Table E1 Pathogenic mutants detected in bronchiectasis patients

Gene.refgene	Func.refgene	Exon	cDNA change	AA change	ExonicFunc.refgene	No. of bronchiectasis patients	No. of healthy subjects	ExAC_Freq	dbSNP	ClinVar_SIG	ClinVar_DIS	SIFT_score	SIFT_pred	Polyphen2_HDIV_score	Polyphen2_HDIV_pred
CFTR	Exonic	Exon 12	c.A1666G	p.I556V	Nonsynsonymous SNV	17	9	0.0029	rs75789129	Pathogenic|probable-pathogenic	Cystic_fibrosis|Nonclassic_cystic_fibrosis	0.88	T	0.334	B
CFTR	Exonic	Exon 20	c.G3205A	p.G1069R	Nonsynsonymous SNV	4	1	0.0002	rs200321110	Untested	Cystic_fibrosis	0.4	T	0.772	P
CFTR	Exonic	Exon 6	c.A650G	p.E217G	Nonsynsonymous SNV	2	1	0.0039	rs121909046	Pathogenic	Cystic_fibrosis	0.31	T	0.053	B
CFTR	Exonic	Exon 25	c.G4056C	p.Q1352H	Nonsynsonymous SNV	5	3	0.001	rs113857788	Pathogenic	Cystic_fibrosis; Congenital_bilateral_absence_of_the_vas_deferens	0	D	1	D
CFTR	Exonic	Exon 17	c.G2770A	p.D924N	Nonsynsonymous SNV	1	0	7.42E-05	rs201759207	Untested	Cystic_fibrosis	0.6	T	0.996	D
CFTR	Intronic	–	–	–	–	8	7	0.0107	rs73715573	Probable-pathogenic|probable-pathogenic	Congenital_bilateral_absence_of_the_vas_deferens; Cystic_fibrosis	–	–	–	–
CFTR	Exonic	Exon 17	c.G2684A	p.S895N	Nonsynsonymous SNV	2	1	0.0003	rs201864483	Untested	Cystic_fibrosis	0.71	T	0	B
CFTR	Exonic	Exon 11	c.G1558A	p.V520I	Nonsynsonymous SNV	1	0	0.0002	rs77646904	Probable-pathogenic|untested|unknown	Nonclassic_cystic_fibrosis|Cystic_fibrosis|not_provided; Cystic_fibrosis	0.02	D	0.063	B
CFTR	Exonic	Exon 18	c.A2936C	p.D979A	Nonsynsonymous SNV	1	0	2.48E-05	rs397508462	Untested	Cystic_fibrosis; Cystic_fibrosis	0	D	1	D
CFTR	Exonic	Exon 14	c.C1909T	p.Q637X	Stopgain	2	0	.	rs397508320	Untested	Cystic_fibrosis	0.9	T	–	–
CFTR	Exonic	Exon 6	c.G601A	p.V201M	Nonsynsonymous SNV	2	0	0.0002	rs138338446	Untested	Cystic_fibrosis	0.01	D	0.945	P
CFTR	Exonic	Exon 4	c.T374C	p.I125T	Nonsynsonymous SNV	1	1	0.0006	rs141723617	Untested	Cystic_fibrosis	.	.	0.17	B
CFTR	Intronic	–	–	–	–	1	0	7.22E-05	–	–	–	–	–	–	–
CFTR	Intronic	–	–	–	–	1	0	3.34E-05	–	–	–	–	–	–	–
CFTR	Intronic	–	–	–	–	1	0	3.30E-05	rs372266030	–	–	–	–	–	–
CFTR	Exonic	Exon 27	c.T4262A	p.V1421E	Nonsynsonymous SNV	1	0	–	–	–	–	–	–	0.837	P
