

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 www.AGDx.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: AGB Code (for Dutch specialists only):
 Hospital: Phone:
 Medical specialty: E-mail:
 Street/PObox: CC report:
 ZIP code + Town:
 Former family members samples known by AGDx: Yes No
 Name: Date of Birth:
 Relation: Family no. (when known):
 Family consanguinity: Yes (see pedigree – 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; targeted analysis (see list v3 for eligible genes) [EPI] (Test code AUA0003)
 Gene Variant

 Is the variant Mosaic Yes No
 Estimated % of Mosaic: Age patient at extraction date:

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 (2 x 6 ml, do not freeze)
 DNA (minimum 5 µg, isolated from EDTA blood) Extraction date:

TO BE COMPLETED BY AGDx DNA-LABORATORY PERSONNEL

<i>Initial for received material</i>	<i>Date arrival</i>	
<i>Amount:</i>		

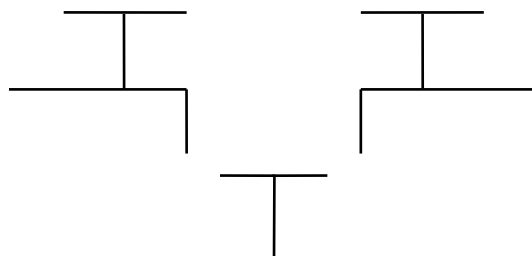
PEDIGREE

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: <https://www.un3373.com/un3373-packaging/un3373/>

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:

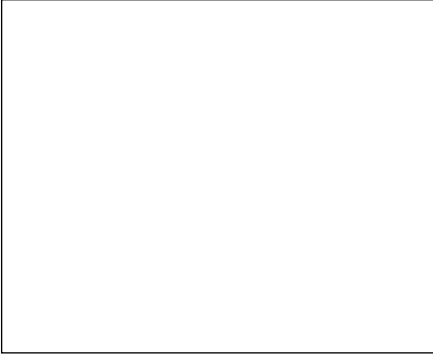
Delivery at LAKC B1-114

See the Disorder and gene list EpiSign v3 for the test content

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

ACCEPTANCE OF FINANCIAL RESPONSIBILITY FOR GENETIC TESTING

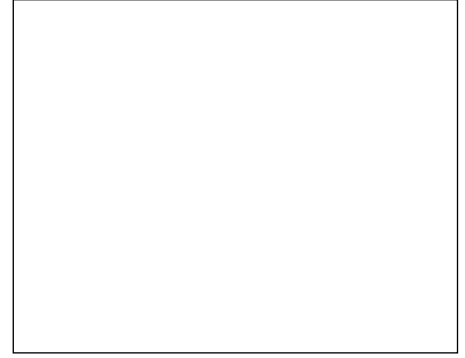
My signature indicates that I accept financial responsibility for all fees associated with this genetic testing order:



Signature of responsible party



Printed name of responsible party



Date

Disclaimer: The listed genes and conditions have been detailed reviewed by Amsterdam UMC Genome Diagnostics, and EpiSign has been validated for clinical use in collaboration with London Health Science Centre, Ontario. Please note that some conditions/genes have been classified as having more moderate signatures based on signature strength, small cohort size, or types of mutations. This test cannot detect females with Fragiel X (FMR1) expansions. Females tested for X-linked conditions may have a moderate signature or a potentially false negative result.

EpiSign produces distinct methylation patterns and will be used as a screenings tool for disorders in the diagnostic work-up or will be applied in a more targeted fashion to help resolve VUS.

As with many clinical tests, uncertain results are possible and additional targeted testing may be required.

ADDRESS LABEL



AGDx Laboratory Genome Diagnostics (H01-114)*

Amsterdam UMC

Meibergdreef 9

1105 AZ AMSTERDAM

The Netherlands



**BIOLOGICAL SUBSTANCE
CATEGORY B**

MEDICAL DIAGNOSTIC SAMPLE

URGENT SHIPPING!

*Outside office hours: delivery at LAKC B1-114

