Print form

EN-ISO 15189:2012

AGDx EpiSign 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/POobox:
ZIP code + Town:
AGB Code:
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 family tree – page 2) ◯ No

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): ◯ Yes ◯ No

Estimated % of Mosaic:

APPLICATION PURPOSE

Suitable for patients with developmental 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 panel)

☐ EDTA blood (ml)
☐ DNA (minimum 2 µg)
☐ Origin of DNA (if known): Extraction date:

TO BE COMPLETED BY AGDx DNA-LABORATORY PERSONNEL

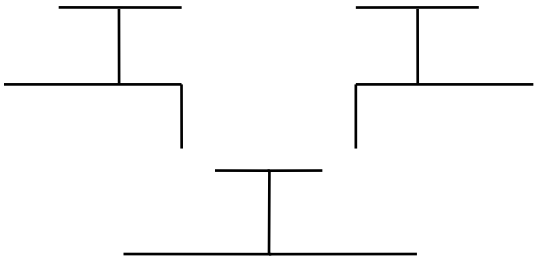
Initial for received material	❒ *P* ❒ *PR*	*Date arrival*	*Application no* *DA*	*Familiy No* *FF*
Amount:	*Initial acceptance*		*Initial registration*	*DNA-no* *D*

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

FAMILY TREE

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/

ADDRESS

Sample delivery address (office hours):
AMSTERDAM GENOME DIAGNOSTICS, Department Clinical Genetics
Postoffice H01-114, Meibergdreef 9
1105 AZ Amsterdam
The Netherlands
Outside office hours:
Delivery at LAKC B1-114

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- 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.

EpiSign Diagnostic DNA Methylation Test VARIANT

	Syndrome:	***Gene:***
☐	*ADCADN syndrome*	*DNMT1 (NB: Late-onset disorder)*
☐	*ADNP related syndrome*	*ADNP*
☐	*ATRX related syndrome*	*ATRX*
☐	*BAFopathies*	*ARID1B, SMARCB1, SMARCA4, SMARCA2*
☐	*Cornelia de Lange syndrome*	*NIPBL, RAD21, SMC3, SMC1A*
☐	*CHARGE syndrome*	*CHD7*
☐	*Claes-Jensen syndrome*	*KDM5C*
☐	*Floating Harbor syndrome*	*SRCAP*
☐	*Genitopatellar syndrome*	*KAT6B*
☐	*Kabuki syndrome*	*KMT2D*
☐	*Sotos syndrome*	*NSD1*
☐	*Williams-Beuren syndrome*	
☐	*Williams-Beuren regio duplication syndrome*	
☐	*Fragile X syndrome*	
☐	*Prader Willi syndrome*	
☐	*Angelman syndrome*	
☐	*Silver- Russell syndrome*	
☐	*Beckwith-Wiedemann syndrome*	
☐	*Temple syndrome*	
☐	*Kagami–Ogata syndrome*	
☐	*Mental retardation*	*FRA12A type*

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

At this moment we are the only laboratory in Europe with licence for EpiSign for diagnostics, in collaboration with Londen Health Sciences Centre, Ontario.

EpiSign has multiple applications in the clinical setting by providing an additional diagnostic tool beyond the current sequencing and copy number technology paradigm.

Assessment of the distinct methylation patterns produced by EpiSign will be used as a screening tool for disorders in the diagnostic work-up or will be applied in a more targeted fashion to help resolve VUS (variants of uncertain clinical significance).

Please note that methylation abnormalties detected using this test, may require additional targeted testing to confirm and further characterize the underlying genomic abnormality. This test will not detect females with Fragile X (FMRl) expansions.

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ADDRESS LABEL

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AGDx Laboratory Genome Diagnostics (H01-114)*

Amsterdam UMC

Meibergdreef 9

1105 AZ AMSTERDAM

The Netherlands

DIAGNOSTISCH MATERIAAL

SPOED!

*Outside office hours: delivery at LAKC B1-114

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