CFTR	Exonic	Exon 2	c.C91T	p.R31C	Nonsynsonymous SNV	1	3	0.0017	rs1800073	Unknown	Cystic_fibrosis	0.14	T	0.989	D
CFTR	Exonic	Exon 20	c.C3289T	p.R1097C	Nonsynsonymous SNV	1	1	0.0002	rs201591901	–	–	0	D	1	D
CFTR	Exonic	Exon 22	c.G3592A	p.V1198M	Nonsynsonymous SNV	1	0	1.65E-05	rs576710089	–	–	0.12	T	0.789	P
SCNN1A	Exonic	Exon 5	c.C1252A	p.P418T	Nonsynsonymous SNV	1	0	8.83E-06	–	–	–	0.52	T	1	D
SCNN1A	Exonic	Exon 3	c.T953G	p.I318S	Nonsynsonymous SNV	1	0	–	–	–	–	0	D	0.794	P
SCNN1A	Exonic	Exon 1	c.C28T	p.R10W	Nonsynsonymous SNV	1	0	0.0001	rs531838244	–	–	0.01	D	0	B
SCNN1B	Exonic	Exon 5	c.A863G	p.N288S	Nonsynsonymous SNV	1	0	8.24E-06	rs137852712	Pathogenic	Bronchiectasis	0.49	T	0.768	P
SCNN1B	Exonic	Exon 5	c.A806T	p.Y269F	Nonsynsonymous SNV	1	0	–	–	–	–	0	D	0.996	D
SCNN1B	Exonic	Exon 2	c.A268G	p.M90V	Nonsynsonymous SNV	1	0	0.0001	rs150466803	–	–	0.04	D	0.101	B
CCDC103	Exonic	Exon 4	c.C508G	p.L170V	Nonsynsonymous SNV	1	0	–	–	–	–	0.05	D	0.998	D
CCDC114	Exonic	Exon 7	c.G697A	p.D233N	Nonsynsonymous SNV	2	1	0.0018	rs145629439	–	–	0.15	T	0.997	D
CCDC114	Exonic	Exon 3	c.G214A	p.E72K	Nonsynsonymous SNV	2	2	0.0001	rs368408933	–	–	0.74	T	0.884	P
CCDC114	Exonic	Exon 10	c.G976A	p.V326M	Nonsynsonymous SNV	1	0	–	–	–	–	0.1	T	0.891	P
CCDC39	Exonic	Exon 6	c.T692G	p.I231R	Nonsynsonymous SNV	1	0	–	–	–	–	0.02	D	1	D
CCDC39	Exonic	Exon 7	c.A800C	p.K267T	Nonsynsonymous SNV	1	0	–	–	–	–	0.5	T	0.958	D
CCDC39	Exonic	Exon 4	c.A455C	p.H152P	Nonsynsonymous SNV	1	0	4.93E-05	rs370141610	–	–	0.04	D	0.963	D
CCDC40	Exonic	Exon 4	c.C607T	p.R203C	Nonsynsonymous SNV	1	0	3.39E-05	rs367978204	–	–	0	D	1	D
CCDC40	Exonic	Exon 7	c.994dupT	p.L331fs	Frameshift insertion	1	0	8.41E-06	–	–	–	–	–	–	–
CCDC40	Exonic	Exon 10	c.G1459A	p.A487T	Nonsynsonymous SNV	2	1	0.0005	rs185157579	–	–	0.45	T	0.522	P
CCDC40	Exonic	Exon 15	c.C2551T	p.R851W	Nonsynsonymous SNV	1	0	1.66E-05	–	–	–	0.02	D	1	D
CCDC40	Exonic	Exon 7	c.G1144A	p.E382K	Nonsynsonymous SNV	7	1	0.0025	rs2289532	–	–	0.15	T	0.976	D
CCDC40	Exonic	Exon 9	c.G1372T	p.A458S	Nonsynsonymous SNV	1	0	1.66E-05	–	–	–	0	D	0.866	P
CCDC40	Exonic	Exon 9	c.C1373T	p.A458V	Nonsynsonymous SNV	1	0	1.66E-05	–	–	–	0	D	0.998	D
CCDC40	Intronic	–	–	–	–	1	0	0	–	–	–	–	–	–	–
CCDC40	Exonic	Exon 7	c.G962A	p.R321Q	Nonsynsonymous SNV	1	1	5.01E-05	rs114740534	–	–	0.08	T	0.517	P
