

Supplementary Table 1: INS-associated IRDs and their corresponding causative genes

Disease	# of Genes	Gene Names
AR-ACHM	6	<i>ATF7, CNGA3, CNGB3, GNAT2, PDE6C, PDE6H</i>
AR-BBS	25	<i>ADIPOR1, ARL6, BBIP1, BBS1, BBS2, BBS4, BBS5, BBS7, BBS9, BBS10, BBS12, C8orf37, CEP19, CEP290, IFT172, IFT27, INPP5E, KCNJ13, LZTFL1, MKK5, MKS1, NPHP1, SDCCAG8, TRIM32, TTC8</i>
AD-CD/CRD	6	<i>CRX, GUCA1A, GUCY2D, PROM1, PRPH2, RAX2</i>
AR-CD/CRD	16	<i>ABCA4, ADAM9, ALMS1, C21orf2, CACNA2D4, CDHR1, CERKL, CNGA3, CNGB3, IFT81, KCNV2, POC1B, RAB28, RAX2, RPGRI1, TTLL5</i>
XL-CD/CRD	2	<i>CACNA1F, RPGR</i>
AD-CSNB	3	<i>GNAT1, PDE6B, RHO</i>
AR-CSNB	11	<i>CABP4, GNAT1, GNB3, GPR179, GRK1, GRM6, LRIT3, RDH5, SAG, SLC24A1, TRPM1</i>
XL-CSNB	2	<i>CACNA1F, NYX</i>
AD-LCA	3	<i>CRX, IMPDH1, OTX2</i>
AR-LCA	23	<i>AIPL1, CABP4, CCT2, CEP290, CLUAP1, CRB1, CRX, GDF6, GUCY2D, IFT140, INPP5E, IQCB1, KCNJ13, LCA5, LRAT, NMNAT1, PRPH2, RD3, RDH12, RPE65, RPGRIP1, SPATA7, TULP1</i>
AD-Optic atrophy	3	<i>AFG3L2, MFN2, OPA1</i>
AR-Optic atrophy	4	<i>ACO2, NBAS, RTN4IP1, TMEM126A</i>
AD-OA-syndromic	2	<i>ELOVL1, MFN2, NR2F1</i>
AR-OA-syndromic	3	<i>ACO2, C12orf65, OPA3</i>
XL-OA	1	<i>TIMM8A</i>
AD-Other IRDs	13	<i>BEST1, CAPN5, FZD4, ITM2B, LRP5, MAPKAPK3, MIR204, OPN1SW, PAX6, RCBTB1, TEAD1, TSPAN12, ZNF408</i>
AR-Other IRDs	19	<i>BEST1, CYP4V2, LRP5, MFRP, NBAS, NR2E3, NRL, OAT, P3H2, PLA2G5, PROM1, RBP4, RCBTB1, RGS9, RGS9BP, RLBP1, SLC38A8, TSPAN12, VSX2</i>
XL-Other IRDs	5	<i>CHM, NDP, OPN1LW, OPN1MW, RS1</i>
AD-RP	26	<i>ADIPOR1, ARL3, BEST1, CA4, CRX, HK1, IMPDH1, IMPG1, KLHL7, NR2E3, NRL, PRPF3, PRPF4, PRPF6, PRPF8, PRPF31, PRPH2, RDH12, RHO, RP1, RP9, RPE65, SAG, SNRNP200, SPP2, TOPORS</i>
AR-RP	68	<i>ABCA4, AGBL5, AHR, ARHGEF18, ARL6, ARL2BP, ASRGL1, BBS1, BBS2, BEST1, C2orf71, C8orf37, C21orf2, CERKL, CLCC1, CNGA1, CNGB1, CRB1, CYP4V2, DHDDS, DHX38, EMC1, EYS, FAM161A, GPR125, HGSNAT, IDH3A, IDH3B, IFT140, IFT172, IMPG2, KIAA1549, KIZ, LRAT, MAK, MERTK, MVK, NEK2, NEUROD1, NR2E3, NRL, PDE6A, PDE6B, PDE6G, POMGNT1, PRCD, PROM1, RBP3, REEP6, RGR, RHO, RLBP1, RP1, RP1L1, RPE65, SAG, SAMD11, SCAPER, SLC4A7, SLC7A14, SNRNP200, SPATA7, TRNT1, TTC8, TULP1, USH2A, ZNF408, ZNF513</i>
XL-RP	3	<i>OFD1, RP2, RPGR</i>

