Category	Disorder	Confirmed underlying genes/regions	Notes
Trinucleotide expansion repeat	Fragile X syndrome (Affected males ONLY)	FMR1 promoter	
Imprinting defect	Angelman syndrome	15q11.2-q13 (SNRPN promoter, SNURF)	
Imprinting defect	Beckwith-Wiedemann syndrome	11p15.5 (IC1 and IC2)	
Imprinting defect	Kagami-Ogata syndrome	14q32 (MEG3 promoter)	
Imprinting defect	Mulchandani-Bhoj-Conlin syndrome	20q11-q13 (GNAS)	
Imprinting defect	Multi-locus imprinting disturbances	All EpiSign imprinting regions	
Imprinting defect	Pseudohypoparathyroidism IA & IB	20q11-q13 (GNAS)	
Imprinting defect	Prader-Willi syndrome	15q11.2-q13 (SNRPN promoter, SNURF)	
Imprinting defect	Silver Russel syndrome 1 & 2	11p15.5 (IC1 and IC2), 7q32.2	
Imprinting defect	Temple syndrome	14q32 (MEG3 promoter)	
Imprinting defect	Diabetes mellitus, transient neonatal 1	6q24 (PLAGL1)	
Episignature	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant	DNMT1	Reduced sensitivity may be observed.
Episignature	ARID1A duplication-related syndrome	ARID1A	The range of validated coordinates is 1p36.11(26,964,202-27,099,490). Reduced sensitivity may be observed.
Episignature	Arboleda-Tham syndrome	KAT6A	Reduced sensitivity may be observed.
Episignature	Alpha-thalassemia/Impaired intellectual development syndrome, X-linked	ATRX	Episignature defined with male cases only. Heterozygotes have been shown to not match the episignature.
Episignature	BAFopathies: Coffin-Siris (CSS1-4) & Nicolaides-Baraitser (NCBRS) syndromes	ARID1A, ARID1B, SMARCB1, SMARCA4, SMARCA2	Patients with other BAFopathy genes may be detected, but not confirmed in our experiments.
Episignature	Beck-Fahrner syndrome	TET3	Healthy carriers and those with incomplete penetrance are detectable. Patients with biallelic variants are distinguishable from those with monoallelic variants.
Episignature	Blepharophimosis-impaired intellectual development syndrome	SMARCA2	
Episignature	Cornelia de Lange syndromes 1-4	NIPBL, RAD21, SMC3, SMC1A	Secondary episignatures available to distuinguish between subtypes.
Episignature	CHARGE syndrome	CHD7	
Episignature	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder	CDK13, CCNK	Reduced sensitivity may be observed.
Episignature	Chromosome 19p13.13 deletion syndrome	Chr19p13.13p13.2 deletion	The range of validated coordinates is 19p13.13p13.2(13,201,983-13,213,144). Only for copy number variants. NFIX sequence variants have been shown to not match the episignature.
Episignature	Chromosome 1p36 deletion syndrome	Chr1p36 deletion	The range of validated coordinates is 1p36.33p36.32(1,019,753-2,867,961). Reduced sensitivity may be observed.
Episignature	Chromosome Xp11.22 duplication syndrome	ChrXp11.22 duplication	The range of validated coordinates is Xp11.22(53,559,057-53,654,518). Episignature defined with male cases only. Heterozygotes have been shown to not match the episignature. Reduced sensitivity may be observed.
Episignature	Börjeson-Forssman-Lehmann, Chung-Jansen and White Kernohan syndromes	PHIP, PHF6, DDB1	Secondary episignatures available to distuinguish between subtypes.
Episignature	Clark-Baraitser syndrome	TRIP12	
Episignature	BAFopathies: Coffin-Siris syndrome 1 & 2	ARID1A, ARID1B	Only for variants near c.6200.
Episignature	Coffin-Siris syndrome 4	SMARCA4	Only for variants near c.2656.
Episignature	Coffin-Siris syndrome 6	ARID2	
Episignature	Developmental and epileptic encephalopathy 54	HNRNPU	
Episignature	Developmental and epileptic encephalopathy 94	CHD2	
Episignature	DEGCAGS syndrome	ZNF699	Heterozygotes have been shown to not match the episignature.