DRC1	Exonic	Exon 12	c.A1519C	p.I507L	Nonsynsonymous SNV	4	0	0.0005	rs188055173	–	–	0.19	T	0.916	P
DRC1	Exonic	Exon 9	c.G1124A	p.R375H	Nonsynsonymous SNV	4	0	0.0004	rs141347582	–	–	0.14	T	1	D
DRC1	Exonic	Exon 3	c.G350A	p.R117H	Nonsynsonymous SNV	2	2	1.00E-04	rs200567817	–	–	0.03	D	0.846	P
DRC1	Exonic	Exon 10	c.C1288T	p.L430F	Nonsynsonymous SNV	1	0	4.95E-05	rs568154812	–	–	0.14	T	0.999	D
DRC1	Exonic	Exon 13	c.A1622G	p.D541G	Nonsynsonymous SNV	1	2	9.06E-05	–	–	–	0.02	D	0.726	P
DRC1	Exonic	Exon 12	c.C1598T	p.S533F	Nonsynsonymous SNV	4	1	0.0002	–	–	–	0.08	T	0.983	D
NME8	Exonic	Exon 8	c.A416T	p.D139V	Nonsynsonymous SNV	1	0	6.61E-05	–	–	–	0.22	T	0.655	P
NME8	Exonic	Exon 6	c.T224C	p.L75S	Nonsynsonymous SNV	3	3	0.0002	rs541494396	–	–	0	D	1	D
NME8	Exonic	Exon 13	c.T1121G	p.L374R	Nonsynsonymous SNV	1	0	8.32E-06	–	–	–	0	D	1	D
NME8	Exonic	Exon 17	c.T1637A	p.L546X	Stopgain	1	0	8.25E-06	rs199902263	–	–	0.38	T	–	–
RPGR	Exonic	Exon 11	c.C1282G	p.L428V	Nonsynsonymous SNV	1	0	0.0001	rs182345461	–	–	0.55	T	0.99	D
RPGR	Exonic	Exon 6	c.G518A	p.G173D	Nonsynsonymous SNV	1	0	–	–	–	–	0	D	1	D
RSPH9	Exonic	Exon 3	c.G467A	p.R156Q	Nonsynsonymous SNV	3	1	0.0006	rs201032669	–	–	0.21	T	0.977	D
RSPH9	Exonic	Exon 6	c.G787A	p.G263R	Nonsynsonymous SNV	1	1	9.08E-05	–	–	–	0.03	D	0.006	B
RSPH9	Exonic	Exon 6	c.C871T	p.L291F	Nonsynsonymous SNV	1	0	0.0006	rs79412180	–	–	0	D	0.047	B
RSPH9	Exonic	Exon 1	c.G128A	p.R43Q	Nonsynsonymous SNV	1	0	3.36E-05	–	–	–	0.39	T	0.722	P
DNAAF1	Exonic	Exon 11	c.G1750A	p.D584N	Nonsynsonymous SNV	3	2	0.0022	rs142345677	–	–	0.01	D	0.998	D
DNAAF1	Exonic	Exon 7	c.A907G	p.K303E	Nonsynsonymous SNV	2	0	–	–	–	–	0.06	T	0.994	D
DNAAF1	Exonic	Exon 11	c.C2063T	p.P688L	Nonsynsonymous SNV	1	0	6.74E-05	rs149659582	–	–	0.01	D	0.023	B
DNAAF2	Exonic	Exon 1	c.C449T	p.A150V	Nonsynsonymous SNV	1	0	–	–	–	–	0.53	T	0.972	D
DNAAF2	Exonic	Exon 1	c.T458C	p.L153P	Nonsynsonymous SNV	1	0	–	–	–	–	0.22	T	1	D
DNAAF2	Exonic	Exon 2	c.C2237T	p.T746M	Nonsynsonymous SNV	1	1	9.07E-05	rs185361306	–	–	0	D	0.895	P
DNAAF2	Exonic	Exon 2	c.G2036T	p.C679F	Nonsynsonymous SNV	1	0	4.97E-05	–	–	–	0.01	D	0.003	B
DNAAF2	Exonic	Exon 2	c.G2059T	p.V687F	Nonsynsonymous SNV	1	0	8.28E-06	rs553179519	–	–	0.02	D	0.834	P
