Table S1. Exome sequencing capture statistics

Sample ID	Total Reads	Valid Reads	Mapped %	Duplicates %	On target (%)	Data in Gb	Bases at 1x (%)	Bases at 2x (%)	Base at 10x (%)	Bases at 20x (%)	Bases at 30X (%)	Mean read length	Average depth
DM1564	61424022	60309910	99	24	98	11	98	98	96	93	87	149	79
DM1565	65139198	64353052	99	24	98	10	98	98	97	94	90	148	96
DM1566	80083656	79151770	99	28	98	11	98	98	97	95	93	148	119
DM1567	88207890	87171118	99	25	98	12	98	98	97	96	94	147	133
DM1568	75949440	75109492	99	24	98	11	98	98	97	95	92	148	113
DM1569	74060974	73188336	99	22	98	11	98	98	97	95	91	147	113
DM1570	78624330	77705910	99	24	98	11	98	98	97	95	92	148	116
DM1571	69445464	68626320	99	24	98	11	98	98	96	94	90	147	102
DM1572	72728660	71932064	99	26	98	11	98	98	97	94	91	147	110
DM1573	79939072	76768058	99	26	98	12	98	98	97	95	92	148	110
DM1574	75651366	74315542	99	24	98	13	98	98	97	95	91	149	100
DM1575	87235650	86126730	99	25	98	13	98	98	97	96	93	148	127
DM1576	67711558	66934712	99	23	98	10	98	98	96	94	88	146	107
DM1582	61417518	60694476	99	24	98	10	98	98	96	94	88	147	92
DM1583	62764800	62029422	99	22	98	10	98	98	96	94	88	147	96
DM1584	80852010	79863346	99	25	98	12	98	98	97	95	92	147	123
DM1585	80057214	78861164	99	24	98	13	98	98	97	95	93	148	110
DM1586	79610260	78615232	99	24	98	11	98	98	97	95	92	148	119
DM1587	94123112	93026136	99	27	98	13	98	98	97	96	94	147	140
DM1588	60401114	58916874	99	25	97	11	98	98	96	93	86	149	74
DM1589	63337038	62591684	99	22	98	7	98	98	96	93	87	146	101
DM1590	41822538	41281306	99	24	98	7	98	97	94	82	64	137	61
DM1591	77566650	76325094	99	26	98	12	98	98	97	95	92	148	107

Table S2. Causal and candidate Bardet-Biedl syndrome genes

No	Gene identifier	Alias symbols	HGNC ID	OMIM number	Chromosomal location	Complex/subcellular localization	Reference
1	**BBS1**	FLJ23590	HGNC:966	209901	11q13.2	BBSome	[1]
2	**BBS2**	RP74	HGNC:967	606151	16q13	BBSome	[2]
3	**ARL6**	BBS3, RP55	HGNC:13210	608845	3q11.2	ARL, ARF GTPase family	[3] [4]
4	**BBS4**		HGNC:969	600374	15q24.1	BBSome	[5]
5	**BBS5**	DKFZp762I194	HGNC:970	603650	2q31.1	BBSome	[6]
6	**MKKS**	BBS6, HMCS, MKS, KMS	HGNC:7108	604896	20p12.2	Chaperonins	[7] [8]
7	**BBS7**	FLJ10715, BBS2L1	HGNC:18758	607590	4q27	BBSome	[9]
8	**TTC8**	BBS8; RP51	HGNC:20087	608132	14q31.3	BBSome	[10]
9	**BBS9**	B1; PTHB1	HGNC:30000	607968	7p14.3	BBSome	[11]
10	**BBS10**	C12orf58; FLJ23560	HGNC:26291	610148	12q21.2	Chaperonins	[12]
11	**TRIM32**	LGMD2H; HT2A; TATIP; BBS11	HGNC:16380	602290	9q33.1	Cilium base	[13]
12	**BBS12**	C4orf24; FLJ35630; FLJ41559	HGNC:26648	610683	4q27	Chaperonins	[14]
13	**MKS1**	BBS13; MKS; FLJ20345; POC12	HGNC:7121	609883	17q22	Transition Zone-MKS	[15]
14	**CEP290**	BBS14; KIAA0373; FLJ13615; 3H11Ag; rd16; NPHP6; JBTS5; SLSN6; LCA10; MKS4; CT87; POC3	HGNC:29021	610142	12q21.32	Transition Zone-MKS/NPHP	[15]
15	**WDPCP**	BBS15; C2orf86; hFrtz; fritz; CPLANE5	HGNC:28027	613580	2p15	Basal body	[16]
16	**SDCCAG8**	BBS16; NY-CO-8; CCCAP; SLSN7; NPHP10	HGNC:10671	613524	1q43-q44	Transition Zone-OFD1	[17] [18]
17	**LZTFL1**	BBS17	HGNC:6741	606568	3p21.31	BBSome regulator	[19]
18	**BBIP1**	BBS18; bA348N5.3; BBIP10	HGNC:28093	613605	10q25.2	BBSome assembly	[20]
19	**IFT27**	BBS19; RAYL; FAP156	HGNC:18626	615870	22q12.3	IFT-B1 complex	[21]
20	**IFT172**	BBS20; SLB; wim; osm-1; NPHP17	HGNC:30391	607386	2p23.3	IFT-B2 complex	[22] [23]
21	**CFAP418**	BBS21; C8orf37; FLJ30600; CORD16; RP64; FAP418; MOT25	HGNC:27232	614477	8q22.1	Cilium base	[24] [25]

