

### General Information

**Patient**

Surname: \_\_\_\_\_

First name: \_\_\_\_\_

Date of birth: \_\_\_\_\_

Sex:  male  female

**Material**

Blood \_\_\_\_ ml (min. 3 ml EDTA-blood)

Dried blood spot cards (at least 10 spots)

DNA \_\_\_\_ µg (min. 5 µg DNA, concentr. ≥ 50 ng/µl) DNA-No.: \_\_\_\_\_

Other specimen \_\_\_\_\_

External ID: \_\_\_\_\_

Date of sample collection: \_\_\_\_\_

Samples can be sent by mail in a cardboard box or air cushion envelope. Samples should not be exposed to direct sunlight. Dried blood spot cards can be ordered for free (info@cegat.com).

**Sender / Clinic**

Surname: \_\_\_\_\_

First name: \_\_\_\_\_

Institution: \_\_\_\_\_

Street: \_\_\_\_\_

Postcode/City: \_\_\_\_\_

Country: \_\_\_\_\_

Phone: \_\_\_\_\_

Email: \_\_\_\_\_

If applicable, please include a VAT number or a copy of your business registration certificate.

VAT: \_\_\_\_\_

**Invoice**

to patient  to sender / clinic

**Declaration of consent**

**By signing this form, I declare that I have received comprehensive information about the genetic background related to the disease in question as well as the possibilities and limitations of molecular genetic testing. I understand that I have the right to withdraw my consent to genetic analyses.**

**I have been informed, and agree, that the data obtained in the analysis will be recorded, evaluated, or stored in an anonymized form in scientific databases, and further, in accordance with data protection and medical confidentiality, that the request, or parts thereof, may be transmitted to a specialized cooperating laboratory. I have been informed, and agree, that all data collected by CeGaT GmbH is electronically stored, processed, and used. I also consent to the data being transmitted electronically (e.g. by e-mail or fax).**

**If you do not check these boxes, your answer will be recorded as “No”.**

I consent to the storage of my genetic material for additional tests and/or quality control (for max. 10 years).  Yes  No

I consent to the storage of my test results beyond the timespan of 10 years (as required by German law).  Yes  No

I consent to the anonymous storage and use of surplus genetic material and/or test results for scientific research.  Yes  No

Genetic variation may sometimes be identified, which does not fit within the scope of the requested genetic analysis (so-called secondary findings report). The reporting of these variants is limited to pathogenic alterations within selected genes, for which a treatment or course of action exists for you or your family (according to the current guidelines of the American College of Medical Genetics and Genomics; ACMG SF V2.0; Kalia et al., 2017, PMID: 27854360). There is no claim of a comprehensive analysis of the genes included within the secondary findings report. An absence of secondary findings cannot be used to indicate a reduced disease risk.

**With regard to secondary findings I would like:**

to be informed

to NOT be informed

**Please Note**

Our panels are regularly updated to reflect current scientific research. It should therefore be recognized that there is the possibility that the list of genes on the order form may have changed slightly (genes added or removed) by the time the sample is analyzed in the laboratory. By signing this form, the patient accepts that the list of genes actually analyzed may be slightly different from what is currently listed. When NGS is utilized more than the requested genes are sequenced for each sample.

**This declaration of consent can be completely or partially withdrawn at any time. I have had sufficient time to consider giving my consent.**

\_\_\_\_\_  
Patient / Legal Guardian (Block letters)



\_\_\_\_\_  
Doctor (Surname, First name)

**X** \_\_\_\_\_  
Patient / Legal Guardian (Date, Signature)

**X** \_\_\_\_\_  
Doctor (Date, Signature)

**Contact**

To discuss the diagnostic strategy please do not hesitate to contact us.  
Phone: +49 7071 565 44-55  
Email: diagnostic-support@cegat.de

CeGaT is accredited by DAKKS according to DIN EN ISO 15189:2014, by the College of American Pathologists (CAP) and CLIA.

