Greenwood Diagnostic Labs

Retinal Disease
Genetic Testing

Greenwood Diagnostic Labs
Giving Greater Care

Molecular Genetic Testing

Retinal Disease Panels

Vision loss can have a significant impact on daily activities and an adverse effect on quality of life. While individual forms of inherited vision loss may be rare, they collectively account for a large number of Mendelian diseases with hundreds of genes implicated in retinal disease and vision loss.

The American Academy of Ophthalmology (AAO) Task Force on Genetic Testing recommends that patients with potentially inherited vision loss be offered genetic testing. The AAO Task Force recommends genetic testing in order to improve the accuracy of diagnosis and prognosis, potentially reduce the recurrence risk in family members, and impact specific medical management and treatment. (AAO Task Force on Genetic Testing; Clinical Statement, Feb 2014)

The Greenwood Genetic Center offers 8 comprehensive panels for a variety of clinical presentations including isolated retinal diseases and syndromic presentations such as Waardenburg and Usher syndromes.

	Bardet-Biedl syndrome	Cone-Rod Dystrophy	Congenital Stationary Night Blindness	Leber Congenital Amaurosis	Macular Degeneration	Ocular Albinism & Hermansky-Pudlak syndrome	Optic Atrophy & Early Glaucoma	Retinititis Pigmentosa
Genes	26 Genes	37 Genes	15 Genes	24 Genes	24 Genes	18 Genes	34 Genes	92 Genes
CPT Code	81479	81479	81479	81479	81479	81479	81479	81434
TAT	8-10 Weeks	8-10 Weeks	8-10 Weeks	8-10 Weeks	8-10 Weeks	8-10 Weeks	8-10 Weeks	8-10 Weeks
Price	$3,500	$3,500	$3,000	$3,500	$3,500	$3,000	$3,500	$3,500

Contact a laboratory genetic counselor

labgc@ggc.org
+1 (800) 473-9411
Monday-Friday from 8am-5pm.

Specimen requirements

4-5 ml of peripheral blood should be collected in EDTA (lavender top) vacutainer tube.
Saliva or extracted DNA will also be accepted.

Shipping requirements

Ship at room temperature to:
106 Gregor Mendel Circle
Greenwood, SC 29646

Blood and Saliva Sample Kits

Available by request

Inheritance Patterns Key

AR: Autosomal Recessive
AD: Autosomal Dominant
XL: X-Linked
XLD: X-Linked Dominant
XLR: X-Linked Recessive
DG: Digenic Recessive

Gene	**Inheritance**	Bardet Biedl	Cone Rod Dystrophy	Congenital Stationary Night Blindness	Leber Congenital Amaurosis	Macular Degeneration	Ocular Albinism & Hermansky-Pudlak syndrome	Optic Atrophy	Retinititis Pigmentosa
ABCA4	AR/AD		x			x			x
ABHD12	AR								x
ACO2	AR							x	
ACVR1	AD							x	
ADAM9	AR		x						
ADIPOR1	AR	x							
AGBL5	AR								x
AIPL1	AR		x		x				x
AP3B1 (HPS2)	AR						x		
ARL2BP	AR								x
ARL3	AD								x
ARL6 (BBS3)	AR	x							x
ASB10	AD							x	
ATF6	AR		x						
AUH	AR							x	
BBIP1 (BBS18)	AR	x							
BBS1	AR/AD	x							x
BBS10	AR	x							
BBS12	AR	x							
BBS2	AR	x							x
BBS4	AR	x							
BBS5	AR	x							
BBS7	AR	x							
BEST1	AR/AD					x		x	x
BEST1/VMD2	AD		x						
BLOC1S3 (HPS8)	AR						x		
BLOC1S6 (HPS9/ PLDN)	AR						x		
C10orf11 (OCA7)	AR						x		
C12orf65	AR							x	
C1QTNF5	AD					x			x
C21orf2	AR		x						
C2orf71	AR								
C8orf37	AR	x	x						x
CA4	AD								x
CABP4	AR			x	x				
CACNA1F	XL		x	x					
CACNA2D4	AR		x						
CANT1	AR							x	
CCDC28B	DR, AR	x							
CDH3	AR					x			
CDHR1	AR		x						x
CEP290 (BBS14)	AR	x			x				x
CERKL	AR		x						x

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CHM	XLD								x
CHST6	AR					x			
CISD2	AR							x	
CLN3	AR								x
CLRN1	AR								x
CLUAP1	AR				x				
CNGA1	AR								x
CNGA3	AR		x						
CNGB1	AR								x
CNGB3	AR		x			x			
CNNM4	AR		x						
COL4A1	AD							x	
CRB1	AR		x		x				x
CRX	AD		x		x				x
CTNNA1	AD					x			
CYP1B1	AR							x	
CYP4V2	AR								x
DHDDS	AR								x
DHX38	AR								x
DRAM2	AR					x			
DTHD1	AR				x				
DTNBP1 (HPS7)	AR						x		
EFEMP1	AD					x			
ELOVL4	AD					x			
EMC1	AR								x
EYS	AR		x						x
FAM161A	AR								x
FBLN5	AD					x			
FLVCR1	AR								x
FOXC1	AD							x	
FOXE3	AR/AD							x	
FSCN2	AD					x			
FSCN2	AD								x
GDF6	AR				x				
GNAT1	AR/AD			x					
GNAT2	AR		x						
GNB3	AR			x					
GPR143	XL						x		
GPR179	AR			x					
GRK1	AR/AD			x					
GRM6	AR			x					
GUCA1A	AD		x						
GUCA1B	AD					x			x
GUCY2D	AR/AD		x		x				x