22	**IFT74**	BBS22; CMG1; CMG-1; FLJ22621	HGNC:21424	608040	9p21.2	IFT-B1 complex	[26]
23	**CEP19**	BBS23; C3orf34; MGC14126	HGNC:28209	615586	3q29	?	[27]
24	**SCAPER**	BBS24; ZNF291; Zfp291	HGNC:13081	611611	15q24.3	Cilium tip	[28]
25	**CEP164**	KIAA1052; NPHP15	HGNC:29182		11q23.3	Distal appendage	[29]
26	**SCLT1**	hCAP-1A; FLJ30655	HGNC:26406		4q28.2	Distal appendage	[30]
27	**TTC21B**	FLJ11457; JBTS11; NPHP12; IFT139B; THM1; FAP60; FLA17; IFT139	HGNC:25660		2q24.3	IFT-A complex	[31]
28	**NPHP1**	NPH1; JBTS4; SLSN1	HGNC:7905	607100	2q13	Transition Zone-NPHP	[32]

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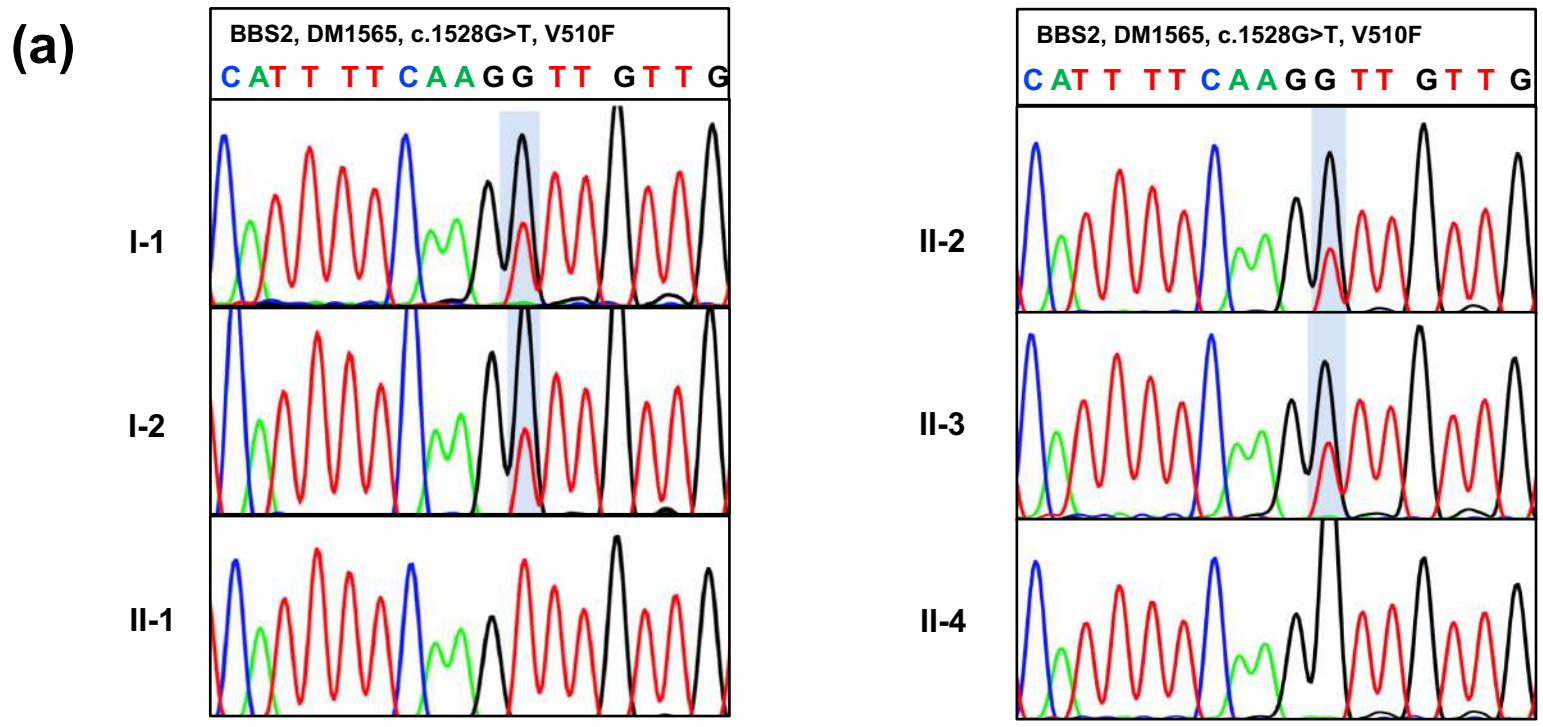
Table S3: List of rare variants identified and their *in silico* predicted pathogenicity scores

