Order Form
Skeletal 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:

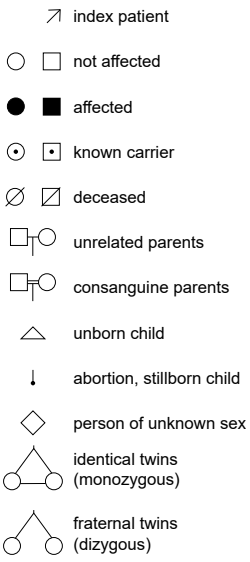
Major Clinical Symptoms:

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

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Indication & Request

Detailed Medical History

Onset of symptoms: ❑ prenatal ❑ postnatal, age at onset: _____

Pregnancy history: ❑ normal ❑ abnormal (please specify): _____

Prenatal ultrasound: ❑ normal ❑ abnormal (please specify): _____

Birth data: size: _____ weight: _____ head circumference: _____

Radiology report: _____
(please attach copy of results if patient allows)

Phenotypic features / other anomalies:

❑ dysmorphic features

❑ developmental delay

❑ abnormal lab results (please specify): _____

❑ other anomalies: _____

Request

❑ **Skeletal Dysplasia: Metaphyseal and Epiphyseal Dysplasia with and without Spinal Involvement (81 Genes, SKT01)**
*ACAN, ACP5, AIFM1, ANKH, B3GALT6, B3GAT3, B4GALT7, BGN, BPNT2, CANT1, CCN6, CDKN1C, CFAP410, CHST3, COL10A1, COL11A1, COL11A2, COL2A1, COL27A1, COL9A1, COL9A2, COL9A3, COMP, CSGALNACT1, DDR2, DDRGK1, DNAJC21, DYM, EFL1, EIF2AK3, ERI1, EXOC6B, EXTL3, FGFR3, FLNB, FN1, GPX4, GZF1, HSPA9, HSPG2, IARS2, INPPL1, KIF22, LBR, LONP1, LTBP3, MATN3, MBTPS1, MMP13, MMP9, NANS, NEPRO, NKX3-2, NPR2, PAPSS2, PCYT1A, PISD, POLE, POP1, PTH1R, RAB33B, RMRP, RNU4ATAC, RPL13, RSPRY1, RUNX2, SBDS, SFRP4, SLC10A7, SLC26A2, SLC39A13, SMARCAL1, SRP54, TMEM165, TONSL, TRAPPC2, TRIP11, TRPS1, TRPV4, UFSP2, XYLT1**
* without repeat analysis

❑ **Micromelic Dysplasia: Acromelic, Acromesomelic, Mesomelic and Rhizo-Mesomelic Dysplasia (24 Genes, SKT04)**
ADAMTS10, ADAMTS17, ADAMTSL2, BMPR1B, DONSON, DVL1, DVL3, FBN1, FZD2, GDF5, GPC6, IHH, LBR, LTBP3, NPR2, PDE4D, PKDCC, PRKAR1A, PRKG2, ROR2, SHOX, SMAD4, TRPS1, WNT5A

❑ **Achondroplasia, Hypochondroplasia, and Pseudoachondroplasia (2 Genes, SKT16)**
FGFR3 (whole gene), *COMP*

❑ **Cleidocranial Dysplasia and Related Disorders (7 Genes, SKT17)**
CBFB, FIG4, LMNA, MSX2, RNU12, RUNX2, ZMPSTE24

❑ **Short-rib Dysplasia (24 Genes, SKT05)**
CEP120, CFAP410, CSPP1, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, DYNLT2B, EVC, EVC2, IFT122, IFT140, IFT172, IFT43, IFT52, IFT80, IFT81, INTU, KIAA0586, KIAA0753, NEK1, TTC21B, WDR19, WDR35

❑ **Chondrodysplasia Punctata (8 Genes, SKT06)**
AGPS, ARSL, EBP, GNPAT, LBR, MGP, NSDHL, PEX7

❑ **Osteogenesis Imperfecta and Related Skeletal Dysplasias with Decreased Bone Density (37 Genes, SKT07)**
ALPL, ANO5, B3GALT6, B4GALT7, BMP1, CCDC134, COL1A1, COL1A2, COPB2, CREB3L1, CRTAP, FKBP10, IFIH1, IFITM5, KDELR2, LRP5, MBTPS2, MESD, NBAS, P3H1, P4HB, PLOD2, PLS3, PPIB, SEC24D, SERPINF1, SERPINH1, SGMS2, SLC34A1, SP7, SPARC, TAPT1, TENT5A, TMEM38B, TNFRSF11B, WNT1, XYLT2

