

# Order Form

CeGaT GmbH  
 Praxis für Humangenetik  
 Paul-Ehrlich Str. 17  
 72076 Tübingen

## CeGaT-Panels

Eye diseases, Primary ciliary dyskinesia, Joubert syndrome,  
 Refsum disease and Stickler syndrome



**Patient / Person**

Surname: .....

First name: .....

Date of birth: .....

DNA-No.: .....

**Sent by**

Name: .....

Mailing address: .....

Phone number: .....

**Date & Signature:** .....

**Indication / Diagnosis**

<input type="checkbox"/> autosomal dominant	<input type="checkbox"/> sporadic
<input type="checkbox"/> autosomal recessive	<input type="checkbox"/> familial
<input type="checkbox"/> X-chromosomal	<input type="checkbox"/> consanguineous
<input type="checkbox"/> unclear	<input type="checkbox"/> segregation

**Material**

2x 5-10 ml EDTA-blood or 5-7 µg DNA

**Invoice**

to patient

to sender

**Panels**

**Panel 1: Usher syndrome:** CDH23, CLRN1, CLRN3, DFNB31, GPR98, MYO7A, PCDH15, PDZD7, USH1C, USH1G, USH2A

**Panel 2: Autosomal dominant retinitis pigmentosa:** BEST1, CA4, CRX, FSCN2, GUCA1B, IMPDH1, KLHL7, NR2E3, NRL, PRPF3, PRPF31, PRPF6, PRPF8, PRPH2, RDH12, RGR, RHO, ROM1, RP1, RP2, RP9, RPE65, RPGR, SEMA4A, SNRNP200, TOPORS

**Panel 3: Autosomal recessive retinitis pigmentosa:** ABCA4, BBS1, BBS8 (TTC8), BEST1, C2ORF71, C8ORF37, CERKL, CNGA1, CNGB1, CRB1, DHDDS, EYS, FAM161A, FLVCR1, GNPTG, IDH3B, IMPG2, LRAT, MAK, MERTK, NR2E3, NRL, PDE6A, PDE6B, PDE6G, PRCD, PROM1, RBP3, RDH12, RGR, RHO, RLBP1, RP1, RP2, RPE65, RPGR, SAG, SPATA7, TULP1, USH2A, ZNF513

**Panel 4: Achromatopsia:** step-by-step-analysis 1. CNGB3: c.1148delC, 2. CNGA3, 3. CNGB3, 4. GNAT2, 5. PDE6C

**Panel 5: Bardet Biedl syndrome:** ALMS1, BBS1, BBS2, BBS3 (ARL6), BBS4, BBS5, BBS6 (MKKS), BBS7, BBS8 (TTC8), BBS9, BBS10, BBS11, BBS12, BBS13 (MKS1), BBS14 (CEP290), CCDC28B, SDCCAG8, WDPCP

**Panel 6: Congenital stationary night blindness:** CABP4, CACNA1F, CACNA2D4, GNAT1, GPR179, GRK1, GRM6, NYX, PDE6B, RBP4, RHO, SAG, SLC24A1, TRPM1

**Panel 7: Joubert syndrome:** AH11, ARL13B, CC2D2A, CEP290, CEP41, INPP5E, KIF7, NPHP1, OFD1, RPGRIP1L, TECT1, TCTN2, TMEM138, TMEM216, TMEM237, TMEM67, TTC21B

**Panel 8: Leber congenital amaurosis:** AIPL1, CEP290, CRB1, CRX, GUCY2D, KCNJ13, LCA5, LRAT, NPHP5, RD3, RDH5, RDH12, RPE65, RPGRIP1, SPATA7

**Panel 9: Primary ciliary dyskinesia:** CCDC39, CCDC40, DNAAF1, DNAAF2 (KTU), DNAH5, DNAH11, DNAI1, DNAI2, DNAL1, RSPH4A, RSPH9, TXNDC3

**Panel 10: Refsum disease:** PHYH, PEX1, PEX2, PEX7, PEX26

**Panel 11: Senior Loken syndrome:** CEP290, NPHP1, NPHP2, NPHP3, NPHP4, NPHP5, SDCCAG8

**Panel 12: Stargardt disease and macular dystrophies:** ABCA4, BEST1, C1QTNF5, CDH3, CNGB3, ELOVL4, FSCN2, PROM1, PRPH2, RDH12, RP1L1, RPGR, TIMP3

**Panel 13: Cone- / Cone-rod dystrophies:** ABCA4, ADAM9, AIPL1, C8ORF37, CABP4, CACNA1F, CACNA2D4, CDHR1, CERKL, CNGB3, CNM4, CRX, GUCA1A, GUCY2D, KCN2, PDE6C, PITPNM3, PROM1, PRPH2, RAX2, RDH5, RGS9, RGS9BP, RIMS1, RPGR, RPGRIP1, SEMA4A

**Panel 14: Flecked retina disorders:** CHM, EFEMP1, PLA2G5, RDH5, RLBP1, RS1, VPS13B

**Panel 15: Familial exudative vitreoretinopathy and Wagner syndrome :** COL2A1, FZD4, LRP5, NDP, TSPAN12, VCAN

**Panel 16: Stickler syndrome:** COL2A1, COL9A1, COL9A2, COL11A1, COL11A2

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**Declaration of consent**

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Patient / Person Medical doctor

**Anonymization for scientific purposes**

I agree that left over material might be anonymously used for research purposes.

yes  no Patient / Person