DNAAF2	Exonic	Exon 1	c.G1273A	p.A425T	Nonsynsonymous SNV	1	1	0.0002	–	–	–	0.49	T	0.573	P
DNAAF3	Exonic	Exon 3	c.G298T	p.V100L	Nonsynsonymous SNV	1	0	–	–	–	–	0.27	T	0.99	D
DNAAF3	Exonic	Exon 3	c.T28A	p.S10T	Nonsynsonymous SNV	2	0	–	–	–	–	0.02	D	0.718	P
DNAAF3	Exonic	Exon 5	c.G547T	p.E183X	Stopgain	1	0	–	–	–	–	0.01	D	–	–
DNAH11	Exonic	Exon 82	c.C13309T	p.R4437C	Nonsynsonymous SNV	2	0	0.0004	rs200466467	–	–	0.09	T	1	D
DNAH11	Exonic	Exon 60	c.G9745A	p.D3249N	Nonsynsonymous SNV	1	0	1.68E-05	rs537824201	–	–	0.09	T	1	D
DNAH11	Exonic	Exon 4	c.G846C	p.M282I	Nonsynsonymous SNV	1	0	8.27E-05	rs375023124	–	–	0.04	D	0.005	B
DNAH11	Exonic	Exon 15	c.A2912G	p.D971G	Nonsynsonymous SNV	8	2	0.0009	rs147700251	–	–	0.17	T	0.518	P
DNAH11	Exonic	Exon 14	c.G2419C	p.D807H	Nonsynsonymous SNV	3	2	0.0004	rs369849556	–	–	0.01	D	0.984	D
DNAH11	Exonic	Exon 75	c.C12258A	p.Y4086X	Stopgain	1	0	–	–	–	–	0.82	T	–	–
DNAH11	Exonic	Exon 14	c.G2542A	p.V848M	Nonsynsonymous SNV	8	4	0.0019	rs80328282	–	–	0.14	T	0.533	P
DNAH11	Exonic	Exon 46	c.G7444A	p.V2482I	Nonsynsonymous SNV	1	0	–	rs545144577	–	–	1	T	0.623	P
DNAH11	Exonic	Exon 20	c.G3791T	p.R1264I	Nonsynsonymous SNV	1	0	–	–	–	–	0.09	T	0.904	P
DNAH11	Exonic	Exon 56	c.A9260G	p.K3087R	Nonsynsonymous SNV	14	6	–	–	–	–	0.28	T	0.991	D
DNAH11	Exonic	Exon 27	c.A4805G	p.D1602G	Nonsynsonymous SNV	1	0	3.20E-05	rs548213797	–	–	0.37	T	0.925	P
DNAH11	Exonic	Exon 45	c.G7292T	p.S2431I	Nonsynsonymous SNV	3	0	9.78E-05	rs185039462	–	–	0.14	T	0.999	D
DNAH11	Exonic	Exon 25	c.G4394T	p.S1465I	Nonsynsonymous SNV	1	0	8.30E-06	–	–	–	0.24	T	0.934	P
DNAH11	Exonic	Exon 55	c.C9017T	p.T3006M	Nonsynsonymous SNV	2	0	6.63E-05	rs537395849	–	–	0.02	D	1	D
DNAH11	Exonic	Exon 42	c.C6983T	p.P2328L	Nonsynsonymous SNV	2	0	0.0002	–	–	–	0	D	0.372	B
DNAH11	Exonic	Exon 55	c.C8968T	p.R2990C	Nonsynsonymous SNV	1	0	8.28E-06	–	–	–	0.01	D	0	B
DNAH11	Exonic	Exon 15	c.A2750T	p.E917V	Nonsynsonymous SNV	2	0	0.0003	rs543944909	–	–	0.02	D	0.651	P
DNAH11	Exonic	Exon 24	c.C4306T	p.R1436W	Nonsynsonymous SNV	3	3	0.0007	rs183489539	–	–	0.01	D	0.964	D
DNAH11	Exonic	Exon 36	c.C6118T	p.R2040C	Nonsynsonymous SNV	1	0	6.69E-05	rs199772877	–	–	0	D	1	D
