MVZ Medizinisches Versorgungszentrum des Universitätsklinikums Köln

Cooperation partner
Institute of Human Genetics
University Hospital of Cologne

PATIENT (use sticker if available)

Last name _____

First name(s) _____

Date of birth _____

Address _____

_____

female o **male o**

Ethnic background _____
(may be important in recessive conditions)

Billing

Test will be paid by ☐ Referring facility ☐ Patient

Please note that international requests must be accompanied by a confirmation of payment. Please contact us for details.

Please ship samples to:

Institute of Human Genetics / MVZ
University Hospital of Cologne
Kerpener Str. 34, 50931 Cologne, Germany

Phone +49-221-478-86811, Fax +49-221-478-86812

Genetic testing can only be performed if samples are accompanied by a completed and signed consent form and the completed request form on page 2

Informed consent form

(according to German Genetic Diagnostics Act)

The planned genetic test, its limitations and possible interpretations of results have been explained to me in detail by the physician stated below. I have had the opportunity to discuss the details and ask questions about this information. By signing this form, I consent that genetic testing will be performed for the following disease/condition/diagnosis:
(to be entered by physician)

_____

I consent that the required sample (e.g. blood, tissue, amniotic fluid) will be taken.
The sample and the results of the testing may be used as follows:

I consent that **remaining sample material** will be **stored** for verification of results and quality management purposes.
I consent that **remaining sample material** will be **stored** to be available for new diagnostic options in the future.
I consent that the **test results and records** will not be destroyed after 10 years — as laid down in German statutory provisions — but will be **stored**.
I consent that the request for testing and all personal details required for the testing are **forwarded** to the Institute of Human Genetics, University Hospital of Cologne or if necessary to a specialized cooperating laboratory.
I wish to be informed on **incidental findings** that may be discovered by the genetic testing, even if they do not directly relate to the above mentioned disease/condition/diagnosis in question.
I consent that **remaining sample material** will be used for **research on the causes** and improved treatment of genetic diseases.
I consent that **data/results** that may be generated on the condition in question will be used in de-identified (pseudonymized) form for **academic or medical teaching** and for **scientific purposes** and will be published in anonymized form in **scientific journals**.
I consent that the results of the testing may be sent to the following physicians: _____

- Please delete as appropriate -

I am free to withdraw any of the above statements at any time. I have had enough time to consider my decision.

Place, Date	Signature Patient/ Parent/Legal Guardian	Name of Physician in printed characters	Stamp and Signature of Physician
_____	_____	_____	_____

►Please give a copy of the completed and signed consent form to the patient◄

Please ship samples to: Institute of Human Genetics / MVZ, University Hospital of Cologne, Kerpener Str. 34, 50931 Cologne, Germany

Request for molecular genetic testing

Contact: Prof. Dr. Christian Netzer, Phone +49-221-478-86811; christian.netzer@uk-koeln.de

Clinical Findings/Diagnosis/Indication/Reason for Testing

_____

_____

_____

Has any previous genetic test been performed in the patient relating to the above Reason for Testing?

☐ no

☐ yes (please specify previous findings/genetic tests)

_____

_____

Is Index Patient Known?

☐ yes (please specify mutation, disorder and relationship)

_____

_____

☐ yes; but there are no or incomplete information on the index patient. In this case a specific reason has to be given for the genetic mutation analysis on your patient. The statement **must** include the probability of predisposition of your patient or the remaining lifetime risk of developing the disease.

_____

_____

_____

☐ no

Type of Testing

☐ diagnostic testing
☐ prenatal testing
☐ predictive testing
☐ heterozygosity/carrier testing

Type of Sample

☐ blood
☐ DNA
☐ buccal swab
☐ saliva
☐ fibroblasts
☐ chorion villi
☐ other: _____

Date of Sample Collection

Sample was collected on: _____

Family History / Pedigree

Familiy history with regard to the current request for testing (please tick box(es) as appropriate)

☐ significant family history (see above information on index patient)

☐ family history not available

☐ no significant family history

☐ both parents clearly not affected

☐ mother clearly not affected, no information on father available

☐ father clearly not affected, no information on mother available

Parental consanguinity

☐ no

☐ yes (please specify):

_____

Pedigree (optional)

