Order Form
Deep Immunogenetics

General Information

Patient

Surname: _____

First name: _____

Date of birth: _____

Sex (assigned at birth): ❑ female ❑ male

Gender (if differs from sex assigned at birth):
❑ man ❑ non-binary ❑ woman ❑ self-described: _____

Material

❑ Blood ____ ml (min. 1-2 ml EDTA-blood)

❑ Dried blood spot cards (at least 5 spots)

❑ DNA ____ µg (min. 1-2 µg DNA, concentr. ≥ 50 ng/µl) DNA-No.: _____

Source material
of extracted DNA: _____ (e.g. EDTA blood, skin biopsy)

❑ 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: _____

VAT: _____

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

Invoice
❑ to sender / clinic
❑ to patient / other (KVA-No.: _____)

Surname: _____

First name: _____

Street: _____

Postcode/City: _____

Country: _____

Email: _____

Declaration of consent

By signing this form, I declare that I have received comprehensive information regarding 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 for genetic analyses.

I have been informed, and agree, that my personal data and the data obtained in the analysis will be recorded, evaluated or stored in an pseudonymized form in scientific databases, and that further, in accordance with data protection and medical confidentiality, the request, or parts thereof, may be transmitted to a specialized cooperating laboratory.

I consent to the re-evaluation of my test results within the data storage period. If significant alterations become apparent, my Physician will be informed by e-mail.

I consent that in addition to the full genetic test as requested, the analysis can be expanded to all pathogenic and likely pathogenic variants (ACMG class 4 and 5) in genes which are related to the indication described for the proband (if applicable, screen for differential diagnosis).

I have been informed, and agree to the electronic storage, processing, use, and transmission of all data collected by CeGaT GmbH.

For more detailed information on data privacy as well as your rights please refer to www.cegat.com/privacy-policy.

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.

I, the referring physician, confirm that I am authorized to request genetic testing for the above-mentioned patient. For predictive testing, I confirm that I am authorized, and that I have fulfilled the requirements, to request this testing. For minors, I declare that I have the consent of all legal guardians.

If the patient did not sign this order form: I, the referring physician, confirm that the patient received genetic counseling and agrees with the genetic testing. The patient's consent has been obtained in writing.

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 pseudonymous storage and use of surplus genetic material and/or test results for scientific research and in scientific literature. ❑ Yes ❑ No

With regard to secondary findings I would like to be informed: ❑ Yes ❑ No

Genetic variation may sometimes be identified, which does not fit within the scope of the requested genetic analysis (so-called secondary findings). The reporting of these variants is limited to pathogenic alterations (ACMG classes 4 and 5) 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; details on genes and associated diseases can be found at www.cegat.com/acmg-genes). There is no claim of a comprehensive analysis of this gene set. An absence of secondary findings cannot be used to indicate a reduced disease risk.

According to German Genetic Diagnostic Act (GenDG) we will issue the medical report to the counselling physician. Please indicate here the contact email of the counselling physician:

Email: _____

Physician's stamp / Barcode

DAkkS Deutsche Akkreditierungsstelle D-ML-13206-01-00

CAP ACCREDITED COLLEGE of AMERICAN PATHOLOGISTS

CLIA CERTIFIED ID: 99D2130225

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

Patient / Legal Guardian (Block letters)	**Physician** (Surname, First name)
X _____ **Patient / Legal Guardian** (Date, Signature)	X _____ **Physician** (Date, Signature)

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Indication

Indication / Suspected diagnosis:

Major Clinical Symptoms:

Preliminary genetic diagnostics:

Transplants (bone marrow, tissue, stem cells) ❑ No ❑ Yes, (please specify)

Please include a copy of all relevant medical reports of your patient.

Pedigree Consanguinity: ❑ Yes ❑ No Ethnic origin:

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

Previous genetic diagnostics:

❑ none

❑ Chromosome analysis / Array-CGH: _____

❑ Sequence analysis of the following genes _____

Previous operations:

❑ Hematopoietic stem cell transplantation*

❑ Splenectomy

❑ none

*if no DNA or blood cells have been collected prior to transplantation, a sample from non-hematopoietic tissue is required.