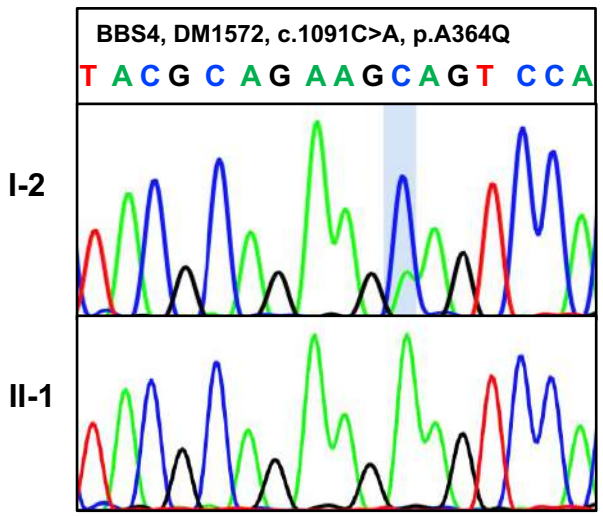
Family ID	BBS gene	Transcript ID	Nucleotide change	Amino acid change	SIFT (Version 6.2.1)	Provean (Version 1.1)	MutationTaster 2021	CADD (version 1.6)
DM1565	*BBS2*	NM_031885.5	c.1528G>T (H)	p.Val510Phe	Damaging	Damaging	Disease causing	29.3
DM1566	*BBS4*	NM_033028.5	c.332+8T>C (h)	p.Ala74Aspfs*7	-	-	Polymorphism	8.67
DM1567	*BBS7*	NM_176824.3	c.712_715del (h)	p.Arg238Glufs*59	-	-	-	-
DM1569	*BBS12*	NM_152618.3	c.1063C>T (H)	p.Arg355*	Damaging	-	Disease causing	35
DM1570	*BBS5*	NM_152384.3	c.619-1G>C (H)	-	-	-	Disease causing	33
DM1571	*BBS7* *BBS7*	NM_176824.3	c.68T>G (h) c.712_715del (h)	p.Leu23Arg p.Arg238Glufs*59	Damaging -	Damaging -	Disease causing -	27.1 -
DM1572	*BBS4*	NM_033028.5	c.1091C>A (H)	p.Ala364Glu	Damaging	Damaging	Disease causing	28.3
DM1573	*BBS12*	NM_152618.3	c.1063C>T (H)	p.Arg355*	Damaging	-	Disease causing	35
DM1574	*BBS10* *BBS10*	NM_024685.4	c.145C>T (h) c.1804G>C (h)	p.Arg49Trp p.Val602Leu	Damaging Damaging	Damaging Neutral	Disease causing Disease causing	31 26.1
	IFT172	NM_015662.3	c.1715G>A (h)	p.Arg572Gln	Damaging	Damaging	Disease causing	28.9
DM1576	*BBS1*	NM_024649.5	c.1169T>G (h)	p.Met390Arg	Damaging	Damaging	Disease causing	25.9
DM1582	*BBS10*	NM_024685.4	c.273C>G (H)	p.Cys91Trp	Damaging	Neutral	Disease causing	24.9
DM1584	*BBS12*	NM_152618.3	c.1063C>T (H)	p.Arg355*	Damaging	-	Disease causing	35
DM1585	*BBS12*	NM_152618.3	c.1063C>T (H)	p.Arg355*	Damaging	-	Disease causing	35
DM1587	*BBS12*	NM_152618.3	c.1063C>T (H)	p.Arg355*	Damaging	-	Disease causing	35
DM1588	*BBS12*	NM_152618.3	c.1063C>T (H)	p.Arg355*	Damaging	-	Disease causing	35
	BBS5	NM_152384.3	c.226A>G (h)	p.Ile76Val	Tolerated	Neutral	Disease causing	14.9
DM1589	*BBS12*	NM_152618.3	c.1063C>T (H)	p.Arg355*	Damaging		Disease causing	35
	SCAPER	NM_020843.4	c.2891G>A (h)	p.Arg1098Gln	Tolerated	Damaging	Disease Causing	32
	NPHP1	NM_000272.5	c.1630A>G (h)	p.Met544Val	Tolerated	Neutral	Polymorphism	11.5
DM1590	*BBS7*	NM_176824.3	c.712_715del (H)	p.Arg238Glufs*59	-	-	-	-
DM1591	*BBS12* *BBS12*	NM_152618.3	c.1392_1395del (h) c.1682_1682del (h)	p.Cys464Trpfs*7 p.Glu561Lysfs*10	- -	- -	- -	- -