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HK1	AD								x
HMCN1	AD					x			
HPS1	AR						x		
HPS3	AR						x		
HPS4	AR/AD						x		
HPS5	AR						x		
HPS6	AR						x		
IDH3B	AR								x
IFT140	AR				x				x
IFT27 (BBS19)	AR	x							
IMPDH1	AD				x				x
IMPG1	AD					x			
IMPG2	AD					x			x
INPP5E	AR	x							
IQCB1	AR				x				
KCNJ13	AR/AD	x			x				
KCNV2	AR		x						
KIZ (PLK1S1)	AR								x
KLHL7	AD								x
LCA5	AR				x				x
LMX1B	AD							x	
LRAT	AR				x				x
LRIT3	AR			x					
LTBP2	AR							x	
LYST	AR						x		
LZTFL1 (BBS17)	AR	x							
MAF	AD							x	
MAK	AR								x
MC1R	AR						x		
MERTK	AR								x
MFRP	AR							x	x
MFSD8	AR					x			
MKKS (BBS6)	AR	x							
MKS1 (BBS13)	AR	x							
MTPAP	AR							x	
MVK	AR								x
MYOC	AD							x	
NDUFS1	AR							x	
NEK2	AR								x
NEUROD1	AR								x
NMNAT1	AR				x				
NPHP1	AR	x							
NR2E3	AR/AD								x
NR2F1	AD							x	
NRL	AD								x

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NYX	XLR			x					
OCA2	AR						x		
OPA1	AR/AD							x	
OPA3	AR/AD							x	
OPTN	AR				x				
OFD1	XLR								x
OPTN	AD							x	
OTX2	AD				x	x			
PAX6	AD							x	
PDE6A	AR								x
PDE6B	AR/AD			x					x
PDE6C	AR		x						
PDE6G	AR								x
PDE6H	AR/AD		x						
PITPNM3	AD		x						
PITX2	AD							x	
PITX3	AD							x	
POC1B	AR		x						
POLG	AR/AD							x	
POMGNT1	AR								x
PRCD	AR								x
PRKCG	AD								x
PROM1	AR		x			x			x
PRPF3	AD								x
PRPF31	AD								x
PRPF4	AD								x
PRPF6	AD								x
PRPF8	AD								x
PRPH2	AR		x		x	x			x
PTHB1 (BBS9)	AR	x							
RAB28	AR		x						
RAX2	AD		x			x			
RBP4	AR/AD								x
RD3	AR				x				x
RDH12	AR				x				x
RDH5	AD/AR		x	x					
RGR	AD/AR								x
RHO	AR			x					x
RIMS1	AD		x						
RLBP1	AD/AR								x
ROM1	AD/AR								x
RP1	AD/AR								x
RP1L1	AR					x			x
RP2	XLR								x
RP9	AD								x
RPE65	AR				x				x

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RPGR	XL		x			x			x
RPGRIP1	AR		x		x				x
SAG	AR			x					x
SBF2	AR							x	
SDCCAG8 (BBS16)	AR	x							
SEMA4A	AR/AD		x						x
SH3PXD2B	AR							x	
SLC24A1	AR			x					
SLC24A5 (OCA6)	AR						x		
SLC45A2 (OCA4)	AR						x		
SLC4A4	AR							x	
SLC7A14	AR								x
SNRNP200	AD								x
SPATA7	AR				x				x
SPG7	AR/AD							x	
SPP2	AD								x
TBK1	AD							x	
TIMP3	AD					x			
TMEM126A (OPA7)	AR							x	
TMEM67	AR	x							
TOPORS	AD								x
TRIM32 (BBS11)	AR	x							
TRPM1	AR			x					
TTC8 (BBS8)	AR	x							x
TTLL5	AR		x						
TULP1	AR		x		x				x
TYR (OCA1A)	AR						x		
TYRP1 (OCA3)	AR						x		
UNC119	AD		x						
USH2A	AR								x
WDPCP (BBS15)	AR	x							
WDR19	AR								x
WFS1	AR/AD							x	
ZNF408	AR								x
ZNF513	AR								x

MORE ANSWERS FOR MORE PATIENTS

Mission Statement

The Greenwood Genetic Center is a nonprofit institute organized to provide clinical genetic services, diagnostic laboratory testing, educational programs and resources and research in the field of medical genetics.

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Phone: 864-941-8110; FAX: 864-941-8141

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