Supplementary Table 2: INS-associated IRD genes and their corresponding inheritance patterns and phenotypes

Genes HGNC	Transcript name	Inheritance	Disease/phenotype	Disease ID (OMIM#)	Gene ID (OMIM#)
<i>ABCA4</i>	NM_000350	AR	STGD, CRD, maculopathy, RP	248200	601691
<i>ACBD5</i>	NM_145698	AR	CRD- syndromic	618863	616618
<i>ACO2</i>	NM_001098	AR	OA, OA- syndromic	614559	100850
<i>ADAM9</i>	NM_003816	AR	CRD	612775	602713
<i>ADAMTS18</i>	NM_199355	AR	Knobloch, Microcornea	615458	607512
<i>ADGRV1</i>	NM_032119	AR	USH2C	605472	602851
<i>AFG3L2</i>	NM_006796	AD/AR	OA, OA- syndromic	618977	604581
<i>AGBL5</i>	NM_001035507	AR	RP	617023	615900
<i>AHI1</i>	NM_001134830	AR	Joubert syndrome, RP	608629	608894
<i>AIPL1</i>	NM_014336	AR	LCA,CRD	604393	604392
<i>ALMS1</i>	NM_015120	AR	Alström Syndrome, CRD	203800	606844
<i>ARHGEF18</i>	NM_015318	AR	RP	617433	616432
<i>ARL2BP</i>	NM_012106	AR	RP	615434	615407
<i>ARL3</i>	NM_004311	AD	RP	618161	604695
<i>ARL6</i>	NM_001278293	AR	BBS, RP	600151	608845
<i>ARSG</i>	NM_014960	AR	USH	618144	610008
<i>ATF6</i>	NM_007348	AR	ACHM	616517	605537
<i>BBIP1</i>	NM_001243783	AR	BBS	615995	613605
<i>BBS1</i>	NM_024649	AR	BBS, RP	209900	209901
<i>BBS10</i>	NM_024685	AR	BBS	615987	610148
<i>BBS12</i>	NM_152618	AR	BBS	615989	610683
<i>BBS2</i>	NM_031885	AR	BBS, RP	615981	606151
<i>BBS4</i>	NM_033028	AR	BBS	615982	600374
<i>BBS5</i>	NM_152384	AR	BBS	615983	603650
<i>BBS7</i>	NM_176824	AR	BBS	615984	607590
<i>BBS9</i>	NM_001033604	AR	BBS	615986	607968
<i>BEST1</i>	NM_001139443	AR/AD	Best, RP, other RD	611809	607854
<i>C12orf65</i>	NM_152269	AR	OA- syndromic	613559	613541
<i>C5orf42</i>	NM_023073	AR	Joubert Syndrome	614615	614571
<i>C8orf37</i>	NM_177965	AR	BBS, RP	617406	614477
<i>CABP4</i>	NM_145200	AR	CSNB, LCA, CRD	610427	608965