Abbreviations: H, homozygous; h, heterozygous; SIFT, Sorting Intolerant From Tolerant; CADD, Combined Annotation Dependent Depletion.

Supp-figure 1:



(e)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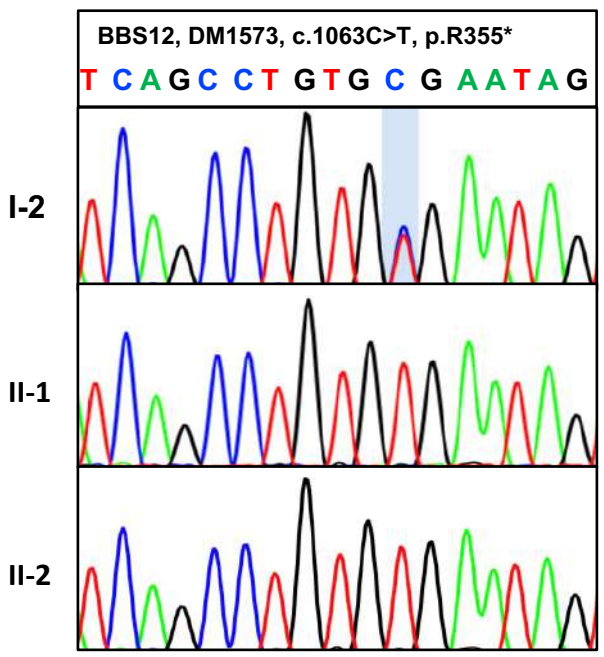
I-2
II-1

(f)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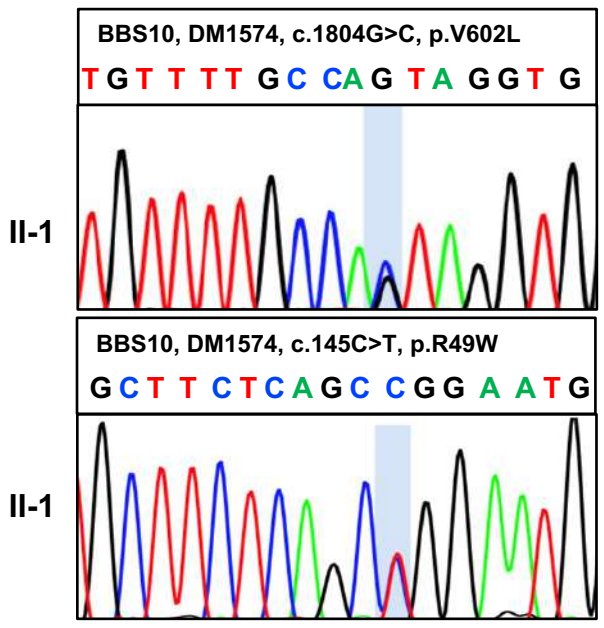
I-2
II-1
II-2