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Request for molecular genetic testing

NEUROMUSCULAR DISORDERS (Contact: Dr. med. Raoul Heller, 0221/478-86832, raoul.heller@uk-koeln.de)

Spinal muscular atrophy type I-IV (SMA); recessive

- ☐ *SMN1* deletion test (MLPA)
- ☐ *SMN1* carrier test (MLPA)
- ☐ *SMN1* point mutation analysis (sequencing, on inquiry)
- ☐ *SMN2* (MLPA)

X-linked SMA; X recessive

- ☐ *UBA1* (sequencing)

Spinal muscular atrophy (SMA); dominant

- ☐ *BICD2* (sequencing)
- ☐ *TRPV4* (sequencing)

Spinal muscular atrophy with respiratory distress type 1 (SMARD1), diaphragmatic SMA (DSMA1); recessive

- ☐ *IGHMBP2* (sequencing, MLPA)

Pontocerebellar hypoplasia (PCH 2D); dominant

- ☐ *SEPSECS* (sequencing)

Pontocerebellar hypoplasia (PCH 2 and 4); recessive

- ☐ *TSEN54 (*sequencing)
- ☐ *TSEN2* (sequencing)
- ☐ *TSEN34* (sequencing)
- ☐ *RARS2* (sequencing)
- ☐ *CASK* (sequencing)
- ☐ other (on inquiry)

Amyotrophic lateral sclerosis (ALS); familial

- ☐ *SOD1* (sequencing)
- ☐ *ALS2* (sequencing)
- ☐ *VAPB* (sequencing
- ☐ *FUS* (sequencing)
- ☐ *TARDBP* (sequencing)

Arthrogryposis (AMC), distal (DA1, DA2A, DA2B, DA3, DA5, DA7); dominant

- ☐ *TPM2* (sequencing)
- ☐ *TNNI2* (sequencing)
- ☐ *TNNT3* (sequencing)
- ☐ *PIEZO2* (sequencing)
- ☐ *MYH3* (sequencing)
- ☐ *MYH8* (sequencing)
- ☐ *MYBPC1* (sequencing)
- ☐ other (on inquiry)

Fetal akinesia (FADS) (Pena-Shokeir); Congenital myasthenia; recessive

- ☐ *RAPSN* (sequencing)
- ☐ *CHRNG* (sequencing)
- ☐ *CHRND* (sequencing)
- ☐ *CHRNE* (sequencing)
- ☐ *CHRNA1* (sequencing)
- ☐ *DOK7* (sequencing)
- ☐ *MUSK* (sequencing)
- ☐ *COLQ* (sequencing)
- ☐ other (on inquiry)

MUSCULAR DISORDERS (Contact: Dr. med. Dipl. Chem. Sebahattin Cirak, 0221/478-86834, sebahattin.cirak@uk-koeln.de)

Congenital myopathy; dominant / recessive

- ☐ *ACTA1* (sequencing)
- ☐ *SEPN1* (sequencing)
- ☐ *MYH7* (sequencing)
- ☐ *MEGF10* (sequencing)
- ☐ other (on inquiry)

Muscular dystrophy-dystroglycanopathy; recessive

- ☐ *ISPD* (sequencing)
- ☐ *FKRP* (sequencing)
- ☐ *POMK* (sequencing)
- ☐ *POMT1* (sequencing)
- ☐ *POMT2* (sequencing)
- ☐ *POMGNT1* (sequencing)
- ☐ *POMGNT2* (sequencing)
- ☐ *CAV3* (sequencing)
- ☐ *ANO5* (sequencing)
- ☐ *B3GNT1* (sequencing)
- ☐ *B3GALNT2* (sequencing)
- ☐ *TMEM5* (sequencing)
- ☐ *LARGE* (sequencing)
- ☐ *FKTN* (sequencing)
- ☐ *GMPPB* (sequencing)
- ☐ *CAPN3* (sequencing)
- ☐ *DYSF* (sequencing)

Duchenne muscular dystrophy; Becker muscular dystrophy; X-recessive

- ☐ *DMD* (sequencing, MLPA)

Ullrich congenital muscular myopathy / Bethlem myopathy; dominant / recessive

- ☐ *COL6A1* (sequencing)
- ☐ *COL6A2* (sequencing)
- ☐ *COL6A3* (sequencing)

