EpiSign V3 List of disorders and genes.

Unique Multi-locus Epigenetic Signatures

Imprinting Abnormalities	Related Region or Gene(s)
Angelman syndrome	15q11.2 methylation abnormality
Beckwith-Wiedemann syndrome	11p15 imprinting abnormality
Diabetes Mellitus, transient neonatal 1	*PLAGL1* imprinting abnormality
Fragile X syndrome	*FMR1* methylation abnormality
Kagami-Ogata syndrome	*MEG3* imprinting abnormality
Mulchandani-Bhoj-Conlin syndrome	20q11-q13 imprinting abnormality involving *GNAS* complex locus
Prader-Willi syndrome	15q11.2 methylation abnormality
Pseudohypoparathyroidism, Type 1A-1C	20q13.32 imprinting abnormality involving *GNAS* complex locus
Russell-Silver syndrome 1	11p15 imprinting abnormality
Russell-Silver syndrome 2	UPD 7 imprinting abnormality
Temple syndrome	*MEG3* imprinting abnormality

Conditions with Strong Signatures	Related Region or Gene(s)
7q11.23 duplication	7q11.23 duplication (Chr7:72,745,0470-74,138,460)
AD Cerebellar Ataxia. Deafness, Narcolepsy (ADCADN)	*DNMT1*
Alpha-thalassemia X-linked Intellectual Disability	*ATRX*
BAFopathy 1: including Coffin-Siris syndrome & Nicolaides-Baraitser [1]	*ARID1A, ARID1B, SMARCB1, SMARCA4, SMARCA2*
BAFopathy 2: Coffin-Siris syndrome 1 & 2	*ARID1A* (c.6133-c.6254), *ARID1B* (c.6133-c.6254)
Beck-Fahrner syndrome [3,4]	*TET3*
Blepharophimosis Intellectual Disability *SMARCA2* syndrome	*SMARCA2*
CHARGE syndrome	*CHD7*
Coffin-Siris syndrome-4	*SMARCA4* (c.2656)
Cornelia de Lange syndrome [2]	*NIPBL, RAD21, SMC3, SMC1A*
Down syndrome	Trisomy 21
Dystonia-28	*KMT2B*
Epileptic Encephalopathy, childhood-onset	*CHD2*
Floating-Harbor syndrome	*SRCAP*
Gabriele-de Vries syndrome	*YY1*
Helsmoortel-Van der Aa syndrome	*ADNP*
Hunter-McAlpine syndrome	5q35.2q35.3 duplication (Chr5:175,728,979-177,047,793) involving *NSD1*
Intellectual development disorder with seizures and language delay	*SETD1B*
Intellectual Disability, X-linked syndromic, Claes-Jensen type [3]	*KDM5C*
Intellectual Disability, X-linked syndromic, Nascimento-type	*UBE2A*
Kabuki syndrome 1 & 2	*KDM6A, KMT2D*
KDM2B-related syndrome	*KDM2B*
Kleefstra syndrome	*EHMT1*
Koolen-De Vries syndrome	*KANSL1*
Luscan-Lumish syndrome	*SETD2*
Menke-Hennekam syndrome 1 & 2 (IDR4 domain only)	*CREBBP* (c.5563-5614), *EP300* (c.5471-5495)
Myopathy, lactic acidosis, and sideroblastic anemia-2	*YARS2*
Phelan-McDermid syndrome	22q13.3 deletion (Chr22:45,277,036-51,244,566)
PRC2 complex disorders: Weaver & Cohen-Gibson syndromes	*EZH2, EED*
Rahman syndrome	*HIST1H1E*
Rubinstein-Taybi syndrome 1	*CREBBP*
Rubinstein-Taybi syndrome 2	*EP300*
Sotos syndrome 1	*NSD1*
Velocardiofacial syndrome	22q11.2 deletion (Chr22: 16,888,899-21,915,509)
Wiedemann-Steiner syndrome	*KMT2A*
Williams-Beuren syndrome	7q11.23 deletion (Chr7:72,744,455-74,142,510)
Wolf-Hirschhorn syndrome	4p16.3 deletion (Chr4:331,568-2,010,962) involving *NSD2*

Genes/conditions listed in blue are new signatures for EpiSign version 3.

Conditions with Moderate Signatures	Related Region or Gene(s)
Arboleda-Tham syndrome	*KAT6A*
Austism, susceptibility to, 18	*CHD8*
Borjeson-Forssman-Lehmann syndrome	*PHF6*
Coffin-Siris syndrome 9	*SOX11*
Genitopatellar syndrome	*KAT6B*
Immunodeficiency-centromeric instability-facial anomalies syndrome, type 1	*DNMT3B*
Immunodeficiency-centromeric instability-facial anomalies syndrome, types 2-4	*CDCA7, ZBTB24, HELLS*
Intellectual Developmental Disorder, autosomal dominant 65	*KDM4B*
Intellectual Developmental Disorder, X-linked, syndromic, Armfield type	*FAM50A*
Intellectual Disability, autosomal dominant 23	*SETD5*
Intellectual Disability, autosomal dominant 51 [3]	*KMT5B*
Intellectual Disability, X-linked 93 [3]	*BRWD3*
Intellectual Disability, X-linked 97	*ZNF711*
Renpenning syndrome	*PQBP1*
SBBYS syndrome	*KAT6B*
Snyder-Robinson syndrome	*SMS*
Tatton-Brown-Rahman syndrome	*DNMT3A*

[1] Patients with variants in other BAFopathy genes may be detected, but this finding has not been confirmed.
[2] *HDAC8* for males may also be detected, but this finding has not been confirmed.
[3] Healthy carriers and those with incomplete penetrance are detectable.
[4] Patients with biallelic variants are distinguishable from those with monoallelic variants.

Note: All coordinates are based on human genome build hg19.

Disclaimer: *Some conditions/genes have been classified as having more moderate signatures based on signature strength, small cohort size, or types of mutations.* Females tested for X-linked conditions may have a moderate signature or a potentially false negative result. As with many clinical tests, uncertain results are possible. Please note that a normal result does not rule out the possibility that the patient is affected with one of these conditions. In some case, specific follow-up testing may be suggested to confirm or rule out a diagnosis.