(g)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T G T T T T G C C A G T A G G T G
II-1
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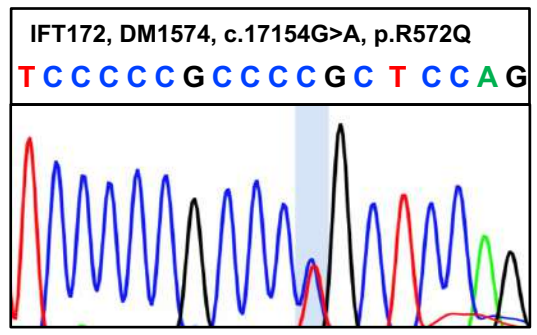
II-1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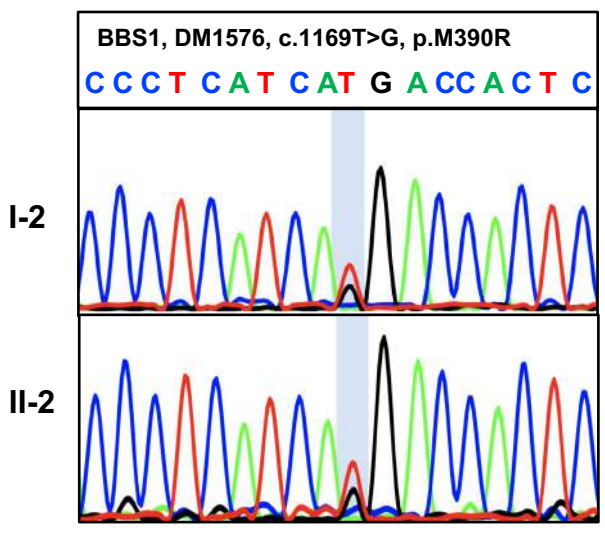
II-1

(h)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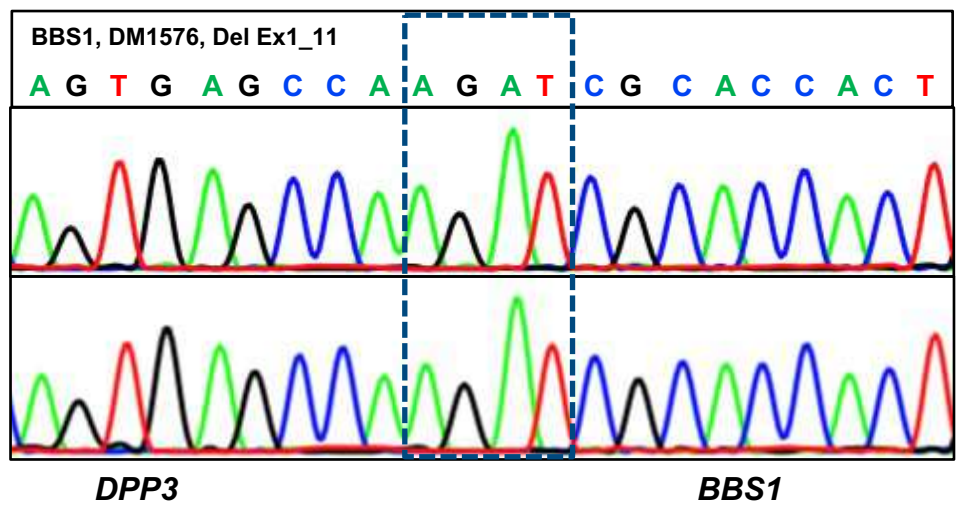
I-2
II-2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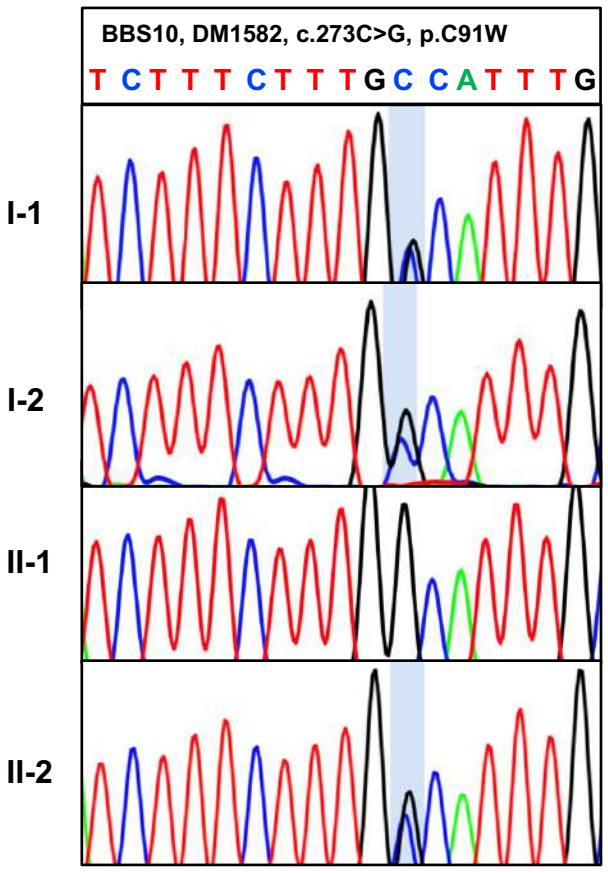
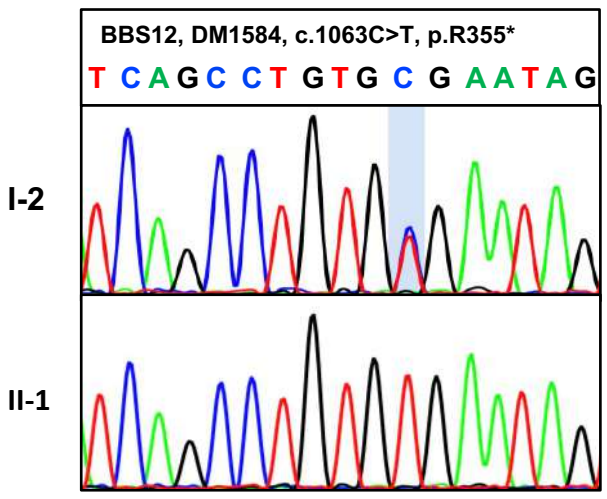
A G T G A G C C A A G A T C G C A C C A C T
I-2
II-2
DPP3
BBS1