Muscular dystrophy; dominant

- ☐ *LMNA* (sequencing)
- ☐ *DNAJB6* (sequencing)

CARDIOMYOPATHIES (Contact: Prof. Dr. med. Christian Netzer, 0221/478-86811, christian.netzer@uk-koeln.de)

Hypertrophic cardiomyopathy; dominant / recessive

- ☐ *MYH7* (sequencing)
- ☐ *MYBPC3* (sequencing)
- ☐ *TNNT2* (sequencing)
- ☐ *TNNI3* (sequencing)
- ☐ *TPM1* (sequencing)

see next page for further tests

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Request for molecular genetic testing

KIDNEY DISORDERS (Contact: Dr. med. Bodo Beck, 0221/478-86824, bodo.beck@uk-koeln.de; Florian Erger, 0221/478-86828, florian.erger@uk-koeln.de)

Nephrotic syndrome (NS); recessive

- ☐ *NPHS1* (sequencing)
- ☐ *NPHS2* (sequencing)
- ☐ *PLCE1 (NPHS3)* (sequencing)
- ☐ *LAMB2* (sequencing)
- ☐ *SMARCAL1* (Schimke dysplasia) (sequencing)
- ☐ *ARHGDIA* (sequencing)
- ☐ *PTPRO* (sequencing)
- ☐ *DGKE* (sequencing)
- ☐ *MYO1E* (sequencing)
- ☐ *COQ2* (sequencing)
- ☐ *COQ6* (sequencing)
- ☐ *ADCK4* (sequencing)
- ☐ *PDSS2* (sequencing)

Nephrotic syndrome (NS); dominant

- ☐ *WT1* (sequencing, MLPA)
- ☐ *LMX1B* (nail patella syndrome) (sequencing)
- ☐ *TRPC6* (sequencing)
- ☐ *INF2* (sequencing)
- ☐ *ACTN4* (sequencing)

Thrombotic miroangiopathy(TMA)

Atypical hemolytic-uremic Syndrome (aHUS); dominant / recessive

- ☐ *CFH* (sequencing)
- ☐ *CD46 (MCP)* (sequencing)
- ☐ *CFI* (sequencing)
- ☐ *C3* (sequencing)
- ☐ *MMACHC* (sequencing)
- ☐ *DGKE* (sequencing)
- ☐ *CFB* (sequencing)
- ☐ *THBD* (sequencing)

Thrombotic thrombocytopenic purpura; recessive

- ☐ *ADAMTS13* (sequencing)

Urolithiasis/Nephrocalcinosis

Primary hyperoxaluria type 1; recessive

- ☐ *AGXT* (sequencing, MLPA)

Primary hyperoxaluria type 2; recessive

- ☐ *GRHPR* (sequencing, MLPA)

Primary hyperoxaluria type 3; recessive

- ☐ *HOGA1* (sequencing)

Dent disease; X-recessive

- ☐ *CLCN5* (sequencing)

Hypercalcemia; recessive

- ☐ *CYP24A1* (sequencing)
- ☐ *SLC34A1* (sequencing)

Cysteinuria; recessive

- ☐ *SLC3A1* (sequencing)
- ☐ *SLC7A9* (sequencing)

CASR diseases; dominant

- ☐ *CASR* (familial hypocalciuric hypercalcemia) (sequencing)
- ☐ *CASR* (familial hypocalcemia with hypercalciuria) (sequencing)

Renal tubular dysgenesis (RTD); recessive

- ☐ *ACE* (sequencing)
- ☐ *AGT* (sequencing)
- ☐ *AGTR1* (sequencing)
- ☐ *REN* (sequencing)

Autosomal dominant tubulointerstitial nephropathy, (ADTKD) / MCKD; dominant

- ☐ *UMOD* (*MCKD2*) (sequencing)
- ☐ *HNF1B* (sequencing, MLPA)
- ☐ *REN* (sequencing)

Interstitial nephritis, karyomegalic; recessive

- ☐ *FAN1* (sequencing)

Urinary tract malformations (CAKUT); dominant

- ☐ *HNF1B* (RCS) (sequencing, MLPA)
- ☐ *PAX2* (RCS) (sequencing)
- ☐ *RET* (sequencing)