DNAH11	Exonic	Exon 72	c.C11804T	p.P3935L	Nonsynsonymous SNV	1	0	0.0005	rs72658814	–	–	0.05	D	1	D
DNAH11	Exonic	Exon 32	c.C5506T	p.R1836X	Stopgain	1	0	–	–	–	–	0.78	T	–	–
DNAH11	Exonic	Exon 81	c.C13175T	p.T4392M	Nonsynsonymous SNV	2	0	0.0002	rs181603431	–	–	0.07	T	1	D
DNAH11	Exonic	Exon 25	c.A4436G	p.Y1479C	Nonsynsonymous SNV	1	0	8.30E-06	–	–	–	0.01	D	0.99	D
DNAH11	Exonic	Exon 70	c.T11396C	p.I3799T	Nonsynsonymous SNV	1	0	2.49E-05	–	–	–	0	D	0.998	D
DNAH11	Exonic	Exon 74	c.C12071T	p.S4024F	Nonsynsonymous SNV	1	0	–	–	–	–	0	D	0.915	P
DNAH11	Exonic	Exon 71	c.C11647T	p.L3883F	Nonsynsonymous SNV	1	1	9.18E-05	–	–	–	0.02	D	0.001	B
DNAH11	Exonic	Exon 71	c.A11624G	p.K3875R	Nonsynsonymous SNV	1	0	–	–	–	–	0.37	T	0.999	D
DNAH11	Exonic	Exon 55	c.A9022G	p.I3008V	Nonsynsonymous SNV	1	0	0.0001	rs369862935	–	–	0.03	D	0.018	B
DNAH11	Exonic	Exon 82	c.C13373T	p.P4458L	Nonsynsonymous SNV	4	0	0.0001	rs72658835	–	–	0	D	1	D
DNAH11	Exonic	Exon 42	c.A6905C	p.H2302P	Nonsynsonymous SNV	1	0	–	–	–	–	0.01	D	1	D
DNAH11	Exonic	Exon 80	c.C13112T	p.P4371L	Nonsynsonymous SNV	1	0	–	–	–	–	0.01	D	1	D
DNAH11	Exonic	Exon 28	c.C4898A	p.S1633Y	Nonsynsonymous SNV	1	0	–	–	–	–	0.01	D	0.983	D
DNAH11	Exonic	Exon 35	c.T5974C	p.F1992L	Nonsynsonymous SNV	1	0	4.14E-05	–	–	–	0	D	0.999	D
DNAH11	Exonic	Exon 77	c.C12595T	p.L4199F	Nonsynsonymous SNV	1	0	–	–	–	–	0.16	T	0.998	D
DNAH11	Exonic	Exon 60	c.C9790T	p.P3264S	Nonsynsonymous SNV	1	0	7.48E-05	rs571709285	–	–	0.91	T	0.998	D
DNAH11	Exonic	Exon 7	c.T1300C	p.F434L	Nonsynsonymous SNV	1	0	–	–	–	–	0.04	D	0.992	D
DNAH5	Exonic	Exon 70	c.C11986T	p.R3996C	Nonsynsonymous SNV	1	0	–	–	–	–	0	D	0.997	D
DNAH5	Exonic	Exon 28	c.G4432A	p.E1478K	Nonsynsonymous SNV	1	0	–	–	–	–	0.05	D	0.997	D
DNAH5	Exonic	Exon 61	c.G10307A	p.R3436H	Nonsynsonymous SNV	1	0	8.24E-06	–	–	–	0.01	D	0.996	D
DNAH5	Exonic	Exon 17	c.G2480A	p.R827H	Nonsynsonymous SNV	1	0	4.97E-05	rs141969815	–	–	0.2	T	0.98	D
DNAH5	Exonic	Exon 14	c.C2047T	p.R683W	Nonsynsonymous SNV	2	1	0.0004	rs182512638	–	–	0.01	D	0.924	P
DNAH5	Exonic	Exon 7	c.C894G	p.N298K	Nonsynsonymous SNV	4	3	0.001	rs116995413	–	–	0.16	T	0.454	P
DNAH5	Exonic	Exon 34	c.C5518T	p.R1840W	Nonsynsonymous SNV	1	0	2.48E-05	.	–	–	0.14	T	0.798	P
DNAH5	Intronic	–	–	–	–	2	1	0.0005	rs151000177	–	–	–	–	–	–