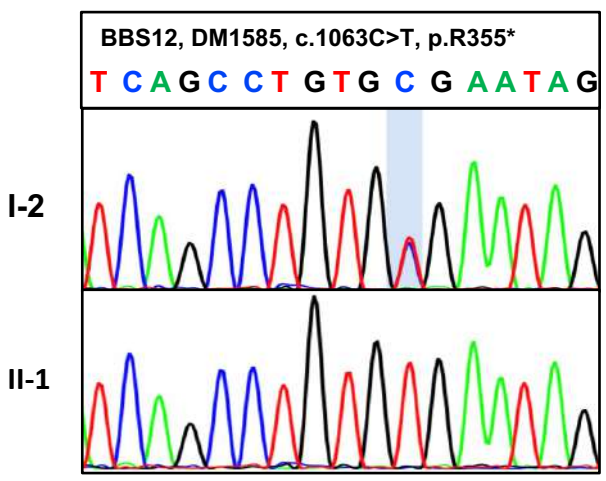
(g)


BBS10, DM1582, c.273C>G, p.C91W
T C T T T C T T T G C C A T T T G
I-1
I-2
II-1
II-2

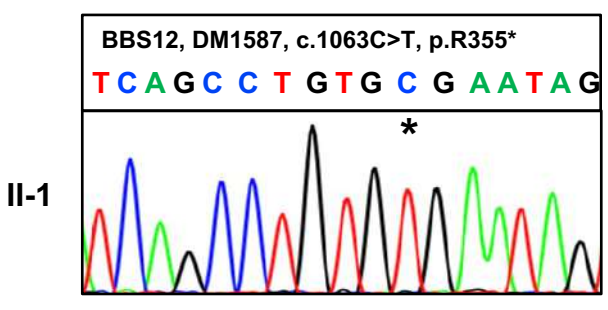
(h)


BBS12, DM1584, c.1063C>T, p.R355*
T C A G C C T G T G C G A A T A G
I-2
II-1

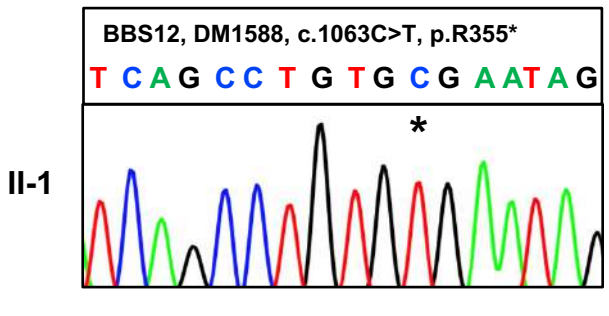
(i)

