Centre of Molecular Diseases

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No molecular diagnosis is done without this registration form, clinical info, clinical photos, x-Rays, signed consent form and billing address.

PATIENT INFORMATION

Family name*:	**First Name*:**		**PMO laboratory #** *(do not fill)*
Date of Birth* (dd/mm/yyyy):	**Gender*:** F M	**Origin/Ethnic Background:**	**Known consanguinity:** *Please enclose pedigree if available* Yes No
Mother`s name: First: Last:			
Father`s name: First: Last:			
Reason for Referral:			
Clinical Diagnosis:			

SAMPLE INFORMATION

DNA *Minimum 10 µg*	Volume _____ µl	Blood in EDTA (3ml)	Fibroblasts	Other: *Please specify*
☐	Concentration _____ng/µl	☐	☐	☐
	Resuspended in (buffer) _____			_____

REQUIRED - PLEASE INCLUDE*:

These information is crucial for accurate interpretation of results.

Clinical info ☐ **Clinical photos** ☐ **X-Rays** ☐ **Signed consent form** ☐

PHYSICIAN INFORMATION

Title:	**Name:**
Address:	**Tel:** _____ **Fax:** _____ **E-mail*:** _____ *Preferred method to receive results* **Secure Email** ☐ **Regular Mail** ☐

BILLING INFORMATION

Billing Address*[1]:	**Patient`s address*:** *(for our internal billing system only if not identical to billing address):*

*Mandatory

[1] Please notify if any agreement has been made with our team on billing.

SAMPLE TREATMENT

DNA/RNA Extraction ☐	DNA Banking ☐	Cell culture[2] ☐	Prenatal Studies[3] ☐
Molecular analysis ☐	Exome *Minimum 10 µg of DNA* ☐	Other: *Please specify* ☐	_____

[2] Please contact the laboratory before sending the cells.
[3] Please contact the laboratory in advance; maternal contamination analysis is required with all prenatal studies.

Next Generation Sequencing – Gene Panels

Connective Tissue & Osteogenesis Imperfecta ☐
Coverage ~96%

ALDH18A1	Cutis laxa, 3A (PYCS)
ANTXR2	Hyaline fibromatosis S.
ATP6V0A2	Cutis laxa, 2A
B3GALT6	SEMD-JL Beighton
B3GAT3	Larsen-like, recessive
B4GALT7	EDS, progeroid
BMP1	OI, recessive
CANT1	Desbuquois
CHST14	EDS, contractural
CHST3	Larsen, AR
COL1A1	OI, dominant
COL1A2	OI, dominant
COL3A1	EDS, vascular
COL5A1	EDS, classical
COL5A2	EDS, classical
CRTAP	OI, recessive
FBN1	Acromicric dysplasia
FBN2	Arachnodactyly, cong.
FKBP10	Bruck syndrome 1
FKBP14	EDS, kyphoscoliotic
FLNB	Larsen, dominant
GORAB	Geroderma osteod.
IFITM5	OI type 5, dominant
IMPAD1	gPAPP dysplasia
KIF22	SEMD-JL, leptodactylic
LEPRE1	OI, recessive
PAPSS2	SEMD
PLOD1	EDS, kyphoscoliotic
PLOD2	Bruck syndrome 2
PPIB	OI, recessive
PYCR1	Cutis laxa, 2B
SERPINF1	OI, recessive
SERPINH1	OI, recessive
SLC26A2	Diastrophic dysplasia
SLC39A13	EDS, spondylocheirod.
SMAD3	Loeys-Dietz, 1C
SP7	OI, recessive
TGFBR1	Loeys-Dietz, 1
TGFBR2	Loeys-Dietz, 2
TNXB	EDS, hypermobile
XYLT1	Desbuquois 2

EDS = Ehlers-Danlos sydrome
OI = Osteogenesis Imperfecta
SEMD = Spondylo-epi-metaphyseal dysplasia
SEMD-JL = Spondylo-epi-metaphyseal dysplasia with joint laxity