<i>CACNA1F</i>	NM_001256789	XL	CSNB,CRD	300071	300110
<i>CACNA2D4</i>	NM_172364	AR	CRD	610478	608171
<i>CC2D2A</i>	NM_001080522	AR	Joubert Syndrome	216360	612013
<i>CDH23</i>	NM_022124	AR	USH	601067	605516
<i>CDHR1</i>	NM_033100	AR	CRD	613660	609502
<i>CEP164</i>	NM_001271933	AR	Senior Löken Syndrome	614845	614848
<i>CEP250</i>	NM_007186	AR	USH	618358	609689
<i>CEP290</i>	NM_025114	AR	LCA, BBS, RD-syndromic	611755	610142
<i>CEP78</i>	NM_032171	AR	CRD	617236	617110
<i>CERKL</i>	NM_001030311	AR	CRD,RP	608380	608381
<i>CFH</i>	NM_000186	AD	Maculopathy	610698	134370
<i>CLCC1</i>	NM_001048210	AR	RP	609913	617539
<i>CLRN1</i>	NM_001195794	AR	USH	276902	606397
<i>CNGA1</i>	NM_001142564	AR	RP	613756	123825
<i>CNGA3</i>	NM_001298	AR	ACHM, CRD	216900	600053
<i>CNGB1</i>	NM_001297	AR	RP	613767	600724
<i>CNGB3</i>	NM_019098	AR	ACHM, CRD	262300	605080
<i>CNNM4</i>	NM_020184	AR	Jalili Syndrome	217080	607805
<i>COL11A1</i>	NM_001854	AD	Stickler Syndrome	604841	120280
<i>COL18A1</i>	NM_030582	AR	Knobloch Syndrome	267750	120328
<i>COL2A1</i>	NM_001844	AR/AD	Stickler Syndrome	108300	120140
<i>COL9A1</i>	NM_001851	AR	Stickler Syndrome	614134	120210
<i>CRB1</i>	NM_001257965	AR	LCA, RP	613835	604210
<i>CRX</i>	NM_000554	AR/AD	CRD, LCA, RP	120970	602225
<i>CSPP1</i>	NM_024790	AR	Joubert Syndrome	615636	611654
<i>CTNNA1</i>	NM_001903	AD	Maculopathy	608970	116805
<i>CWC27</i>	NM_005869	AR	RP- syndromic	250410	617170
<i>CYP4V2</i>	NM_207352	AR	Bietti, RP	210370	608614
<i>DFNB31</i>	NM_001083885	AR	USH	611383	607928
<i>DHX38</i>	NM_014003	AR	RP	618220	605584
<i>DRAM2</i>	NM_178454	AR	Maculopathy	616502	613360
<i>EFEMP1</i>	NM_001039349	AD	Maculopathy	126600	601548
<i>ELOVL1</i>	NM_001256402	AD	OA- syndromic	618527	611813
<i>ELOVL4</i>	NM_022726	AD	Maculopathy	600110	605512
<i>EMC1</i>	NM_015047	AR	RP	616875	616846
<i>ESPN</i>	NM_031475	AR	USH	609006	606351
<i>EXOSC2</i>	NM_014285	AR	RP- syndromic	617763	602238

<i>EYS</i>	NM_001142800	AR	RP	602772	612424
<i>FAM161A</i>	NM_001201543	AR	RP	606068	613596
<i>FLVCR1</i>	NM_014053	AR	RP- syndromic	609033	609144
<i>GDF6</i>	NM_001001557	AR	LCA	615360	601147
<i>GNAT1</i>	NM_144499	AR/AD	CSNB	616389	139330
<i>GNAT2</i>	NM_005272	AR	ACHM	613856	139340
<i>GNB3</i>	NM_002075	AR	CSNB	617024	139130
<i>GPR179</i>	NM_001004334	AR	CSNB	614565	414515
<i>GRK1</i>	NM_002929	AR	CSNB	613411	180381
<i>GRM6</i>	NM_000843	AR	CSNB	257270	604096
<i>GUCA1A</i>	NM_000409	AD	CRD	602093	600364
<i>GUCY2D</i>	NM_000180	AR/AD	CRD, LCA	204000	600179
<i>HARS</i>	NM_001258042	AR	USH	614504	142810
<i>HGSNAT</i>	NM_152419	AR	RP, RP-syndromic	616544	610453
<i>HK1</i>	NM_033500	AD	RP	617460	142600
<i>HMCN1</i>	NM_031935	AD	Maculopathy	603075	608548
<i>IDH3A</i>	NM_005530	AR	RP	619007	601149
<i>IDH3B</i>	NM_006899	AR	RP	612572	604526
<i>IFT140</i>	NM_014714	AR	LCA, RP, RD- syndromic	266920	614620
<i>IFT172</i>	NM_015662	AR	RP, BBS	616394	607386
<i>IFT27</i>	NM_006860	AR	BBS	615996	615870
<i>IMPDH1</i>	NM_183243	AD	RP,LCA	613837	146690
<i>IMPG1</i>	NM_001563	AR/AD	Maculopathy, RP	616151	602870
<i>IMPG2</i>	NM_016247	AR	RP	613581	607056
<i>INPP5E</i>	NM_019892	AR	BBS, RD-syndromic, LCA	610156	613037
<i>IQCB1</i>	NM_001023570	AR	LCA	609254	609237
<i>ITM2B</i>	NM_021999	AD	RD- other	616079	603904
<i>JAG1</i>	NM_000214	AD	Alagille syndrome	118450	601920
<i>KCNJ13</i>	NM_002242	AR/AD	BBS, LCA, RD- syndromic	614186	603208
<i>KIZ</i>	NM_018474	AR	RP	615780	615757
<i>KLHL7</i>	NM_001172428	AD	RP	612943	611119
<i>LCA5</i>	NM_001122769	AR	LCA	604537	611408
<i>LRAT</i>	NM_004744	AR	LCA, RP	613341	604863
<i>LRIT3</i>	NM_198506	AR	CSNB	615058	615004
<i>LZTFL1</i>	NM_001276379	AR	BBS	615994	606568
<i>MAK</i>	NM_001242957	AR	RP	614181	154235
<i>MERTK</i>	NM_006343	AR	RP	613862	604705

