

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 & Anamnise

Indication / Suspected Diagnosis:

Clinical Major Symptoms:

Preliminary genetic diagnostics:

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

 Large Panel Diagnostic Option (253 Genes, DRM-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 (253 genes in total).

 Oculocutaneous albinism (8 Genes, DRM01)

C10orf11 (LRMDA), GPR143, MC1R, OCA2, SLC24A5, SLC45A2, TYR, TYRP1

 Syndromic albinism and related disorders: Hermansky-Pudlak syndrome, Griscelli syndrome, Waardenburg syndrome (21 Genes, DRM02)

AP3B1, BLOC1S3, BLOC1S6, DTNBP1, EDN3, EDNRB, EPG5, HPS1, HPS3, HPS4, HPS5, HPS6, KIT, LYST, MITF, MLPH, MYO5A, PAX3, RAB27A, SOX10, TYR

 Hyperpigmentation: Dowling-Degos disease and related disorders (16 Genes, DRM03)

ABCB6, ADAM10, ADAR, BRAF, KITLG, KRT14, KRT5, MAP2K1, POFUT1, POGlut1, PSENNEN, PTPN11, RAF1, SASH1, ST3GAL5, STK11

 Ichthyosis, palmoplantar keratoderma, and related disorders of cornification (64 Genes, DRM04)

AAGAB, ABCA12, ABHD5, ALDH3A2, ALOX12B, ALOXE3, AP1S1, AQP5, ATP2A2, ATP2C1, CARD14, CDSN, CERS3, CLDN1, CSTA, CTSC, CYP4F22, DSG1, DSP, ELOVL4, ENPP1, ERCC2, ERCC3, FLG, GJA1, GJB2, GJB3, GJB4, GJB6, GTF2E2, GTF2H5, JUP, KDSR, KRT1, KRT10, KRT14, KRT16, KRT17, KRT2, KRT6A, KRT6B, KRT6C, KRT9, LOR, MBTPS2, MPLKIP, MVK, NIPAL4, NSDHL, PNPLA1, SDR9C7, SERPINB7, SLC27A4, SLURP1, SNAP29, SPINK5, ST14, STS, SULT2B1, SUMF1, TGM1, TGM5, TRPV3, WNT10A

 Epidermolysis bullosa and related genetic blistering disorders (23 Genes, DRM05)

CDSN, COL17A1, COL7A1, CSTA, DSP, DST, EXPH5, FERMT1, ITGA3, ITGA6, ITGB4, JUP, KLHL24, KRT1, KRT10, KRT14, KRT5, LAMA3, LAMB3, LAMC2, PKP1, PLEC, TGM5

 Ectodermal dysplasia, selective tooth agenesis, trichothiodystrophy, and hypotrichosis (61 Genes, DRM08)

ANTXR1, APCDD1, AXIN2, CDH3, CDSN, CTSC, DLX3, DSG4, DSP, DYNC2LI1, EDA, EDAR, EDARADD, ERCC2, EVC, EVC2, FGF10, FGFR2, FGFR3, GJA1, GJB6, GRHL2, GTF2E2, GTF2H5, HOXC13, HR, IFT122, IFT43, IFT52, IKBKG, KCTD1, KRT14, KRT74, KRT81, KRT83, KRT85, KRT86, LIPH, LPAR6, LRP6, LTBP3, MPLKIP, MSX1, NFKBIA, OFD1, PAX9, PKP1, POC1A, PORCN, PVRL1 (NECTIN1), PVRL4 (NECTIN4), SNRPE, SOX18, ST14, TP63, TRPS1, UBR1, WDR19, WDR35, WNT10A, WNT10B

 Dyskeratosis congenita (12 Genes, DRM09)

ACD, CTC1, DKC1, NHP2, NOP10, PARN, RTEL1, TERC, TERT, TINF2, USB1, WRAP53

 Photodermatosis: Xeroderma pigmentosum, Cockayne syndrome, COFS syndrome and related disorders (19 Genes, DRM10)

BLM, DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FERMT1, GTF2E2, GTF2H5, MPLKIP, POLH, RECQL4, USB1, UVSSA, XPA, XPC

 Neurofibromatosis 1 and related disorders (5 Genes, DRM11)

LZTR1, NF1, NF2, SMARCB1, SPRED1

 Vascular disorders: hereditary hemorrhagic telangiectasia, cerebral cavernous malformations, association with MoyaMoya, and related disorders (21 Genes, DRM12)

ACTA2, ACVRL1, CBL, CCM2, CECR1 (ADA2), COL3A1, ENG, EPHB4, GDF2, GUCY1A3 (GUCY1A1), JAG1, KRIT1, PDCD10, PTEN, RASA1, RNF213, SAMHD1, SHOC2, SMAD4, SOX18, THSD1

 Progeria syndromes and lipodystrophy (39 Genes, DRM13)

AGPAT2, ALDH18A1, ATP6V0A2, B4GALT7, BANF1, BLM, BSCL2, C1R, C1S, CAV1, COL3A1, CTC1, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FBN1, GTF2E2, GTF2H5, KCNJ6, LMNA, MPLKIP, OBFC1, PIK3R1, PLIN1, PNPLA2, POLD1, PPARG, PSMB8, PTSS1, PTRF(CAVIN1), PYCR1, RECQL4, SLC25A24, WRN, ZMPSTE24

 Lymphedema (12 Genes, DRM14)

ABCC9, CCBE1, FAT4, FLT4, FOXC2, GATA2, GJC2, IKBKG, KIF11, PIEZO1, SOX18, VEGFC

Additional analyses

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

www.cegat.de/en/diagnostic-support · diagnostic-support@cegat.de · Phone +49 7071 565 44-55