Nephronophtisis; recessive

- ☐ *NPHP1* (sequencing, MLPA)
- ☐ *NPHP2* (sequencing)
- ☐ *NPHP3* (sequencing)
- ☐ *BBS10* (sequencing)

Alport syndrome; X-dominant

- ☐ *COL4A5* (sequencing, MLPA)

Fechtner syndrome (Alport-like-syndrome); dominant

- ☐ *COL4A5* (sequencing, MLPA)

Alport syndrome; recessive

- ☐ *COL4A3* (sequencing)
- ☐ *COL4A4* (sequencing)

Polycystic kidney and hepatic disease (ARPKD); recessive

- ☐ *PKHD1* (sequencing)

Polycystic kidney and hepatic disease (ADPKD); recessive

- ☐ *PKD1* (sequencing, MLPA)
- ☐ *PKD2* (sequencing, MLPA)

Tubulopathies / Hereditary hypertension

Gitelman syndrome; recessive

- ☐ *SLC12A3* (sequencing)

EAST (SESAME) syndrome; recessive

- ☐ *KCNJ10* (sequencing)

Polyhydramnion / transient Bartter syndrome; X-recessive

- ☐ *KCNJ10* (sequencing)

Hereditary hypertension

Adrenal adenoma / hyperaldosteronism

- ☐ *KCNJ5* (adrenal aldosterone producing adenoma (APA) (sequencing)
- ☐ *RMND1* (sequencing)
- ☐ *KCNJ5* (familial hyperaldosteronism)

Liddle syndrome; dominant

- ☐ *SCNN1B* (ß-ENac) (sequencing)
- ☐ *SCNN1G* (γ-ENac) (sequencing)

Pseudohypoaldosteronism; dominant

- ☐ *NR3C2* (MCR) (sequencing)

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Request for molecular genetic testing

TUMOR DISEASES (Contact: Dr. med. Julia Schreml, 0221/478-86612, julia.schreml@uk-koeln.de (1) Dr. med. Bodo Beck, 0221/478-86824, bodo.beck@uk-koeln.de (2)

Hereditary kidney tumors [2]

Renal cell carcinoma (RCC) / neurocutane syndrome

- ☐ *VHL* (sequencing, MLPA)
- ☐ *FLCN* (Birt Hogg Dubé) (sequencing, MLPA)
- ☐ *MAX* (PCC syndrome) (sequencing)
- ☐ *TSC1* (sequencing, MLPA)
- ☐ *TSC2* (sequencing, MLPA)

Nephroblastoma

- ☐ *WT1* (Denys Drash) (sequencing, MLPA)
- ☐ *DIS3L2* (Perlman syndrome) (sequencing)

Endocrine tumors[1]

Multiple endocrine neoplasia; dominant

- ☐ *VHL* (sequencing, MLPA)
- ☐ *MEN1* (sequencing)
- ☐ *RET* (sequencing)
- ☐ *CDKN1B* (sequencing)

Hereditary paraganglioma-pheochromocytoma syndrome; dominant

- ☐ *RET* (sequencing)
- ☐ *VHL* (sequencing, MLPA)
- ☐ *SDHA* (sequencing)
- ☐ *SDHB* (sequencing)
- ☐ *SDHC* (sequencing)
- ☐ *SDHD* (sequencing)
- ☐ *SDHAF2* (sequencing)
- ☐ *MAX* (sequencing)

MICROCEPHALIES (Contact: Prof. Dr. med. Christian Netzer, 0221/478-86811, christian.netzer@uk-koeln.de)

Primary microcephaly; recessive

- ☐ *CEP152* (sequencing)
- ☐ *WDR62* (sequencing)
- ☐ *ASPM* (sequencing)
- ☐ *MCPH1* (sequencing)
- ☐ *CDK5RAP2* (sequencing)
- ☐ *ZNF335* (sequencing)
- ☐ *CENPJ* (sequencing)
- ☐ *CASC5* (sequencing)
- ☐ *STIL* (sequencing)
- ☐ *CEP63* (sequencing)
- ☐ *CEP135* (sequencing)
- ☐ *PNKP* (sequencing)