DNAH5	Exonic	Exon 72	c.C12367T	p.H4123Y	Nonsynsonymous SNV	3	3	0.0007	rs151145750	–	–	0	D	0.888	P
DNAH5	Exonic	Exon 27	c.G4139A	p.R1380Q	Nonsynsonymous SNV	1	0	0.0001	–	–	–	0.05	D	0.992	D
DNAH5	Exonic	Exon 24	c.C3733T	p.R1245C	Nonsynsonymous SNV	1	3	0.0002	rs149609746	–	–	0	D	1	D
DNAH5	Exonic	Exon 19	c.C2980T	p.R994W	Nonsynsonymous SNV	1	0	0.0002	rs560275492	–	–	0.18	T	0.738	P
DNAH5	Exonic	Exon 39	c.C6556T	p.R2186W	Nonsynsonymous SNV	1	1	4.94E-05	–	–	–	0	D	1	D
DNAH5	Exonic	Exon 41	c.A6835G	p.M2279V	Nonsynsonymous SNV	1	0	–	–	–	–	0.05	D	0.155	B
DNAH5	Exonic	Exon 38	c.A6428T	p.E2143V	Nonsynsonymous SNV	1	0	1.65E-05	–	–	–	0	D	0.999	D
DNAH5	Exonic	Exon 31	c.G4961A	p.R1654Q	Nonsynsonymous SNV	1	0	0.0003	rs150868941	–	–	0.01	D	0.916	P
DNAH5	Exonic	Exon 11	c.A1421G	p.E474G	Nonsynsonymous SNV	1	0	–	–	–	–	0.05	D	0.005	B
DNAH5	Exonic	Exon 41	c.A6790G	p.S2264G	Nonsynsonymous SNV	1	0	3.30E-05	–	–	–	0.01	D	0.958	D
DNAH5	Exonic	Exon 75	c.G13073A	p.R4358Q	Nonsynsonymous SNV	2	0	0.0005	rs144949095	–	–	0.38	T	0.61	P
DNAH5	Exonic	Exon 67	c.A11569G	p.R3857G	Nonsynsonymous SNV	2	0	–	–	–	–	0.01	D	0.935	P
DNAH5	Exonic	Exon 48	c.C7892T	p.T2631M	Nonsynsonymous SNV	1	0	0.0002	rs375200875	–	–	0.01	D	1	D
DNAH5	Intronic	–	–	–	–	1	0	9.66E-05	rs200608915	–	–	–	–	–	–
DNAH5	Exonic	Exon 5	c.C586T	p.R196C	Nonsynsonymous SNV	1	0	1.65E-05	rs200314274	–	–	0	D	0.972	D
DNAI1	Exonic	Exon 11	c.G1028T	p.C343F	Nonsynsonymous SNV	1	0	2.48E-05	–	–	–	0.02	D	0.577	P
DNAI1	Exonic	Exon 19	c.A2011G	p.K671E	Nonsynsonymous SNV	1	0	0.0002	rs560719255	–	–	0.07	T	0.79	P
DNAI2	Exonic	Exon 7	c.T773C	p.I258T	Nonsynsonymous SNV	1	0	3.32E-05	rs560319154	–	–	0.51	T	0.65	P
DNAI2	Exonic	Exon 2	c.G124A	p.V42M	Nonsynsonymous SNV	4	3	0.002	rs151176313	–	–	0.05	D	0.13	B
DNAI2	Exonic	Exon 5	c.G535T	p.A179S	Nonsynsonymous SNV	1	0	3.30E-05	–	–	–	0.05	D	1	D
HEATR2	Exonic	Exon 6	c.G1394A	p.R465Q	Nonsynsonymous SNV	8	0	0.0008	rs192289643	–	–	0.58	T	0.543	P
HEATR2	Exonic	Exon 3	c.C787T	p.R263W	Nonsynsonymous SNV	2	0	0.0005	rs143045012	–	–	0.02	D	0.94	P
HEATR2	Exonic	Exon 8	c.C1686G	p.H562Q	Nonsynsonymous SNV	1	0	9.13E-05	rs201303241	–	–	0.02	D	0.982	D
HEATR2	Exonic	Exon 3	c.G811T	p.G271C	Nonsynsonymous SNV	1	0	–	–	–	–	0.09	T	0.999	D
