medgen.at
Ambulatorium für Humangenetische Diagnostik & Beratung

Auftrag zur labordiagnostischen Syndromdiagnostik

PATIENT
Zuname:
Vorname:
☐ männlich ☐ weiblich

SV-Nr.: _ _ _ _ Geburtsdatum: _ _ _ _ _ _

EINSENDER
Ansprechpartner (Druckschrift):

Telefon:
Abnahmedatum:
Unterschrift:
Klinikstempel/Praxisstempel:

KOSTENTRÄGER:

☐ Einsender

☐ Patient (*Bitte unbedingt vom Patienten unterschriebene* ***Kostenübernahmeerklärung*** *inkl. Kontaktadresse beilegen!)*

Screeninguntersuchungen

☐ **Chromosomen** (5 ml Heparinblut)
☐ **CGH/SNP Array** (2 ml EDTA Blut)
☐ **MLPA Subtelomere** (für alle MLPA Untersuchungen insgesamt 2 ml EDTA Blut)
☐ **MLPA Mikrodeletions-/Mikroduplikationssyndrome**
☐ **MLPA X-assoziierte mentale Retardierungssyndrome**

Spezifische Fragestellungen

(*H*: 5 ml Heparinblut; *E*: 2 ml EDTA Blut)

- o Achondroplasie *E*
- o Adrenogenitales Sy CYP21A2 *HE*
- o Alagille Sy *E*
- o Angelman *HE*
- o Apert *E*
- o Azoospermie AZF *HE*
- o Beckwith-Wiedemann *HE*
- o Blackfan-Diamond ° RPS19 *E*
- o CATCH 22/di George *HE*
- o Cong. nephrot. Sy *E:*
  ° NPHS1; ° NPHS2; ° WT1
- o Cornelia de Lange *HE*
- o Cri du Chat *HE*
- o CVID *E:*
  ° TNFRSF13B; ° TNFRSF13C
- o Cystische Fibrose *E*
- o Denys-Drash-/Frasier *E*
- o Fam. Mittelmeerfieber *E*
- o Fragiles X *HE*
- o Fructoseintoleranz *E*
- o Hämochromatose *E*
- o Hered. Albright Osteodystr. *E*
- o Hered. Pankreatitis *E*
- o Holt-Oram *E*
- o Hyperbilirub., Meulengracht *E*
- o Hyperhomocysteinämie *E*
- o Hyper-IgM Immundef, X-chr *E*
- o ICF *H*
- o Kallmann Sy 1 *HE*
- o Kallmann Sy 2 *HE*
- o Laktoseintoleranz *E*
- o Lissenzephalie 1 *E:*
  ° DCX; ° PAFAH1B1; ° TUBA1A
- o Miller-Dieker *HE*
- o MODY Typ 2 *E*
- o MODY Typ 3 *E*
- o Nijmegen *HE*
- o Noonan *HE*
- o Osteopetrosis *E*:
  ° TCIRG1; ° OSTM1; ° ClCN7
- o Pelizaeus Merzbacher *HE*
- o Period. Fieber (Hyper-IgD) *E*
- o Period. Fieber (Muckle-Wells) *E*
- o Period. Fieber (TRAPS) *E*
- o Persist. Müller-Gang II *E*
- o Pfeiffer Sy *E*
- o Prader Willi *HE*
- o Premature Ovarian Failure *E:*
  ° FMR1; ° BMP15
- o Prog. myokl. Epilepsie (Unverricht-Lundborg) *E*
- o Pseudohypoaldost. I (MLR) *E*
- o Pseudohypoparathyr. I *E*
- o Rett *HE*
- o Rubinstein-Taybi *HE*
- o Silver-Russell *HE*
- o Sotos *HE*
- o Torsionsdystonie, aut. dom. *E*
- o Transthyretin Amyloidose *E*
- o Tumoröse Calcinose *E:*
  ° FGF23; ° GALNT3; ° KL
- o Wachstumshormoninsensitivitätssy *E*
- o WHIM *E*
- o Williams-Beuren *HE*
- o Wiskott Aldrich *E*
- o Wolf-Hirschhorn *HE*
- o X-chr. lymphoprolif. Erkr. (XLP) *E*
  ° SH2D1A; ° XIAP
- o X-Inaktivierungsmuster *E*
- o Zöliakie *E*
- o Zuckerintoleranz (LCT, ALDOB) *E*

Vollständiges Leistungsangebot entnehmen Sie bitte der Preisliste auf www.medgen.at

Andere Verdachtsdiagnose/Fragestellung:

Bemerkungen/klinische Daten:

Bitte legen Sie die EINVERSTÄNDNISERKLÄRUNG bei und beachten Sie die jeweils gültigen gesetzlichen Versandvorschriften der Post für biologisches Material!

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Informed consent for genetic testing

I, ... DOB ..... confirm that
(Last name, first name)

Mrs / Mr Dr. ..... has informed me about the nature,
(name of referring clinician)

the purpose, the effectiveness, the limitations, the benefits and risks of the planned genetic test.

I consent to have a sample from myself taken for the following genetic testing:

.....

I know that I can withdraw this consent at any time. In that case, I will notify medgen.at in written form.

I know that the remaining sample will be kept at medgen.at GmbH to allow verifiability of the results and to perform additional tests that may perhaps be necessary.

.....

(Place, date) (Signature of Patient) (Signature of referring clinician present)

The Austrian "Gentechnikgesetz" §69 states that genetic testing is solely allowed if there is a written statement of the person to be tested which confirms that beforehand the person has been informed by a specialized doctor (human genetics, medical genetics or a specialist for the medical field in question) about the nature, purpose and effectiveness of the test. Based on this information the person consents with his/her free will to the genetic test.