Seckel syndrome; recessive

- ☐ *CEP152* (sequencing)
- ☐ *CENPJ* (sequencing)
- ☐ *CDK5RAP2* (sequencing)
- ☐ *RBBP8* (sequencing)
- ☐ *ATR* (sequencing)
- ☐ *ATRIP* (sequencing)
- ☐ *DNA2* (sequencing)
- ☐ *NIN* (sequencing)

Microcephalic osteodysplastic primordal dwarfism type I (MOPD1); recessive

- ☐ *RNU4atac* (sequencing)

LIG4-syndrome/ LIG4-syndrome-like

- ☐ *LIG4* (sequencing)
- ☐ *XRCC4* (sequencing)

Microcephalic osteodysplastic primordal dwarfism type II (MOPD2); recessive

- ☐ *PCNT* (sequencing)

Nijmegen breakage syndrome; recessive

- ☐ *NBS1* (sequencing)

Nijmegen breakage syndrome-like

- ☐ *RAD50* (sequencing)

Other microcephaly / dwarfism sindrome; recessive

- ☐ *NHEJ1* (sequencing)
- ☐ *MRE11* (sequencing)
- ☐ *BRAT1* (sequencing)

KABUKI SYNDROME AND MENTAL RETARDATION SYNDROMES (Contact: Prof. Dr. med. Christian Netzer, 0221/478-86811, christian.netzer@uk-koeln.de)

Kabuki syndrome; dominant

- ☐ *MLL2* (sequencing)
- ☐ *KDM6A* (sequencing)

Coffin Siris syndrome; dominant

- ☐ *ARID1B* (sequencing, MLPA)
- ☐ *ARID1A* (sequencing, MLPA)
- ☐ *SMARCA2* (sequencing)
- ☐ *SMARCA4* (sequencing)
- ☐ *SMARCB1* (sequencing)
- ☐ *SMARCE1* (sequencing)
- ☐ *PHF6* (clinically overlapping phenotype in female patients in first years of life) (sequencing)

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Request for molecular genetic testing

SKELETAL DISORDERS (Contact: Prof. Dr. med. Christian Netzer, 0221/478-86811, christian.netzer@uk-koeln.de)

Osteogenesis imperfecta (OI) type I — IV; dominant

- ☐ *COL1A1* (sequencing, MLPA)
- ☐ *COL1A2* (sequencing, MLPA)

Osteogenesis imperfecta (OI) type V; dominant

- ☐ *IFITM5* (sequencing)

Osteogenesis imperfecta (OI) type IIB, VI, VII, X, XIV; recessive

- ☐ *CRTAP* (sequencing)
- ☐ *FKBP10* (sequencing)
- ☐ *LEPRE1* (sequencing)
- ☐ *PPIB* (sequencing)
- ☐ *SERPINH1* (sequencing)
- ☐ *SPARC* (sequencing)
- ☐ *SERPINF1* (sequencing)
- ☐ *SP7* (sequencing)
- ☐ *BMP1* (sequencing)
- ☐ *WNT1* (sequencing)
- ☐ *TMEM38B* (sequencing)
- ☐ *SEC24D* (sequencing)

Osteoporosis with fractures; X-dominant

- ☐ *PLS3* (sequencing)

Bruck syndrome; recessive

- ☐ *PLOD2* (sequencing)

Hypophosphatasia; dominant, recessive

- ☐ *ALPL* (sequencing)

Juvenile Osteoporosis; dominant

- ☐ *LRP5* (sequencing)
- ☐ *WNT1* (sequencing)
- ☐ *COL1A1* (sequencing, MLPA)
- ☐ *COL1A2* (sequencing, MLPA)

Osteoporosis pseudo glioma syndrome; recessive

- ☐ *LRP5* (sequencing)

Osteopetrosis type 1; dominant

- ☐ *LRP5* (sequencing)

Cole-Carpenter syndrome; dominant

- ☐ *P4HB* (sequencing)

Cole-Carpenter syndrome; recessive

- ☐ *SEC24D* (sequencing)
- ☐ *LEPRE1* (sequencing)

Craniolenticulosutural dysplasia; recessive

- ☐ *SEC23A* (sequencing)

HEARING DISORDERS (Contact: Prof. Dr. med. Christian Netzer, 0221/478-86811, christian.netzer@uk-koeln.de)