Clinical information

Please provide the following clinical information for your patient. Detailed clinical information – also on the absence of phenotypic findings - will increase the likelihood of identifying causative alterations during NGS analysis and significantly improve the interpretation of identified variants. Please specify if applicable:

General symptoms

Age at disease onset: _____

For newborns / infants:
Prenatal medical history:

❑ normal

❑ preterm-birth: _____WG

❑ Other information on the course of pregnancy (e. g. medication): _____

❑ Recurrent fever

❑ with elevated inflammation markers ❑ infection-associated

❑ without further abnormalities

❑ accompanied by further symptoms, namely: _____

❑ Autoimmunity / Autoinflammation: _____

❑ Susceptibility to infection:
(pathogen and localization) _____

❑ Miscellaneous:
(e.g. skin or lung alterations) _____

❑ Previous and current therapy: _____

Blood values & immunological parameters

In case of abnormal test results please specify altered parameters, optionally using symbols such as ↑ elevated ↑↑ strongly elevated ↓ decreased ↓↓ strongly decreased

Inflammation markers: ❑ normal ❑ abnormal: _____

Immunoglobulins: ❑ normal ❑ abnormal: _____

Pathogen detection: ❑ not analyzed ❑ none detected

❑ detection of the following pathogens: _____

SCID newborn screening (TREC level) ❑ normal ❑ abnormal: _____

Hematology: ❑ normal ❑ lymphopenia

❑ thrombocytopenia ❑ anemia ❑ neutropenia

Immune phenotyping: ❑ normal ❑ abnormal: _____

_____

_____

Autoantibodies: ❑ none ❑ yes, the following: _____

Oxidative burst (DHR assay): ❑ normal ❑ abnormal: _____

Complement activity (CH50, AP50): ❑ normal ❑ abnormal: _____

Enzyme activities: ❑ normal ❑ abnormal: _____
(e. g. ADA, PNP)

Other clinical chemistry ❑ normal ❑ abnormal: _____

Other specialized (immune) diagnostics: _____
(e. g. lymphocyte function, telomere length, chromosome breakage)

Physical appearance / other abnormalities

❑ no abnormalities of the physical appearance

❑ Signs of (partial) albinism: _____

❑ Facial dysmorphism: _____

❑ Skeletal anomalies: _____

❑ Developmental delay: _____

❑ Mental retardation: _____

❑ Other abnormalities: _____

_____

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Inquiry

We recommend the high-coverage Deep Immunogenetics enrichment as a second-tier analysis after exome or panel diagnostics.

Optional step 1: Exome enrichment

❑ **Abnormal Lymphoproliferation (29 Genes, AID05)**
EBV susceptibility with lymphoproliferation / ALPS

CARMIL2, CASP10, CASP8, CD27, CD70, CORO1A, CTPS1, FADD, FAS, FASLG, IL2RA, IL2RB, ITK, KRAS, LAT, LRBA, MAGT1, NFKB1, NRAS, PIK3CD, PIK3R1, PRKCD, RASGRP1, SH2D1A, STAT3, STK4, TET2, TNFRSF9, XIAP

❑ **Periodic Fever Syndromes with/without Urticaria (9 Genes, AID01)**
ELANE, MEFV, MVK, NLRP12, NLRP3, NOD2, OTULIN, PSTPIP1, TNFRSF1A

❑ **Further Autoinflammatory Disorders without Type I Interferonopathies (35 Genes, AID02)**
Incl. inflammation of the skin, joints and bones, vasculitis

ADA2, ADAM17, AP1S3, ARPC1B, CARD11, CARD14, COPA, DPP9, ELF4, HAVCR2, HCK, IL1RN, IL36RN, LPIN2, LYN, NCKAP1L, NFKB1, NLRC4, NLRP1, NOD2, OTULIN, PLCG2, POMP, PSMA3, PSMB10, PSMB4, PSMB8, PSMB9, PSMG2, PSTPIP1, RELA, RIPK1, SLC29A3, SYK, TNFAIP3

Further gene sets can be found on the order forms for immune disorders and blood disorders.

Step 2: Deep Immunogenetics enrichment

❑ **Lymphoproliferation and Autoimmunity (47 Genes, DIG01)**
ADA2, CARD11, CARMIL2, CASP10, CASP8, CBLB, CCL22, CD27, CD70, CDC42, CTLA4, CTPS1, DEF6, FADD, FAS, FASLG, HAVCR2, IKZF1, IKZF3, IL2RA, IL2RB, ITK, JAK2, JAK3, KRAS, LAT, LRBA, MAGT1, NCKAP1L, NFKB1, NRAS, PIK3CD, PIK3R1, PRKCD, PTEN, PTPN11, RASGRP1, SH2D1A, SOCS1, STAT1, STAT3, STAT5B, STK4, TET2, TNFRSF9, XIAP, ZAP70

❑ **Autoinflammation (60 Genes, DIG02)**
ADA2, AP3B1, BACH2, CARD11, CARD14, CDC42, CEBPE, COPA, CTLA4, GATA2, HAVCR2, HCK, IFIH1, IL6ST, JAK1, LYN, LYST, NCKAP1L, NFKB1, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, OAS1, PDGFRA, PDGFRB, PIK3CG, PLCG1, PLCG2, POMP, PRF1, PSMA3, PSMB10, PSMB4, PSMB8, PSMB9, PSMG2, PSTPIP1, RAB27A, RC3H1, RELA, RIPK1, SLC7A7, SOCS1, STAT1, STAT2, STAT4, STAT6, STING1, STX11, STXBP2, SYK, TLR8, TNFAIP3, TNFRSF1A, TREX1, UBA1, UNC13D, XIAP

The Deep Immunogenetics enrichment comprises 337 genes. Please contact us if you are interested in an individual selection of the genes listed below.

