Print form

EN-ISO 15189:2012

AGDx Application form
Laboratory Genome Dx and Genetic Metabolic Disorders
Amsterdam UMC, **locations AMC and VUmc**

Sample delivery address (office hours):
Postoffice H01-114, Meibergdreef 9, 1105 AZ AMSTERDAM
Outside office hours: delivery at LAKC B1-114

Tel. nr.: +31 20 566 5110 Fax nr.: +31 20 566 93 89
kg-dna@amc.nl GenomeDiagnostics.AmsterdamUMC.NL

Print and include this form when sending the patients sample

PATIENT INFORMATION

Last name:
First name:
Initials:
Date of birth:
Gender: ◯ Male ◯ Female
Your reference:
PO number:

ORDERING PHYSICIAN INFORMATION

Name:
Hospital:
Medical specialty:
Street/PObox:
ZIP code + Town:
AGB Code (for Dutch specialists only)**:**
Phone:
E-mail:
CC report:

Former family members samples known by AGDx: ◯ Yes ◯ No
Name:
Relation:
Date of Birth:
Family no. (when known)**:**

Family consanguinity: ◯ Yes (see pedigree – page 2) ◯ No

TESTS REQUEST (Invoice according to the actual website prices)

1) **Test name:** 1) **Test code:**
2) **Test name:** 2) **Test code:**
3) **Test name:** 3) **Test code:**

APPLICATION PURPOSE

☐ Confirmation clinical diagnosis
☐ Confirmation clinical suspicion
☐ Concluding diagnosis unknown
☐ Carrier testing (recessive disorder) Presymptomatic research
☐ Genotyping apropos of prenatal research
☐ Interpretation previously found sequence variation
☐ Other:
☐ Known familial variant (gene + variant):
☐ Whole gene or panel scanning:

EXPEDITED TESTING
◯ No ◯ Yes (Contact by phone obligatory)
Report needed before:
Phone contact with:.

Note: Prenatal research: 2 - 3 weeks
Familial mutation: 3 weeks (see page3)

☐ **Storage for future diagnostics**
Please indicate the patients' syndrome

☐ **Storage for research**
Project/code:
Project leader:

SAMPLE MATERIAL (Note: Fresh EDTA blood sample is required for cnv analysis within a panel)

☐ Blood
☐ EDTA
☐ PAX (RNA)
Extraction date:

☐ DNA (indicate DNA source):

☐ Biopsy / Tissue (NO muscle tissue)
Type:
☐ In paraffin

☐ Saliva
☐ Cheek mucosa / Swab
☐ Other material:

☐ Fibroblasts
☐ Chorionic Villi
☐ Amnios
☐ Cell culture

TO BE COMPLETED BY AGDx DNA-LABORATORY PERSONNEL

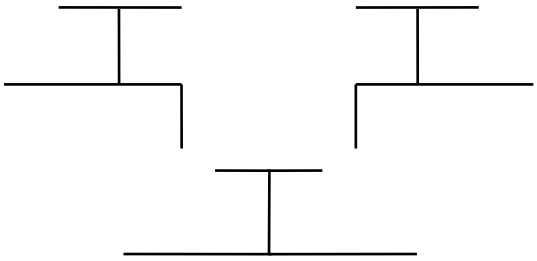
Initial for received material	*Date arrival*	
Amount:		

We only accept specimen with a COMPLETED APPLICATION FORM and each specimen container must be labelled with a NAME, DATE OF BIRTH and GENDER

PEDIGREE

Indicate patient with an arrow (↗)

Affected persons in full shading ■ Carriers in half shading ◧



CLINICAL INFORMATION

INFORMED CONSENT

The patient or his or her legal representative is informed by the applicant concerning the use and storage of the patients'sample.
See form Conditions for application AGDx. If there are any objections concerning the conditions, the applicant can indicate this below:

☐ The patient or his or her legal representative wishing to object concerning the use and storage of the patients' sample.
For additional questions contact kg-dna@amc.nl

SPECIMEN

Collect 2x 6-7 ml EDTA blood (**DO NOT FREEZE**; do not use 4 ml tubes). Infants 5-10 ml. Label all specimen containers with the patient's **NAME, DATE of BIRTH** and **GENDER**. For additional questions contact kg-dna@amc.nl

SHIPPING AND HANDLING INSTRUCTIONS

See form Shipping and handling instructions AGDx

Commercial site, for information only: http://www.un3373.com/un3373-packaging/

We only accept specimen with a COMPLETED APPLICATION FORM and each specimen container must be labelled with a NAME, DATE OF BIRTH and GENDER