Episignature	Developmental delay with variable intellectual disability and dysmorphic facies	JARID2	Reduced sensitivity may be observed.
Episignature	Diets-Jongmans syndrome	KDM3B	
Episignature	Down syndrome	Chr21 trisomy	
Episignature	Williams-Beuren region duplication syndrome	Chr7q11.23 duplication	The range of validated coordinates is 7q11.23(73,953,518-74,138,459).
Episignature	Dystonia 28, childhood-onset	KMT2B	
Episignature	Fanconi anemia	FANCA, FANCC, FANCD2, FANCG, FANCI, FANCL	Heterozygotes have been shown to not match the episignature. Patients with other FANC genes may be detected, but not confirmed in our experiments.
Episignature	Floating Harbour syndrome	SRCAP	
Episignature	Gabriele-de Vries syndrome	YY1	Reduced sensitivity may be observed.
Episignature	Genitopatellar syndrome	KAT6B	Reduced sensitivity may be observed.
Episignature	Hao-Fountain syndrome	USP7	

Episignature	Hunter McAlpine craniosynostosis syndrome	Chr5q35 duplication involving NSD1	The range of validated coordinates is 5q35.2q35.3(175,839,681-176,904,798).
Episignature	Helsmoortel-van der Aa syndrome	ADNP	Central episignature for variants within the coding nucleotide range of c.2054-2340. Terminal episignature for variants outside of the coding nucleotide range of c.2054-2340.
Episignature	Immunodeficiency-centromeric instability-facial anomalies syndrome 1	DNMT3B	Reduced sensitivity may be observed.
Episignature	Immunodeficiency-centromeric instability-facial anomalies syndrome 2-4	CDCA7, ZBTB24, HELLS	Reduced sensitivity may be observed.
Episignature	Intellectual developmental disorder with autism and macrocephaly	CHD8	Reduced sensitivity may be observed.
Episignature	Intellectual developmental disorder with microcephaly and with or without ocular malformations or hypogonadotropic hypogonadism	SOX11	Reduced sensitivity may be observed.
Episignature	Intellectual developmental disorder with seizures and language delay	SETD1B	
Episignature	Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities	BCL11B	Reduced sensitivity may be observed.
Episignature	Kabuki syndrome 1 & 2	KMT2D, KDM6A	Secondary episignatures available to distuinguish between subtypes.
Episignature	Intellectual developmental disorder, autosomal dominant 23; KBGS syndrome	SETD5, ANKRD11	Secondary episignatures available to distuinguish between subtypes.
Episignature	KDM2B-related syndrome	KDM2B	
Episignature	Koolen de Vreis syndrome	KANSL1	
Episignature	Kleefstra syndrome 1	EHMT1	
Episignature	KMT2D-related syndrome	KMT2D	Only for variants within the amino acid range of 3400-3700. Reduced sensitivity may be observed.
Episignature	Luscan-Lumish syndrome	SETD2	
Episignature	Menke-Hennekam syndrome 1 & 2	CREBBP, EP300	Only for domain ID4. Other domains of MKHK1/2 are not available.
Episignature	Mowat-Wilson syndrome	ZEB2	
Episignature	Intellectual developmental disorder, autosomal dominant 21	CTCF	
Episignature	Intellectual developmental disorder, autosomal dominant 51	KMT5B	Healthy carriers and those with incomplete penetrance are detectable. Reduced sensitivity may be observed.
Episignature	Intellectual developmental disorder, autosomal dominant 7	DYRK1A	
Episignature	Intellectual developmental disorder, X-linked, syndromic, Armfield type	FAM50A	Episignature defined with male cases only. Heterozygotes have been shown to not match the episignature. Reduced sensitivity may be observed.
Episignature	Intellectual developmental disorder, X-linked, syndromic, Claes-Jensen type	KDM5C	Healthy carriers and those with incomplete penetrance are detectable. Heterozygotes have a distinct profile from hemizygotes.
Episignature	Intellectual developmental disorder, X-linked syndromic, Nascimento type	UBE2A	Episignature defined with male cases only. Heterozygotes have been shown to not match the episignature. Reduced sensitivity may be observed.
Episignature	Intellectual developmental disorder, X-linked, syndromic, Snyder-Robinson type	SMS	Episignature defined with male cases only. Reduced sensitivity may be observed.
