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 (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

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 (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:

TO BE COMPLETED BY AGDx DNA-LABORATORY PERSONNEL

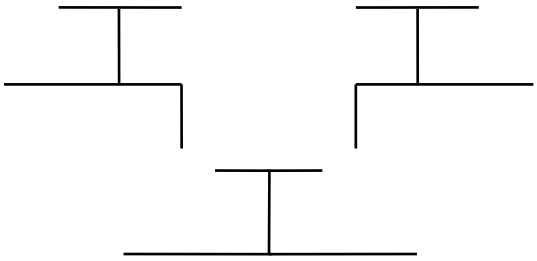
Initial for received material *Amount:*	*Date arrival*	

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: 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

CONDITION **RELATED REGION OR GENE(S)**

Imprinting and Trinucleotide Repeat Disorders	
Disorder	Confirmed underlying gene(s)
Mental retardation, FRA12A type	*DIP2B* promoter
Fragile X syndrome (Affected males ONLY)	*FMR1* promoter
Angelman syndrome	*SNRPN* promoter
Beckwith-Wiedemann syndrome	Both IC1 and IC2 on Chr11
Kagami-Ogata syndrome	*MEG3* promoter
Prader-Willi syndrome	*SNRPN* promoter
Silver-Russell syndrome	UPD11 *(*both IC1 and IC2*),* UPD7
Temple syndrome	*MEG3* promoter
EpiSignature Disorders	
EpiSignature	Confirmed gene(s)
Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant	*DNMT1*
Alpha-thalassemia mental retardation syndrome	*ATRX*
Autism, susceptibility to, 18	*CHD8*
BAFopathies: Coffin-Siris types 1-4,8 & Nicolaides-Baraitser syndromes*	*ARID1A, ARID1B, SMARCB1, SMARCA4, SMARCA2, SMARCC2*
Blepharophimosis Intellectual Disability *SMARCA2* syndrome	*SMARCA2*
Börjeson-Forssman-Lehmann syndrome	*PHF6*
Cornelia de Lange syndrome**	*NIPBL, RAD21, SMC3, SMC1A*
CHARGE syndrome	*CHD7*
Down syndrome	Chr21 trisomy
Chr7q11.23 duplication syndrome	Chr7q11.23 duplication
Epileptic encephalopathy, childhood-onset	*CHD2*
Floating Harbour syndrome	*SRCAP*
Genitopatellar syndrome	*KAT6B*

* *SMARCE1; ARID2; DPF2* may also be detected.
***HDAC8* for males may also be detected.

Hunter McAlpine syndrome	Chr5q35-qter duplication involving *NSD1*
ADNP syndrome, Helsmoortel-van der Aa syndrome (Terminal and Central)	*ADNP*
Immunodeficiency-centromeric instability-facial anomalies syndrome type 1	*DNMT3B*
Immunodeficiency-centromeric instability-facial anomalies syndrome types 2-4	*CDCA7, ZBTB24, HELLS*
Kabuki syndrome	*KMT2D, KDM6A*
Koolen-De Vries syndrome	*KANSL1*
Kleefstra syndrome	*EHMT1*
Mental retardation, autosomal dominant 23	*SETD5*
Mental retardation, autosomal dominant 51	*KMT5B*
Mental retardation, X-linked 93	*BRWD3*
Mental retardation, X-linked 97	*ZNF711*
Claes-Jensen syndrome	*KDM5C*
Nascimento syndrome	*UBE2A*
Snyder-Robinson syndrome	*SMS*
Weaver and Cohen-Gibson syndromes, PRC2 complex disorders	*EZH2, EED*
Rahman syndrome	*HIST1H1E*
Rubinstein-Taybi syndrome	*CREBBP, EP300*
SBBYSS syndrome	*KAT6B*
SETD1B-related syndrome	*SETD1B*
Sotos syndrome	*NSD1*
Tatton-Brown-Rahman syndrome	*DNMT3A*
Wiedemann-Steiner syndrome	*KMT2A*
Wolf-Hirschhorn syndrome	Chr4p16.3 deletion
Williams syndrome	Chr7q11.23 deletion

NOTE: Highlighted conditions represent new EpiSignatures available as part of EpiSign v2. Genes listed in blue are new additions to existing EpiSign v1 EpiSignatures.

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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

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boration 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 unc ertain 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.

ADDRESS LABEL

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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

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