- NGS panel genes are analysed with either quality A or quality C. For more transparency of NGS testing in a diagnostics setting see Matthijs G et al., Eur J Hum Genet 2015; doi: 10.1038.
- Quality A: Genes must be covered completely for 100%. Regions with low coverage (<30 reads) in the NGS test are analysed by addiotnal Sanger sequencing.
- Quality C: Gene coverage >95%. No addtional analyses in case a regions with a low coverage (<30 reads).
- CNV analyses, when included in the NGS panel for the detection of exon deletions /duplications, is exclusively possible on EDTA blood specimen.
- Analysis of familial variants, in genes not included in our standard panel (for example Index via WES analyses), might overdue 3 weeks concerning primer design and validation with a max TAT of 6 weeks.

OBESITY

Characteristics:

Length		cm	Autism	◯ Yes	◯ No
Weight		kg	Therapy resistance	◯ Yes	◯ No
Head circumference		cm	Early onset (<5 year)	◯ Yes	◯ No
Organ disorders (specify)			Hyperphagy	◯ Yes	◯ No
Dysmorphology (specify)			Dominant inheritance	◯ Yes	◯ No
			Mental disability	◯ Yes	◯ No
			Developmental delay	◯ Yes	◯ No

AGDx NGS – Obesitome panel (Test code: D0066)
MC4R- Gene sequencing (Test code: V00139)
MC4R – Mutation Specific testing (Test code: V00138)

DYSLIPIDEMIA

Cholesterol levels and medication

Date measurement lipid levels:			Medication:	◯ Yes ◯ No
Total cholesterol		mmol/l	Medication start date:	
LDL–cholesterol		mmol/l	Specify medication:	
HDL–cholesterol		mmol/l	Dose:	
Triglycerides		mmol/l	Specify medication:	
Apolipoprotein A1		g/l	Dose:	
Apolipoprotein B		g/l	Start CVD:	
Lipoprotein (a)		mg/l	Height:	
			Weight:	

◯ Treated ◯ Untreated

Clinical signs

Xanthoma	◯ Yes ◯ No	PTCA	◯ Yes ◯ No
Arcus lipoides	◯ Yes ◯ No	CABG	◯ Yes ◯ No
Xanthelasmata	◯ Yes ◯ No	Claudicatio	◯ Yes ◯ No
Myocardial Infarction	◯ Yes ◯ No	CVA	◯ Yes ◯ No
Angina pectoris	◯ Yes ◯ No	Hypertension	◯ Yes ◯ No

AGDx NGS – Dyslipidemia panel (Test code D00463)
AGDx NGS – Dyslipidemia panel plus CNV (Test code D00471)
MTHRF – Mutation specific testing (Test code D00063)

We only accept specimen with a COMPLETED APPLICATION FORM and each specimen container must be labelled with a NAME, DATE OF BIRTH and GENDER

EpiSign Diagnostic DNA Methylation Test

☐ EpiSign Complete including late onset disorders [EPI] **(Test code AUA0001)**

☐ EpiSign Complete excluding late onset disorders [EPI] **(Test code AUA0002)**

☐ EpiSign Variant [EPI] **(Test code AUA0003)**

Gene Variant

Is the variant Mosaic (See list of genes, page 3) ◯ ... ◯ No

Estimated % of Mosaic:

APPLICATION PURPOSE

Suitable for pa..... ntal delay or with one or more overlapping features, suggestive of one of the represented epigenetic signature conditions or imprinting disorders.

SAMPLE MATERIAL (Note: Fresh EDTA blood sample is required for cnv analysis within a .....

☐ EDTA blood (2 x 6 ml, do not freeze)

☐ DNA (minimum 5 µg, isolated from EDTA blood) Extraction date:

☐ Origin of DNA (if known): Age patient at extraction date:

ACCEPTANCE OF FINANCIAL RESPONSIBILITY FOR GENETIC TESTING

My signature indicates that I accept financial responsibility for all fees associated with this genetic testing order:

Signature of responsible party	Printed name of responsible party	Date

Print form

ADDRESS LABEL

---------✂----------

AGDx Laboratory Genome Diagnostics (H01-114)*

Amsterdam UMC

Meibergdreef 9

1105 AZ AMSTERDAM

The Netherlands

MEDICAL DIAGNOSTIC SAMPLE

URGENT SHIPPING!

*Outside office hours: delivery at LAKC B1-114

---------✂----------