Order Form
Immune Disorders

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 _____

Transplants (bone marrow, tissue, stem cells):

❑ No ❑ Yes (please specify): _____

Please Note: If you have previously had a stem cell transplant, please **do not send in EDTA blood**, but a **saliva sample.**

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.

Targeted analysis of the ACMG genes according to current recommendations can be requested as "additional analyses".

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

Patient / Legal Guardian
(Block letters)

Physician
(Surname, First name)

X _____
Patient / Legal Guardian
(Date, Signature)

X _____
Physician
(Date, Signature)

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, the College of American Pathologists (CAP) and CLIA.

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Indication

Analysis type: ❑ Proband **is affected** ❑ Proband is **NOT affected** (predictive testing)

Indication / Suspected diagnosis:

Major Clinical Symptoms:

Previous genetic testing:
(If performed, please specify test and results)

Please include a copy of all existing 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 testing:

❑ 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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Indication & Request

Request Immune Disorders

- ❑ **Primary Antibody Deficiencies (40 Genes, PID01)**
  Incl. hyper IgM syndrome, CVID, agammaglobulinemia, activated PI3Kdelta syndromes
  AICDA, ARHGEF1, ATP6AP1, BLNK, BTK, CARD11, CD19, CD40, CD40LG, CD79A, CD79B, CD81, CR2, CTNNBL1, FNIP1, ICOS, IGLL1, IKZF1, IRF4, IRF2BP2, MOGS, MS4A1, NFKB1, NFKB2, PIK3CD, PIK3CG, PIK3R1, PLCG2, PTEN, RAC2, SEC61A1, SH3KBP1, SLC39A7, SPI1, TCF3, TNFRSF13B, TNFRSF13C, TOP2B, TRNT1, UNG

- ❑ **Severe Combined Immunodeficiencies (SCID) (23 Genes, PID02)**
  ADA, AK2, ATM, BCL11B, CD247, CD3D, CD3E, CORO1A, DCLRE1C, IL2RG, IL7R, JAK3, LAT, LIG4, NHEJ1, PRKDC, PTPRC, RAC2, RAG1, RAG2, RMRP, RPSA, TBX1

- ❑ **Combined Immunodeficiencies (CID) (59 Genes, PID03)**
  ARPC1B, ARPC5, B2M, BCL10, CARD11, CD27, CD3E, CD3G, CD40, CD40LG, CD8A, CHD7, CIITA, DOCK2, DOCK8, FCHO1, FOXI3, FOXN1, GINS1, ICOS, IKBKB, IKZF1, IKZF2, ICOSLG, IL21, IL21R, ITK, LCK, LCP2, LIG1, MALT1, MAP3K14, MCM10, MCM4, MSN, MTHFD1, NFKBIA, ORAI1, PAX1, PNP, REL, RELA, RELB, RFX5, RFXANK, RFXAP, SASH3, SLC46A1, SP110, STIM1, STK4, TAP1, TAP2, TAPBP, TBX1, TBX2, TFRC, TNFRSF4, ZAP70

- ❑ **Hyper-IgE Syndrome (13 Genes, PID04)**
  ARPC1B, CARD11, DOCK8, ERBIN, IL6R, IL6ST, PGM3, SPINK5, STAT3, STAT6, TGFBR1, TGFBR2, ZNF341

- ❑ **Defects of the Complement System (17 Genes, PID06)**
  C1QA, C1QB, C1QC, C1R, C1S, C2, C3, C5, C6, C7, C8A, C8B, CFD, CFH, CFI, CFP, FCN3

- ❑ **Congenital Neutropenia (23 Genes, PID07)**
  CEBPE, CLPB, CSF3R, CXCR2, CXCR4, DNAJC21, EFL1, ELANE, G6PC3, GFI1, GINS1, HAX1, JAGN1, LAMTOR2, SBDS, SLC37A4, SMARCD2, SRP54, TAFAZZIN, USB1, VPS13B, VPS45, WAS

- ❑ **Chronic Granulomatous Disease (Defects of Respiratory Burst) (7 Genes, PID08)**
  CYBA, CYBB, CYBC1, G6PD, NCF1 (c.75_76delGT), NCF2, NCF4

- ❑ **Chronic Mucocutaneous Candidiasis (8 Genes, PID10)**
  AIRE, CARD9, IL17F, IL17RA, IL17RC, RORC, STAT1, TRAF3IP2

- ❑ **Susceptibility to Mycobacterial Infections (17 Genes, PID11)**
  CYBB, IFNG, IFNGR1, IFNGR2, IL12B, IL12RB1, IL23R, IRF1, IRF8, ISG15, JAK1, MCTS1, RORC, SPPL2A, STAT1, TBX21, TYK2

- ❑ **Susceptibility to Viral Infections (22 Genes, PID12)**
  DBR1, GATA2, GINS1, IFIH1, IFNAR1, IFNAR2, IRF3, IRF7, IRF9, MCM4, POLR3A, POLR3F, RANBP2, SNORA31, STAT1, STAT2, TBK1, TICAM1, TLR3, TRAF3, UNC93B1, ZNFX1