<i>MFN2</i>	NM_014874	AD	OA, OA- syndromic	609260	608507
<i>MFRP</i>	NM_031433	AR	RD-other	611040	606227
<i>MFSD8</i>	NM_001371596	AR	Maculopathy	616170	61124
<i>MIR204</i>	NR_029621	AD	Retinal dystrophy with iris coloboma	616722	610942
<i>MKKS</i>	NM_170784	AR	BBS	605231	604896
<i>MKS1</i>	NM_017777	AR	BBS	615990	609883
<i>MYO7A</i>	NM_001127179	AR	USH1	276900	276903
<i>NBAS</i>	NM_015909	AR	OA, RD-other	614800	608025
<i>NEK2</i>	NM_001204182	AR	RP	615565	604043
<i>NMNAT1</i>	NM_022787	AR	LCA	608553	608700
<i>NPHP1</i>	NM_000272	AR	Joubert	609583	607100
<i>NR2E3</i>	NM_016346	AR/AD	RP, ESCS, CPRD	611131	604485
<i>NR2F1</i>	NM_005654	AD	OA-syndromic	615722	132890
<i>NRL</i>	NM_006177	AR/AD	RP, ESCS	613750	162080
<i>NYX</i>	NM_022567	XL	CSNB	310500	300278
<i>OFD1</i>	NM_001330209	XL	RP, Joubert Syndrome	300424	300170
<i>OPA1</i>	NM_130837	AD	OA	165500	605290
<i>OPA3</i>	NM_001017989	AR	OA-syndromic	165300	606580
<i>OPN1LW</i>	NM_020061	XL	BCM	303700	300822
<i>OPN1MW</i>	NM_000513	XL	BCM	303700	300821
<i>OTX2</i>	NM_021728	AD	LCA, pattern dystrophy	610125	600037
<i>P3H2</i>	NM_018192	AR	High myopia, retinal degeneration	614292	610341
<i>PAX2</i>	NM_003987	AD	renal-coloboma syndrome	120330	167409
<i>PAX6</i>	NM_000280	AD	Aniridia, foveal hypoplasia	136520	607108
<i>PCDH15</i>	NM_033056	AR	USH1	601067	605514
<i>PDE6A</i>	NM_000440	AR	RP	613810	180071
<i>PDE6B</i>	NM_000283	AR/AD	CSNB, RP	613801	180072
<i>PDE6C</i>	NM_006204	AR	ACHM	613093	600827
<i>PDE6G</i>	NM_002602	AR	RP	613582	180073
<i>PDE6H</i>	NM_006205	AR	ACHM	610024	601190
<i>PLA2G5</i>	NM_000929	AR	LORD	228980	601192
<i>PLK4</i>	NM_014264	AR	RD- syndromic	616171	605031
<i>POC1B</i>	NM_172240	AR	CRD,RD-syndromic	615973	614784
<i>PROM1</i>	NM_001145849	AR/AD	CRD, maculopathy, RP	612657	604365