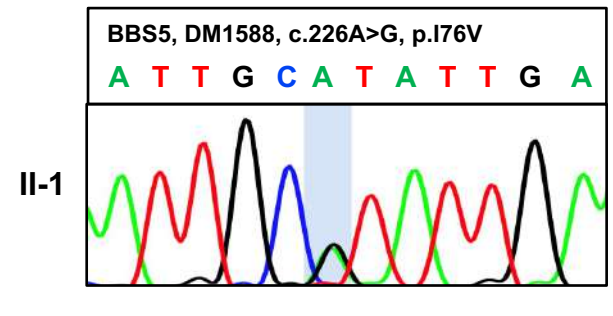

BBS12, DM1585, c.1063C>T, p.R355*
T C A G C C T G T G C G A A T A G
I-2
II-1

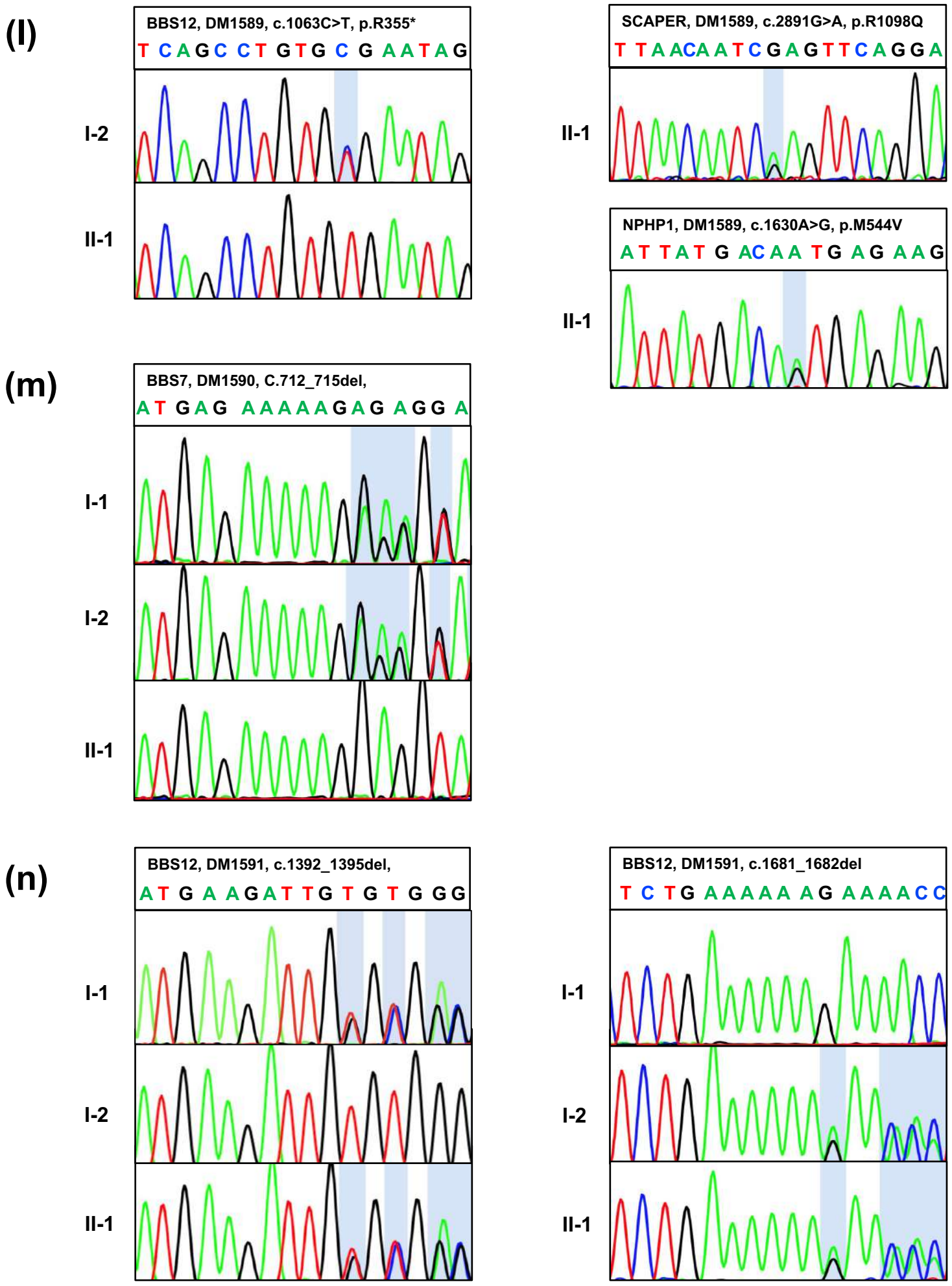
(j)


BBS12, DM1587, c.1063C>T, p.R355*
T C A G C C T G T G C G A A T A G
*
II-1

(k)