Chondrodysplasias ☐
Coverage ~97%

ACP5	Spondyloenchondrodysplasia
ADAMTS10	Weill-Marchesani 1
ADAMTSL2	Geleophysic dysplasia
ALPL	Hypophosphatasia
COL10A1	Metaphyseal dysplasia Schmid
COL11A1	Stickler 2
COL11A2	Stickler 3, OSMED
COL2A1	SEDC group
COL9A1	MED, dominant
COL9A2	MED, dominant
COL9A3	MED, dominant
COMP	Pseudoachondroplasia
CUL7	3-M syndrome
DLL3	Spondylocostal dysplasia 1
DYM	Dyggve-Melchior-Clausen dysplasia
EXT1	Exostoses, multiple, 1
EXT2	Exostoses, multiple, 2
FAM111A	Kenny-Caffey syndrome
FGFR3	FGFR3 dysplasia group
FLNA	Filamin A dysplasia group
GDF5	Grebe dysplasia
GNAS	Fibrous dysplasia
GPC6	Omodysplasia
HES7	Spondylocostal dysplasia 4
HSPG2	Dyssegmental dysplasia
LFNG	Spondylocostal dysplasia 3
LRP5	Osteoporosis-pseudoglioma syndrome
MAFB	Multicentric carpotarsal osteolysis
MATN3	MED, dominant
MESP2	Spondylocostal dysplasia 2
MMP2	Torg-Winchester syndrome
NPR2	Acromesomelic dysplasia, Maroteaux
OBSL1	3-M syndrome
PDE4D	Acrodysostosis 2
PHEX	Hypophosphatemic rickets
PRKAR1A	Acrodysostosis 1
RMRP	Cartilage-Hair-Hypoplasia
ROR2	Robinow syndrome, recessive
RUNX2	Cleidocranial dysplasia
SBDS	Shwachman-Bodian-Diamond syndrome
SLC35D1	Schneckenbecken dysplasia
SOX9	Campomelic dysplasia
TBX6	Spondylocostal dysplasia 5
TRAPPC2	SED tarda, X-linked
TRIP11	Achondrogenesis 1A
TRPV4	Metatropic dysplasia
WISP3	Progressive pseudorheumatoid dysplasia

Lysosomal Storage Diseases ☐
Coverage ~98%

AGA	Aspartylglucosaminuria
ARSA	Metachromatic leucodystrophy
ARSB	MPS VI
ASAH	Farber disease
CLN3	Neuronal ceroid lipofuscinosis
CLN5	Neuronal ceroid lipofuscinosis
CLN6	Neuronal ceroid lipofuscinosis
CLN8	Neuronal ceroid lipofuscinosis
CTNS	Cystinosis
CTSA	Galactosialidosis
CTSC	Haim-Munk syndrome
CTSD	Neuronal ceroid lipofuscinosis
CTSK	Pycnodysostosis
DNAJC5	Neuronal ceroid lipofuscinosis
FUCA1	Alpha-Fucosidosis
GAA	Pompe disease
GALC	Krabbe disease
GALNS	MPS IV
GBA	Gaucher disease
GLA	Fabry disease
GLB1	GM1 gangliosidosis
GNE	Sialuria
GNPTAB	Mucolipidosis II
GNPTG	Mucolipidosis II
GNS	MPS IIID
GUSB	MPS VII
HEXA	GM2 gangliosidosis
HEXB	Sandhoff disease
HGSNAT	MPS IIIC (Sanfilippo C)
HYAL1	MPS IX
IDS	MPS II
IDUA	MPS I
LAMP2	Danon disease
LIPA	Wolman disease
MAN2B1	Alpha-Mannosidosis
MANBA	Beta-Mannosidosis
MCOLN1	Mucolipidosis IV
MFSD8	Neuronal ceroid lipofuscinosis
NAGA	Schindler disease
NAGLU	MPS IIIB
NEU1	Mucolipidosis I
NPC1	Niemann-Pick C
NPC2	Niemann-Pick C
PPT1	Neuronal ceroid lipofuscinosis
PSAP	Krabbe-like, MLD-like, Gaucher-like
SGSH	MPS IIIA
SLC17A5	Sialic acid storage disease
SMPD1	Niemann-Pick A+B
SUMF1	Multiple sulfatase deficiency
TPP1	Neuronal ceroid lipofuscinosis

Please note that results on NGS of whole panel regions are given on the indicate coverage (%).
For selected genes we complete uncovered coding regions if specifically required.

Without complete coverage ☐ With complete coverage (additional costs) ☐

Sanger Sequencing

GATM (AGAT)	AGAT deficiency
DLL3	Spondylocostal dysostosis 1
FAM111A - Exon 5	Kenny-Caffey syndrome
GAMT	GAMT deficiency
HES7	Spondylocostal dysostosis 4
IDH1 - Exon 4	Metaphyseal chondromatosis
IDH2 - Exon 4	Metaphyseal chondromatosis
IFITM5	OI, dominant, type 5
KIF22 - Exon 4	SEMD-JL leptodactylic
LFNG	Spondylocostal dysostosis 3
MAFB	Multicentric carpotarsal osteolysis
MESP2	Spondylocostal dysostosis 2
MMP2	Torg-Winchester syndrome
MMP13	Metaphyseal dysplasia, Spahr
MMP14	Winchester syndrome
RMRP	Cartilage-hair hypoplasia
ROR2	Robinow syndrome, recessive
SMAD3	Loeys-Dietz syndrome 3
TGFB1	Camurati-Engelmann disease
WISP3	Progressive pseudorheumatoid dysplasia
GAA	Pompe disease
SLC6A8	Creatine transporter deficiency
SLC2A1 (GLUT1)	Glucose transporter type 1 deficiency
Other gene[4]	_____

Testing for Known Mutation/Variant

Please provide copy of report if testing done at another laboratory

Name of Relative(s) *(father, mother, sister, cousin.etc…)* ***Familial Mutation*** *(gene, exon, cDNA, protein)*

[4] Only if discussed with the laboratory