
146	chr16:23641461	C/G	C	SNV	PALB2	exonic	5	c.2014G>C	p.Glu672Gln	missense	probable-non-pathogenic		784	C	405	G	379	48,3	C=0.5166, G=0.4834	0.00001
149	**chr11:108175462**	**G/A**	**G**	**SNV**	**ATM**	**exonic**	**37**	**c.5557G>A**	**p.Asp1853Asn**	**missense**		**Pathogenic**	**3967**	**G**	**2079**	**A**	**1888**	**47,6**	**G=0.5241, A=0.4759**	**0.00001**
149	**chr16:89985844**	**G/T**	**G**	**SNV**	**MC1R**	**exonic**	**1**	**c.178G>T**	**p.Val60Leu**	**missense**	**pathogenic**		**168**	**G**	**111**	**T**	**57**	**33,9**	**G=0.6607, T=0.3393**	**0.00001**
150	**chr3:52436850**	**A/G**	**A**	**SNV**	**BAP1**	**exonic**	**15**	**c.1928T>C**	**p.Ile643Thr**	**missense**		**Pathogenic**	**967**	**A**	**356**	**G**	**611**	**63,2**	**A=0.3681, G=0.6319**	**0.00001**
151	**chr11:108160480**	**T/G**	**T**	**SNV**	**ATM**	**exonic**	**29**	**c.4388T>G**	**p.Phe1463Cys**	**missense**		**Pathogenic**	**267**	**T**	**156**	**G**	**111**	**41,6**	**T=0.5843, G=0.4157**	**0.00001**
152	**chr16:89986546**	**G/C**	**G**	**SNV**	**MC1R**	**exonic**	**1**	**c.880G>C**	**p.Asp294His**	**missense**	**pathogenic**		**2876**	**G**	**1389**	**C**	**1487**	**51,7**	**G=0.4830, C=0.5170**	**0.00001**
152	chr16:89986091	G/A	G	SNV	MC1R	exonic	1	c.425G>A	p.Arg142His	missense			580	G	393	A	187	32,2	G=0.6776, A=0.3224	0.00001
155	**chr16:89986144**	**C/T**	**C**	**SNV**	**MC1R**	**exonic**	**1**	**c.478C>T**	**p.Arg160Trp**	**missense**	**pathogenic**		**1839**	**C**	**1089**	**T**	**750**	**40,8**	**C=0.5922, T=0.4078**	**0.00001**
155	chr7:124469337	GT/G	GT	INDEL	POT1	exonic	16	c.1564delA	p.Thr522fs	frameshiftDeletion			144	GT	92	G	52	36,1	GT=0.6389, G=0.3611	0.00001
157	**chr3:52436850**	**A/G**	**A**	**SNV**	**BAP1**	**exonic**	**15**	**c.1928T>C**	**p.Ile643Thr**	**missense**		**Pathogenic**	**887**	**A**	**393**	**G**	**494**	**55,7**	**A=0.4431, G=0.5569**	**0.00001**
157	**chr16:89986546**	**G/C**	**G**	**SNV**	**MC1R**	**exonic**	**1**	**c.880G>C**	**p.Asp294His**	**missense**	**pathogenic**		**1962**	**G**	**956**	**C**	**1006**	**51,3**	**G=0.4873, C=0.5127**	**0.00001**
159	**chr3:52436850**	**A/G**	**A**	**SNV**	**BAP1**	**exonic**	**15**	**c.1928T>C**	**p.Ile643Thr**	**missense**		**Pathogenic**	**525**	**A**	**82**	**G**	**443**	**84,4**	**A=0.1562, G=0.8438**	**0.00001**
160	chr11:108202222	T/A	T	SNV	ATM	exonic	51	c.7567T>A	p.Leu2523Met	missense			302	T	236	A	66	21,9	T=0.7815, A=0.2185	0.00038
160	**chr16:23632778**	**CA/C**	**CA**	**INDEL**	**PALB2**	**exonic**	**10**	**c.3017delT**	**p.Leu1006Ter**	**nonsense**		**Stop Codon**	**411**	**CA**	**279**	**C**	**132**	**32,1**	**CA=0.6788, C=0.3212**	**0.00001**