Hearing loss; recessive

- ☐ DFNB1 locus (*GJB2* (sequencing) and if appropriate *GJB6* MLPA of junction fragment in case of Δ[GJB6-D13S1830] and Δ[GJB6-D13S1854]

Aminoglycoside-induced hearing loss

- ☐ *MT-RNR1* (12s rRNA) (sequencing)

Pendred syndrome/DFNB4; recessive

- ☐ *SLC26A4* (sequencing)

MULTISYSTEM DISORDERS (Contact: Prof. Dr. med. Christian Netzer, 0221/478-86811, christian.netzer@uk-koeln.de (1)
Dr. med. Bodo Beck, 0221/478-86824, bodo.beck@uk-koeln.de (2)

Cystic fibrosis; recessive [1]

- ☐ *CFTR* (OLA, hot spots)
- ☐ *CFTR* (sequencing, MLPA)

MODY diabetes (Maturity onset diabetes of the young); dominant [2]

- ☐ *HNF1A* (sequencing, MLPA)
- ☐ *HNF1B* (sequencing, MLPA)
- ☐ *POMC* (sequencing)
- ☐ *HNF4A* (sequencing, MLPA)
- ☐ *GCK* (sequencing, MLPA)

Pancreatitis; dominant [1]

- ☐ *PRSS1* (sequencing)

Aniridia / WAGR syndrome (Wilms tumor, aniridia, urogenital anomalies, mental retardation); dominant [2]

- ☐ *PAX6* (sequencing, MLPA)

Porphyria; recessive [1]

- ☐ *ALAD* (Doss porphyria) (sequencing)

Methylmalonic aciduria/homocysteinuria; recessive [2]

- ☐ *MMACHC* (sequencing)
- ☐ *MMADHC* (sequencing)
- ☐ *CBS* (sequencing)

Loeys Dietz syndrome; dominant [1]

- ☐ *TGFBR1* (sequencing)
- ☐ *TGFBR2* (sequencing, MLPA)
- ☐ *TGFB2* (sequencing)
- ☐ *SMAD3* (sequencing)

Marfan syndrome; dominant [1]

- ☐ *FBN1* (sequencing, MLPA)

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Request for molecular genetic testing

METABOLIC DISORDERS (Contact: Dr. med. Raoul Heller, 0221/478-86832, raoul.heller@uk-koeln.de)

Molybdenum cofactor deficiency; recessive

- ☐ *MOCS1* (sequencing)
- ☐ *MOCS2* (sequencing)
- ☐ *GPHN* (sequencing)

Sulfite oxidase deficiency, recessive

- ☐ *SUOX* (sequencing)

CRANIOFACIAL MALFORMATION SYNDROMES (Contact: Prof. Dr. med. Christian Netzer, 0221/478-86811, christian.netzer@uk-koeln.de)

Syndromic craniosynostosis; dominant
(e.g. Apert, Pfeiffer, Crouzon, Saethre-Chotzen, Muenke syndromes)

- ☐ *FGFR1* (sequencing, hot spots)
- ☐ *FGFR2* (sequencing, hot spots)
- ☐ *FGFR3* (sequencing, hot spots)
- ☐ *TWIST* (sequencing, MLPA)

Craniosynostosis and dental anomalies; recessive

- ☐ *IL11RA* (sequencing)

Craniosynostosis type 4; dominant

- ☐ *ERF* (sequencing)

Hypochondroplasia, achondroplasia; dominant

- ☐ *FGFR3* (sequencing, hot spots)

LADD syndrome, ALSG syndrome; dominant

- ☐ *FGF10* (sequencing, MLPA)
- ☐ *FGFR2* (sequencing, TK domain)
- ☐ *FGFR3* (sequencing, TK domain)

Sample and shipping requirements

5-10 ml EDTA blood / ≥10 ml amniotic fluid/chorion villi / ≥500 ng DNA / for RNA analysis 5-10 ml PAXgene blood;
1-2 ml EDTA blood acceptable for newborns and infants (please contact us).
Please contact us before shipping samples for prenatal diagnosis/in ongoing pregnancy.

Ship samples at room temperature. Please make sure that samples are **correctly labeled (name and dob)** !
Testing will only be performed if samples are accompanied by a completed and signed informed consent form (see page 1).