<i>PRPF3</i>	NM_004698	AD	RP	601414	607301
<i>PRPF31</i>	NM_015629	AD	RP	600138	606419
<i>PRPF4</i>	NM_004697	AD	RP	615922	607795
<i>PRPH2</i>	NM_000322	AR/AD	RP, CRD, LCA	608133	179605
<i>PRPS1</i>	NM_002764	XL	RD- syndromic	311070	311850
<i>RAB28</i>	NM_001017979	AR	CRD	615374	612994
<i>RAX2</i>	NM_032753	AD	CRD	610381	610362
<i>RBP3</i>	NM_002900	AR	RP	268000	180290
<i>RCBTB1</i>	NM_018191	AR	RD- other	617175	607867
<i>RD3</i>	NM_001033560	AR	LCA	610612	180040
<i>RDH11</i>	NM_001252650	AR	RD- syndromic	616108	607849
<i>RDH12</i>	NM_152443	AR/AD	LCA, RP	612712	608830
<i>RDH5</i>	NM_001199771	AR	FA, CD	136880	601617
<i>RHO</i>	NM_000539	AR/AD	RP, CSNB	613731	180380
<i>RIMS2</i>	NM_001348484	AR	Syndromic CSNB	618970	606630
<i>RLBP1</i>	NM_000326	AR	RP, RPA	607475	180090
<i>ROM1</i>	NM_000327	DIGENIC	RP	608133	180721
<i>RP1</i>	NM_006269	AR/AD	RP	180100	603937
<i>RP1L1</i>	NM_178857	AR/AD	Maculopathy, RP	613587	608581
<i>RP2</i>	NM_006915	XL	RP	312600	300757
<i>RP9</i>	NM_203288	AD	RP	180104	607331
<i>RPE65</i>	NM_000329	AR/AD	LCA, RP	204100	180069
<i>RPGR</i>	NM_000328	XL	CRD, XL- RP,	304020	312610
<i>RPGRIP1</i>	NM_020366	AR	CRD, LCA, RD- syndromic	613826	605446
<i>RPGRIP1L</i>	NM_001127897	AR	Joubert Syndrome	611560	610937
<i>RS1</i>	NM_000330	XL	Retinoschisis	312700	300839
<i>RTN4IP1</i>	NM_032730	AR	OA	616732	610502
<i>SAG</i>	NM_000541	AR/AD	RP,CSNB	613758	181031
<i>SCAPER</i>	NM_020843	AR	RP	618195	611611
<i>SDCCAG8</i>	NM_006642	AR	BBS, RP-syndromic	615993	613524
<i>SLC24A1</i>	NM_004727	AR	CSNB	613830	603617
<i>SLC25A46</i>	NM_138773	AR	RD- syndromic	619303	610826
<i>SLC38A8</i>	NM_001080442	AR	Foveal hypoplasia	609218	615585
<i>SLC7A14</i>	NM_020949	AR	RP	615725	615720
<i>SNRNP200</i>	NM_014014	AD/AR	RP	610359	601664
<i>SPATA7</i>	NM_018418	AR	LCA,RP	604232	609868
<i>TIMP3</i>	NM_000362	AD	Maculopathy	136900	188826
<i>TMEM126A</i>	NM_032273	AR	OA	612989	612988

<i>TMEM216</i>	NM_001173990	AR	Joubert Syndrome	608091	613277
<i>TMEM231</i>	NM_001077416	AR	RP-syndromic	614970	614949
<i>TMEM237</i>	NM_001044385	AR	Joubert Syndrome	614424	614423
<i>TOPORS</i>	NM_005802	AD	RP	609923	609507
<i>TRIM32</i>	NM_012210	AR	BBS	615988	602290
<i>TRNT1</i>	NM_182916	AR	RP,RP-syndromic	616959	612907
<i>TRPM1</i>	NM_001252020	AR	CSNB	613216	603576
<i>TTC8</i>	NM_144596	AR	BBS,RP,CRD	615985	608132
<i>TTLL5</i>	NM_015072	AR	CRD	615860	612268
<i>TUB</i>	NM_177972	AR	RD- syndromic	616188	601197
<i>TULP1</i>	NM_003322	AR	LCA,RP	613843	602280
<i>USH1C</i>	NM_005709	AR	USH1, atypical USH	276904	605242
<i>USH1G</i>	NM_173477	AR	USH1	606943	607696
<i>USH2A</i>	NM_206933	AR	USH2, RP	276901	608400
<i>VCAN</i>	NM_004385	AD	Wagner disease	143200	118661
<i>VSX2</i>	NM_182894	AR	Microphthalmia	610093	142993
<i>WFS1</i>	NM_006005	AR	Wolfram Syndrome	222300	606201
<i>ZNF423</i>	NM_001271620	AR	Joubert Syndrome	614844	604557