HEATR2	Exonic	Exon 6	c.C1325T	p.S442L	Nonsynsonymous SNV	1	0	0.0003	rs146938307	–	–	0.09	T	0.837	P
HEATR2	Exonic	Exon 6	c.C1381T	p.R461W	Nonsynsonymous SNV	1	0	3.34E-05	rs373336873	–	–	0.01	D	1	D
HEATR2	Exonic	Exon 6	c.G1382A	p.R461Q	Nonsynsonymous SNV	1	0	3.34E-05	–	–	–	0.36	T	0.605	P
LRRC6	Exonic	Exon 2	c.G131A	p.R44Q	Nonsynsonymous SNV	7	3	0.0011	rs184555568	–	–	0.04	D	1	D
LRRC6	Exonic	Exon 7	c.C863A	p.P288H	Nonsynsonymous SNV	6	6	0.0021	rs76147813	–	–	0.03	D	0.986	D
LRRC6	Exonic	Exon 9	c.A1037G	p.K346R	Nonsynsonymous SNV	1	0	2.48E-05	rs200879838	–	–	0	D	0.998	D
EFEMP2	Exonic	Exon 8	c.G781A	p.E261K	Nonsynsonymous SNV	1	0	7.43E-05	rs200340088	–	–	0.19	T	0.983	D
ELN	Exonic	Exon 26	c.G1771T	p.A591S	Nonsynsonymous SNV	1	0	–	–	–	–	0.08	T	0.998	D
ELN	Exonic	Exon 14	c.C806T	p.A269V	Nonsynsonymous SNV	1	1	4.33E-05	–	–	–	0.02	D	0.996	D
ELN	Exonic	Exon 9	c.G455T	p.G152V	Nonsynsonymous SNV	1	3	–	–	–	–	0.08	T	1	D
ELN	Exonic	Exon 22	c.G1525A	p.A509T	Nonsynsonymous SNV	1	0	–	–	–	–	0.47	T	0.931	P
FBLN5	Exonic	Exon 11	c.T1229C	p.I410T	Nonsynsonymous SNV	2	0	9.89E-05	rs201297086	–	–	0	D	0.763	P
FLCN	Exonic	Exon 4	c.G190C	p.A64P	Nonsynsonymous SNV	1	0	–	–	–	–	0.19	T	0.999	D
FLCN	Exonic	Exon 5	c.C256T	p.R86W	Nonsynsonymous SNV	1	0	–	–	–	–	0	D	1	D
FLCN	Exonic	Exon 14	c.G1580A	p.R527Q	Nonsynsonymous SNV	1	0	3.30E-05	–	–	–	0.13	T	0.998	D
LTBP4	Exonic	–	–	–	Unknown	1	0	0.0002	rs376368630	–	–	0.04	D	1	D
LTBP4	Exonic	–	–	–	Unknown	1	1	0.0004	rs187093021	–	–	0.06	T	0.999	D
LTBP4	Exonic	–	–	–	Unknown	1	0	.	–	–	–	0.26	T	0.998	D
LTBP4	Exonic	–	–	–	Unknown	1	1	9.30E-05	rs574587568	–	–	0.1	T	1	D
SLC34A2	Exonic	Exon 2	c.76_84del	p.26_28del	Nonframeshift deletion	1	1	3.30E-05	–	–	–	–	–	–	–
SLC34A2	Exonic	Exon 12	c.G1339A	p.V447M	Nonsynsonymous SNV	2	0	8.24E-05	–	–	–	0.06	T	1	D
SLC34A2	Exonic	Exon 6	c.C616T	p.R206W	Nonsynsonymous SNV	1	0	1.67E-05	–	–	–	0	D	1	D
SLC34A2	Exonic	Exon 11	c.G1285A	p.V429I	Nonsynsonymous SNV	1	1	0.0002	rs201539404	–	–	0.11	T	0.99	D
SLC34A2	Exonic	Exon 11	c.T1259A	p.L420H	Nonsynsonymous SNV	1	0	2.47E-05	–	–	–	0	D	0.997	D
ATP7A	Exonic	Exon 5	c.T1409G	p.F470C	Nonsynsonymous SNV	1	0	–	–	–	–	0.12	T	0.808	P

T, tolerated; D, damaging; B, benign; P, probably damaging.