ABL1, ACD, ADA, ADA2, AK2, AP3B1, ARID1A, ASXL1, ASXL2, ATM, ATR, ATRX, B2M, BACH2, BCL10, BCL11B, BCL2, BCOR, BCORL1, BIRC3, BLM, BRAF, BRCA1, BRCA2, BRCC3, BRIP1, BTK, CALR, CARD11, CARD14, CARMIL2, CASP10, CASP8, CBL, CBLB, CBLC, CCL22, CCND1, CD247, CD27, CD28, CD3D, CD3E, CD3G, CD48, CD70, CD79B, CD8A, CDC42, CDKN2A, CEBPA, CEBPE, CHD7, CIITA, COPA, COPG1, CREBBP, CSF1R, CSF3R, CSNK1A1, CTC1, CTCF, CTLA4, CTPS1, CUX1, CXCR4, CYBA, CYBB, CYBC1, DCLRE1C, DDX41, DEF6, DIS3, DKC1, DNAJC21, DNMT3A, DOCK2, DUT, EFL1, EGR1, EP300, ERBIN, ERCC4, ERCC6L2, ETNK1, ETV6, EXOC3L2, EZH2, FADD, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FAS, FASLG, FBXW7, FCHO1, FERMT1, FLT3, FOXI3, FOXN1, FOXO1, FYN, G6PD, GATA1, GATA2, GNAS, GNB1, GRHL2, HAVCR2, HCK, HRAS, ICOS, ID3, IDH1, IDH2, IFIH1, IKBKB, IKZF1, IKZF3, IL21R, IL2RA, IL2RB, IL2RG, IL6R, IL6ST, IL7R, IRF4, ITK, JAK1, JAK2, JAK3, KDM6A, KIT, KLF2, KLHL6, KMT2A, KMT2D, KRAS, LAT, LCK, LCP2, LIG1, LIG4, LIPA, LRBA, LYN, LYST, MAD2L2, MAGT1, MALT1, MAP2K1, MAP3K14, MECOM, MEF2B, MPL, MPO, MSN, MTHFD1, MTR, MYC, MYD88, MYSM1, NBN, NCF1, NCF2, NCF4, NCKAP1L, NF1, NFE2L2, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, NOP10, NOTCH1, NOTCH2, NPM1, NRAS, OAS1, ORAI1, PALB2, PARN, PAX1, PAX5, PDGFRA, PDGFRB, PGM3, PHF6, PIGA, PIK3CD, PIK3CG, PIK3R1, PLCG1, PLCG2, PNP, POMP, POT1, PPM1D, PRF1, PRKCD, PRKDC, PRPF40B, PRPF8, PSMA3, PSMB10, PSMB4, PSMB8, PSMB9, PSMG2, PSTPIP1, PTEN, PTPN11, PTPRC, RAB27A, RAC2, RAD21, RAD51, RAD51C, RAF1, RAG1, RAG2, RASGRP1, RBM8A, RBSN, RC3H1, RECQL4, RELA, RELB, RFWD3, RFX5, RFXANK, RFXAP, RHOA, RIPK1, RMRP, RPL11, RPL15, RPL26, RPL27, RPL31, RPL35A, RPL5, RPS10, RPS15, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, RPS7, RPSA, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SETBP1, SF1, SF3A1, SF3B1, SGK1, SH2B3, SH2D1A, SLC19A2, SLC46A1, SLC7A7, SLX4, SMC1A, SMC3, SOCS1, SRP54, SRP72, SRSF2, STAG2, STAT1, STAT2, STAT3, STAT4, STAT5B, STAT6, STIM1, STING1, STK4, STX11, STXBP2, SUZ12, SYK, TAP1, TAP2, TAPBP, TBX1, TCN2, TERC, TERT, TET2, TFRC, THPO, TINF2, TLR8, TNFAIP3, TNFRSF1A, TNFRSF9, TP53, TREX1, TSR2, U2AF1, U2AF2, UBA1, UBE2T, UBR5, UNC13D, USB1, VAV1, WAS, WIPF1, WRAP53, WT1, XIAP, XPO1, XRCC2, ZAP70, ZCCHC8, ZEB2, ZRSR2

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

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