**Doctor's stamp / Barcode**

Indication

**Indication / Suspected Diagnosis:**

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**Clinical Major Symptoms:**

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**Preliminary genetic diagnostics:**

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**Transplants (bone marrow, tissue, stem cells)**    No    Yes, (please specify) \_\_\_\_\_

**Please include a copy of all existing reports of your patient.**

**Pedigree**                      Consanguinity:  Yes    No            Ethnic origin: \_\_\_\_\_

-  index patient
- not affected
- affected
- known carrier
- deceased
- unrelated parents
- consanguine parents
- unborn child
- abortion, stillborn child
- person of unknown sex
- identical twins (monozygous)
- fraternal twins (dizygous)

**Family medical history**

Are there other family members who currently have or have had the same or a similar disease as the patient?

Yes       No

If yes, please list the affected family members:

Name (not required)	Relationship to the patient (e.g. mother)	Age of onset	Diagnosis / Symptoms

## Inquiry

All genes listed on this order form, and the entire mitochondrial genome, are sequenced in parallel. This allows you to select multiple gene sets, or choose individual genes in addition to your selected gene set. As there is no additional laboratory expenditure, there is only a moderate price increase due to the subsequent analysis and interpretation. We are happy to answer your questions or send you an individual quote. Please contact us at [info@cegat.com](mailto:info@cegat.com).

 **Large Panel Diagnostic Option (272 Genes, KID-D45)**

In addition to the full analysis of the requested gene set(s), we will expand the analysis to all pathogenic and likely pathogenic variants (ACMG class 4 and 5) in genes which are related to the indication described for the proband on the entire diagnostic panel (272 genes in total).

 **Nephronophthisis (29 Genes, KID01)**

NPHP1 – Deletion/duplication analysis NOT required

AHI1, ANKS6, ATXN10, CC2D2A, CEP164, CEP290, CEP83, CTNS, DCDC2, FAN1, GLIS2, INVS, IQCB1, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, RPGRIP1L, SDCCAG8, SLC41A1, TMEM216, TMEM67, TRAF3IP1, TTC21B, WDR19, WDR35, XPNPEP3, ZNF423

 **Cystic Kidney Disease (16 Genes, KID02)**

PKD1, PKD2

ANKS6, BICC1, ETFA, GANAB, HNF1B, INVS, LRP5, NPHP3, OFD1, PAX2, PKHD1, PMM2, SEC61A1, UMOD

 **Renal Tubular Dysgenesis (4 Genes, KID03)**

ACE, AGT, AGTR1, REN

 **Renal Dysplasia, Renal Agenesis, CAKUT (48 Genes, KID04)**

ALDH1A2, BICC1, BMP4, BMP7, CDC5L, CHD1L, DACH1, DSTYK, EYA1, FGF20, FIBP, FOXC1, FRAS1, FREM1, FREM2, GATA3, GDNF, GREM1, GRIP1, HNF1B, ITGA8, KIF14, NEK8, NPHP3, OSR1, PAX2, RET, ROBO2, SALL1, SDCCAG8, SIX1, SIX2, SIX5, SLIT2, SOX17, SPRY1, SRGAP1, TBC1D1, TBX18, TFAP2A, TNS3, TRAP1, UMOD, UPF3B, UPK3A, WNT4, WNT5A, WT1

 **Nephrotic Syndrome (40 Genes, KID05)**

ACTN4, ADCK4, ANLN, APOE, APOL1, ARHGAP24, ARHGDI1, CD2AP, COQ2, COQ6, CRB2, CUBN, DGKE, EMP2, FAT1, INF2, ITGA3, ITGB4, KANK1, KANK2, KANK4, LAMB2, LMX1B, MAGI2, MYH9, MYO1E, NPHS1, NPHS2, NUP107, NUP205, NUP93, PDSS2, PLCE1, PTPRO, SMARCAL1, TRPC6, TTC21B, WDR73, WT1, XPO5

 **Focal Segmental Glomerulosclerosis (32 Genes, KID06)**

ACTN4, ADCK4, ANLN, APOL1, ARHGAP24, CD2AP, COL4A3, COL4A4, COL4A5, COQ2, COQ6, CRB2, FBXW7, INF2, LAMA5, LAMB2, LMNA, LMX1B, MYH9, MYO1E, NPHP1, NPHP4, NPHS1, NPHS2, NXF5, PAX2, PDSS2, PLCE1, SYNPO, TRPC6, TTC21B, WT1