BBS12, DM1588, c.1063C>T, p.R355*
T C A G C C T G T G C G A A T A G
*
II-1

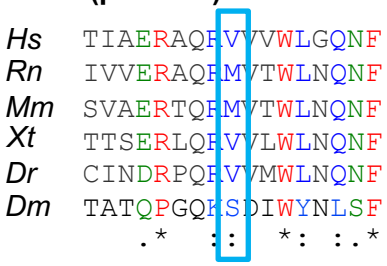

BBS5, DM1588, c.226A>G, p.I76V
A T T G C A T A T T G A
II-1



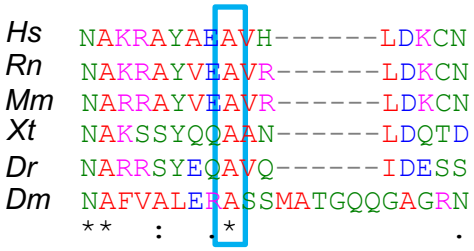
Suppl Figure 1: Chromatograms showing sequence traces of 16 different variants and one CNV identified in this study. Gene name, pedigree identifier, nucleotide substitution or deletions, and amino acid sequence change are mentioned above the sequence traces. Two novel heterozygous variants were identified in *BBS12:* c.1392_1395del; c.1681_1682del. followed by heterozygous allele in *BBS7:* c.68T>G. The position of identified variants is indicated by a light blue shaded line, and sequence chromatograms of the proband were shown below parents' sequence traces. Abbreviations; I-1, father; I-2, mother; II-1, proband.

Supp-figure 2: Multiple protein sequence alignment of regions affected by missense variants.

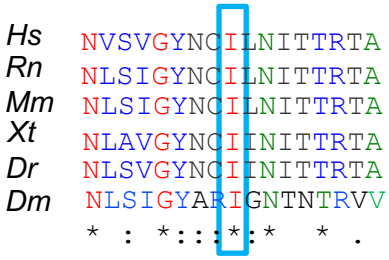
BBS2 (p.V510F)



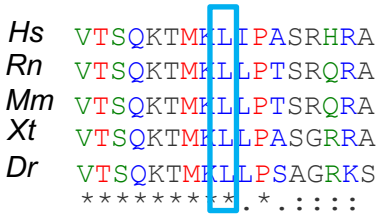
BBS4 (p.A364E)



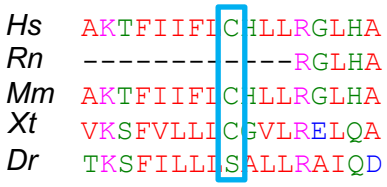
BBS5 (p.I76V)



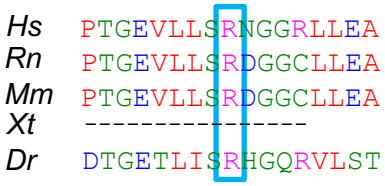
BBS7 (p.L23R)



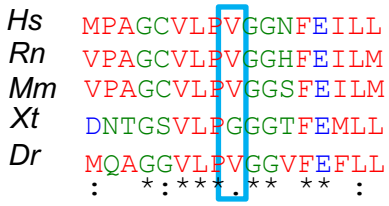
BBS10 (p.C91W)



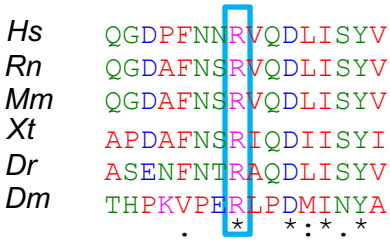
BBS10 (p.R49W)



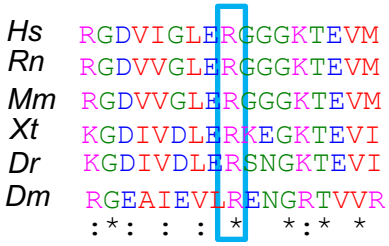
BBS10 (p.V602L)



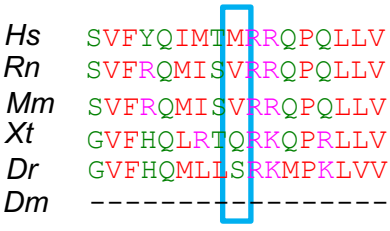
SCAPER (p.R1098E)



IFT172 (p.R572Q)



NPHP1 (p.M544V)



Multiple sequence alignment generated with Clustal Omega shows amino acid conservation around the residues affected by variants in known BBS genes. Blue boxes indicate the position of missense variants identified in this cohort. Asterisk (*) indicates amino acid sequence identity and (:) indicates amino acid similarity. Abbreviations: *HS*, *Homo sapiens*; *Rn, Rattus norvegicus*; *Mm, Mus musculus*; *Xt, Xenopus tropicalis*; *Dr, Danio rerio*; *Dm, Drosophila melanogaster*