- ❑ **Periodic Fever Syndromes with/without Urticaria (10 Genes, AID01)**
  ELANE, F12, MEFV, MVK, NLRP12, NLRP3, NOD2, OTULIN, PSTPIP1, TNFRSF1A

- ❑ **Further Autoinflammatory Disorders (38 Genes, AID02)**
  Incl. inflammation of the skin, joints and bones, vasculitis
  ADA2, ADAM17, AP1S3, ARPC1B, CARD11, CARD14, COPA, DPP9, ELF4, HAVCR2, HCK, IL1RN, IL36RN, LACC1, LPIN2, LYN, NCKAP1L, NFKB1, NLRC4, NLRP1, NOD2, OTULIN, PLCG2, POMP, PSMA3, PSMB10, PSMB4, PSMB8, PSMB9, PSMG2, PSTPIP1, RELA, RIPK1, SLC29A3, STING1, SYK, TNFAIP3, UBA1

- ❑ **Type I Interferonopathies (25 Genes, AID07)**
  Incl. interferonopathies with leading neurological symptoms, Aicardi-Goutières-syndrome and systemic lupus erythematosus (SLE) / chilblain lupus
  ADA2, ADAR, C1QA, C1QB, C1QC, C2, C2orf69, C3, DNASE1L3, DNASE2, IFIH1, LSM11, OAS1, POLA1, PRKCD, RNASEH2A, RNASEH2B, RNASEH2C, RNU7-1, SAMHD1, STAT2, STING1, TLR7, TREX1, UNC93B1

- ❑ **Immune Dysregulation with Early Onset Colitis (IBD) (34 Genes, AID03)**
  AICDA, ARPC1B, BACH2, CD3G, CD40LG, CD55, CTLA4, CYBA, CYBB, CYBC1, ELF4, FOXP3, GUCY2C, IL10, IL10RA, IL10RB, LRBA, MALT1, NCF1 (c.75_76delGT), NCF2, NCF4, NLRC4, PIK3CD, RIPK1, SKIC2, SKIC3, SLC37A4, SLCO2A1, STAT3, TGFB1, TNFAIP3, TRIM22, TTC7A, XIAP

- ❑ **Primary Hemophagocytic Lymphohistiocytosis (HLH) (11 Genes, AID04)**
  AP3B1, AP3D1, HAVCR2, LIPA, LYST, PRF1, RAB27A, SLC7A7, STX11, STXBP2, UNC13D

- ❑ **Abnormal Lymphoproliferation (33 Genes, AID05)**
  EBV susceptibility with lymphoproliferation / ALPS
  CARMIL2, CASP10, CASP8, CBLB, CD27, CD70, CDC42, CORO1A, CTLA4, CTPS1, FADD, FAS, FASLG, IL2RA, IL2RB, ITK, KRAS, LAT, LRBA, MAGT1, NFKB1, NRAS, PIK3CD, PIK3R1, PRKCD, RASGRP1, SH2D1A, SOCS1, STAT3, STK4, TET2, TNFRSF9, XIAP
  For patients with autoimmune lymphoproliferative diseases, we offer a comprehensive deep sequencing panel to detect somatic mosaic variants (see the **Deep Immunogenetics** order form for details).

- ❑ **Regulatory T-Cells Defects / IPEX (16 Genes, AID06)**
  BACH2, CARMIL2, CTLA4, DEF6, DOCK8, FAS, FASLG, FOXP3, IKBKB, IKZF1, IL2RA, IL2RB, LRBA, MALT1, STAT3, STAT5B

Additional analyses (additional fees may apply)

- ❑ **HLA-Typing (HLA01)**
  I would like to receive an additional report stating the HLA alleles (HLA class I (Gene A, B, C) and HLA class II (Gene DPA1, DPB1, DQA1, DQB1, DRB1, DRB3, DRB4, DRB5)).

- ❑ **ACMG Genes Diagnostics**
  I would like to be informed of relevant alterations within the list of recommended genes for secondary analysis, according to the current guidelines of the American College of Medical Genetics and Genomics. The analysis is restricted to the sequence data, re-sequencing of regions with poor sequence coverage will not typically be performed. A negative “ACMG genes” report cannot be used to rule out (genetic) disease risk. Additional fees may apply. According to German legislation, predictive tests for minors may not be performed for diseases which have an onset in adulthood. Therefore, some genes will not be analyzed for minors, unless the phenotypic spectrum is within the scope of the primary medical indication of the patient.
  Details on genes and associated diseases can be found at www.cegat.com/acmg-genes

- ❑ **Pharmacogenetics (PGX)**
  I would like to receive an additional report analyzing known variants that are involved in the metabolism of pharmaceutical products. Details can be found at www.cegat.com/pgx

- ❑ _____

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

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