❑ **Osteopetrosis and Related Skeletal Dysplasias with Increased Bone Density (31 Genes, SKT08)**
AMER1, ANKH, CA2, CLCN7, CSF1R, CTSK, DLX3, FAM20C, FERMT3, GJA1, HPGD, LEMD3, LRP4, LRP5, LRRK1, OSTM1, PLEKHM1, PTDSS1, SFRP4, SLC29A3, SLCO2A1, SNX10, SOST, SQSTM1, TBXAS1, TCIRG1, TGFB1, TNFRSF11A, TNFRSF11B, TNFSF11, ZNF687

❑ **Hypophosphatemic Rickets and Related Skeletal Dysplasias with Abnormal Mineralization (20 Genes, SKT09)**
ALPL, ANKH, AP2S1, CASR, CLCN5, CYP27B1, CYP2R1, CYP3A4, DMP1, ENPP1, FAM20C, FGF23, OCRL, PHEX, PTH1R, SLC2A2, SLC34A1, SLC34A3, TRPV6, VDR

❑ **Limb Malformations: Brachydactyly, Reduction Defects, Synostoses, Ectrodactyly, Polydactyly, Syndactyly, and Selected Genetic Syndromes with Limb Malformations (73 Genes, SKT10)**
ARHGAP31, BHLHA9, BMP2, BMPR1B, CCNQ, CDH3, CHSY1, CKAP2L, DHODH, DLL4, DLX5, DOCK6, DONSON, EFNB1, EFTUD2, EOGT, ERI1, ESCO2, EVC2, FGF10, FGF9, FGFR1, FGFR2, FGFR3, FLNA, FLNB, GDF5, GDF6, GJA1, GLI1, GLI2, GLI3, GPC4, HOXA13, HOXD13*, IHH, IQCE, KIAA0825, KIF7, LMBR1, LRP4, MAP3K20, MAP3K7, MECOM, MEGF8, MYCN, NOG, NOTCH1, PDE3A, PDE4D, PRKAR1A, PTHLH, RAB23, RBM8A, RBPJ, RECQL4, ROR2, SALL1, SALL4, SF3B4, SMO, SMOC1, TBX15, TBX3, TBX4, TBX5, TOP2B, TP63, TRPV4, UBA2, WNT10B, WNT7A, YY1AP1*
* without repeat analysis

❑ **Craniosynostosis (40 Genes, SKT11)**
AHDC1, ALPL, ALX4, ASXL1, CDC45, COLEC11, CYP26B1, EFNB1, ERF, FGF9, FGFR1, FGFR2, FGFR3, FREM1, HNRNPK, IFT122, IFT140, IFT43, IHH, IL11RA, KAT6A, MASP1, MEGF8, MSX2, P4HB, POR, RAB23, RECQL4, RNU12, RUNX2, SCARF2, SEC24D, SKI, SMAD6, TCF12, TFAP2B, TWIST1, WDR19, WDR35, ZIC1

■ **Lysosomal Storage Disorders with Skeletal Involvement (SKT14)**
Please note: Replaced as part of **MET-02 „Lysosomal Storage Disorders“**. Please use the order form **„Metabolic incl. Mitochondrial Disorders“**.

❑ **Craniofacial and Patellar Dysostoses; Dysostoses with Vertebral (and Costal) Inolvement: Klippel Feil Syndrome, Meier Gorlin Syndrome, and Related Disorders (50 Genes, SKT15)**
ABCC9, ALX1, ALX3, ALX4, BMPER, CDC45, CDC6, CDK10, CDT1, DHODH, DLL3, DONSON, EDN1, EDNRA, EFNB1, EFTUD2, EIF4A3, EVC2, FOXI3, GDF6, GNAI3, HES7, KAT6B, LFNG, LMX1B, MEOX1, MESP2, MNX1, MYO18B, ORC1, ORC4, ORC6, PDE4D, PLCB4, POLR1A, POLR1B, POLR1C, POLR1D, PRKAR1A, SF3B2, SF3B4, SNRPB, SPECC1L, TBX4, TBX6, TCOF1, TMCO1, TMEM53, TWIST1, TXNL4A*
* without repeat analysis

❑ **Multiple Exostoses (2 Genes, SKT18)**
EXT1, EXT2

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Skeletal Disorders

Indication & Request

If a genetic syndrome with skeletal involvement is suspected, we recommend a trio exome or single exome analysis. We are also happy to accept individual requests and put together a suitable gene set.

Request Array-CGH

- ❑ Please perform array-CGH diagnostics
  - ❑ prior **or**
  - ❑ parallel

  to panel diagnostics.

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