 **Alport Syndrome and Disorders of Glomerular Basement Membrane (GBM) (7 Genes, KID07)**

CD151, COL4A3, COL4A4, COL4A5, FN1, MYH9, PXDN

 **C1q Deficiency (3 Gene, KID08)**

C1QA, C1QB, C1QC

 **Renal Tubular Acidosis (9 Genes, KID09)**

ATP6V0A4, ATP6V1B1, BCS1L, CA2, OCRL, SLC4A1, SLC4A4, VIPAS39, VPS33B

 **Bartter Syndrome and Differential Diagnosis (17 Genes, KID10)**

BSND, CASR, CLCN5, CLCNKA, CLCNKB, CLDN16, CLDN19, CNM2, EGF, FXD2, GNA11, INSR, KCNJ1, MAGED2, SLC12A1, SLC12A3, TRPM6

 **Hypophosphatemic Rickets (15 Genes, KID11)**

ALPL, CLCN5, CYP27B1, CYP2R1, DMP1, ENPP1, FAH, FGF23, KL, OCRL, PHEX, SLC34A1, SLC34A3, SLC9A3R1, VDR

 **Pseudohypaldosteronism (9 Genes, KID12)**

CUL3, HSD11B2, KLHL3, NR3C2, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4

 **Diabetes insipidus, nephrogenic (3 Genes, KID13)**

AQP2, AVP, AVPR2

 **Hyperoxaluria (3 Genes, KID14)**

AGXT, GRHPR, HOGA1

 **Atypical Hemolytic Uremic Syndrome and Differential Diagnosis\* (19 Genes, KID15)**

ADAMTS13, C3, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CLU, DGKE, MMACHC, MMADHC, MUT, PIGA, PLG, THBD

\* Please note that a molecular genetic test for the detection of known CFHR1/CFHR4 and CFHR3/CFHR1 deletions as well as CFHR1/CFH and CFH/CFHR1 hybrid allele is currently in validation phase and may be requested shortly.

 **Primary Inherited Aminoacidurias (8 Genes, KID16)**

SLC1A1, SLC2A2, SLC36A2, SLC3A1, SLC6A19, SLC6A20, SLC7A7, SLC7A9

 **Branchiootorenal Syndrome (5 Genes, KID17)**

EYA1, SALL1, SIX1, SIX5, TFAP2A

 **Bardet-Biedl Syndrome (30 Genes, KID18)**

ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8ORF37, CCDC28B, CEP164, CEP290, IFT172, IFT27, IFT74, KIF7, LZTFL1, MKKS, MKS1, NPHP1, SDCCAG8, TMEM67, TRAPPC3, TRIM32, TTC21B, TTC8, WDPCP

 **Joubert Syndrome (39 Genes, KID19)**

AHI1, ARL13B, ARMC9, B9D1, B9D2, C2CD3, C5ORF42, CC2D2A, CELSR2, CEP104, CEP120, CEP164, CEP290, CEP41, CSPP1, EXOC8, HYLS1, INPP5E, KIAA0556, KIAA0586, KIAA0753, KIF7, MKS1, NPHP1, OFD1, PDE6D, PDP1, PIBF1, RPGRIP1L, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B, ZNF423

 **Meckel Syndrome (24 Genes, KID20)**

AHI1, B9D1, B9D2, CC2D2A, CEP120, CEP290, CEP55, CSPP1, KIAA0586, KIF14, MKS1, NPHP3, RPGRIP1L, TCTN1, TCTN2, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B, TXNDC15, WDPCP

 **Senior-Loken Syndrome (11 Genes, KID21)**

CEP164, CEP290, INVS, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19, ZNF423

## Additional analyses

For further information and advice please do not hesitate to contact our Diagnostic Support team.

[www.cegat.de/en/diagnostic-support](http://www.cegat.de/en/diagnostic-support) · [diagnostic-support@cegat.de](mailto:diagnostic-support@cegat.de) · Phone +49 7071 565 44-55