Episignature	MSL2-related syndrome	MSL2	Reduced sensitivity may be observed.
Episignature	Neurodevelopmental disorder with dysmorphic facies and behavioral abnormalities	SRSF1	
Episignature	Neurodevelopmental disorder with hypotonia, stereotypic hand movements, and impaired language	MEF2C	
Episignature	NSD2 duplication-related syndrome	NSD2	The range of validated coordinates is 4p16.3(1,832,733-1,975,031). CNVs overlapping or expanding this region may also be detected.
Episignature	Phelan-McDermid syndrome	Chr22q13.3 deletion	The range of validated coordinates is 22q13.3(49,238,268-50,248,907). CNVs overlapping or expanding this region may also be detected. Only for copy number variants. SHANK3 sequence variants have been shown to not match the episignature.
Episignature	PRC2 complex disorders (Weaver and Cohen-Gibson syndromes)	EZH2, EED	Shared episignatures between PRC2 complex syndromes WVS and COGIS.

Episignature	Neuroocular syndrome	PRR12	Healthy carriers and those with incomplete penetrance are detectable. Reduced sensitivity may be observed.
Episignature	Pitt-Hopkins syndrome	TCF4	
Episignature	Potocki-Lupski syndrome	Chr17p11.2 duplication	The range of validated coordinates is 17p11.2(16,779,412-20,231,379). CNVs overlapping or expanding this region may also be detected. Reduced sensitivity may be observed.
Episignature	Renpenning syndrome	PQBP1	Episignature defined with male cases only. Heterozygotes have been shown to not match the episignature. Reduced sensitivity may be observed.
Episignature	Rahman syndrome	H1-4	
Episignature	Rubinstein-Taybi syndrome 1 and 2	CREBBP, EP300	Secondary episignatures available to distuinguish between subtypes.
Episignature	Ohdo syndrome, SBBYSS variant	KAT6B	Reduced sensitivity may be observed.
Episignature	Sifrim-Hitz-Weiss syndrome	CHD4	
Episignature	SLC32A1-related syndrome	SLC32A1	Reduced sensitivity may be observed.
Episignature	Smith-Magenis syndrome	Chr17p11.2 deletion	The range of validated coordinates is 17p11.2(17,322,913-18,515,769). CNVs overlapping or expanding this region may also be detected. Only for copy number variants. RAI1 sequence variants have been shown to not match the episignature.
Episignature	Sotos syndrome	NSD1	
Episignature	Tatton-Brown-Rahman syndrome	DNMT3A	Reduced sensitivity may be observed.
Episignature	Turner syndrome	ChrX deletion; 45,X	
Episignature	Velocardiofacial syndrome	Chr22q11.2 deletion	The range of validated coordinates is 22q11.21(19,510,547-20,285,090). CNVs overlapping or expanding these regions may be detected.
Episignature	Wiedemann-Steiner syndrome	KMT2A	
Episignature	Wolf-Hirschhorn syndrome & Rauch-Steindel syndrome	Chr4p16.3 deletion, NSD2	The range of validated coordinates is 4p16.3(679,715-2,169,001). CNVs overlapping or expanding this region may also be detected. NSD2 sequence variants have been shown to match the episignature.
Episignature	White-Sutton syndrome	POGZ	
Episignature	Williams-Beuren syndrome	Chr7q11.23 deletion	CNVs overlapping or expanding 7q11.23 may also be detected.
Episignature	Witteveen-Kolk syndrome	SIN3A	Reduced sensitivity may be observed.
Episignature	Wieacker-Wolff syndrome	ZC4H2	Episignature defined with male cases only. Heterozygotes have been shown to not match the episignature. Reduced sensitivity may be observed.
Episignature	Intellectual developmental disorder, X-linked 93	BRWD3	Healthy carriers and those with incomplete penetrance are detectable. Reduced sensitivity may be observed.
Episignature	Intellectual developmental disorder, X-linked 97	ZNF711	Heterozygotes have been shown to match the episignature. Reduced sensitivity may be observed.
Episignature	Klinefelter syndrome	ChrX duplication; 47,XXY	XXX cases may also be detected.
Episignature - targeted only	Fetal Valproate syndrome	NA	Available as a targeted request only.