EPISIGN™ TESTING MENU

Version 6

EpiSign™ METRIC is a research use only (RUO) software not intended for standalone use in diagnostic procedures.

Locus	Associated OMIM Condition (#)	Methylation Category
45,X	Turner syndrome	Locus
47,XXX	Trisomy X	Family
47,XXX	Trisomy X	Locus
47,XXY	Klinefelter syndrome	Family
47,XXY	Klinefelter syndrome	Locus
47,XYY	XYY syndrome/Jacobs syndrome	Family
47,XYY	XYY syndrome/Jacobs syndrome	Locus
ADNP	Helsmoortel-van der Aa syndrome (615873)	Family
ADNP	Helsmoortel-van der Aa syndrome (615873)	Variant Consequence
ADNP	Helsmoortel-van der Aa syndrome (615873)	Region
ADNP	Helsmoortel-van der Aa syndrome (615873)	Region
AHCY	Hypermethioninemia with S-adenosylhomocysteine hydrolase deficiency (613752)	Locus
AHDC1	Xia-Gibbs syndrome (615829)	Locus
ANKRD11	KBG syndrome (148050)	Family, Secondary
ARID1A	Coffin-Siris syndrome 2 (614607)	Family
ARID1A	Coffin-Siris syndrome 2 (614607)	Region
ARID1A	Coffin-Siris syndrome 2 (614607)	Locus
ARID1A	ARID1A duplication-related syndrome (603024)	Variant Class
ARID1B	Coffin-Siris syndrome 1 (135900)	Family
ARID1B	Coffin-Siris syndrome 1 (135900)	Region
ARID1B	Coffin-Siris syndrome 1 (135900)	Locus
ARID2	Coffin-Siris syndrome 6 (617808)	Locus
ATRX	Alpha-thalassemia/impaired intellectual development syndrome, X-linked (301040)	Locus
ATRX	Intellectual disability-hypotonic facies syndrome, X-linked (309580)	Locus
BCL11B	Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities (618092)	Locus
BCLAF3	BCLAF3-related syndrome (301159)	Promoter
BICRA	Coffin-Siris syndrome 12 (619325)	Locus

BPTF	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies (617755)	Family
BRWD3	Intellectual developmental disorder, X-linked 93 (300659)	Locus
BRWD3	Intellectual developmental disorder, X-linked 93 (300659)	Variant Zygosity
CCNK	Intellectual developmental disorder with hypertelorism and distinctive facies (618147)	Family
CDCA7	Immunodeficiency-centromeric instability-facial anomalies syndrome 3 (616910)	Family
CDK13	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder (617360)	Family
CDK13	CDK13-related syndrome (603309)	Variant Class
CHAMP1	Neurodevelopmental disorder with hypotonia, impaired language, and dysmorphic features (616579)	Family
CHD2	Developmental and epileptic encephalopathy 94 (615369)	Locus
CHD4	Sifrim-Hitz-Weiss syndrome (617159)	Family
CHD4	Sifrim-Hitz-Weiss syndrome (617159)	Variant Class
CHD4	CHD4-related syndrome (603277)	Variant Class
CHD6	CHD6-related syndrome (616114)	Locus
CHD6	CHD6-related syndrome (616114)	Variant Consequence
CHD7	CHARGE syndrome (214800)	Locus
CHD8	Intellectual developmental disorder with autism and macrocephaly (615032)	Locus
Chr1p36.11del	Xia-Gibbs syndrome (615829)	Locus
Chr1p36.11dup	ARID1A duplication-related syndrome (603024)	Variant Class
Chr1p36del_distal	Chromosome 1p36 deletion syndrome, distal (607872)	Locus
Chr1q43q44del	Developmental and epileptic encephalopathy 54 (617391)	Locus
Chr2p13.2del	Beck-Fahrner syndrome (618798)	Locus
Chr2q22q22.3del	Mowat-Wilson syndrome (235730)	Locus
Chr2q36.1q36.2del	Neurodevelopmental disorder with or without autism or seizures (619239)	Locus
Chr2q36.3del	Clark-Baraitser syndrome (617752)	Locus
Chr3p21.31del	Luscan-Lumish syndrome (616831)	Locus
Chr3p25.3del	Intellectual developmental disorder, autosomal dominant 23 (615761)	Family, Secondary
Chr3p25.3del	Intellectual developmental disorder, autosomal dominant 23 (615761)	Family, Secondary

Chr4p16.3del	Wolf-Hirschhorn syndrome (194190)	Family
Chr4p16.3del	Wolf-Hirschhorn syndrome (194190)	Locus
Chr4p16.3dup	NSD2 duplication-related syndrome (602952)	Variant Class
Chr5p13.2del	Cornelia de Lange syndrome 1 (122470)	Family
Chr5p13.2del	Cornelia de Lange syndrome 1 (122470)	Locus, Secondary
Chr5q31.2del	Diets-Jongmans syndrome (618846)	Locus
Chr5q35.2q35.3del	Sotos syndrome (117550)	Locus
Chr5q35.2q35.3dup	Hunter-McAlpine craniosynostosis syndrome (601379)	Locus
Chr6p24.2p22.3del	Developmental delay with variable intellectual disability and dysmorphic facies (601594)	Locus
Chr6q24dup(pat)	Diabetes mellitus, transient neonatal 1 (601410)	Imprinting
Chr7q11.23del	Williams-Beuren syndrome (194050)	Locus
Chr7q11.23dup	Williams-Beuren region duplication syndrome (609757)	Locus
Chr7q21del(pat)	Silver-Russell syndrome 2 (618905)	Imprinting
Chr7q36.1del	Weaver syndrome (277590)	Family
Chr7q36.1del	Weaver syndrome (277590)	Locus, Secondary
Chr8 inv dup del	Chromosome 8p invdupdel syndrome	Locus
Chr9q34.3del	Kleefstra syndrome 1 (610253)	Locus
Chr9q34.3dup	EHMT1 duplication-related syndrome (607001)	Variant Class
Chr10p12.1del	DeSanto-Shinawi syndrome (616708)	Family
Chr10p12.1del	DeSanto-Shinawi syndrome (616708)	Locus
Chr10q22.2del	Ohdo syndrome, SBBYSS variant (603736)	Family, Secondary
Chr10q22.3q23.2del	WAPL-related syndrome (610754)	Family
Chr10q22.3q23.2del	WAPL-related syndrome (610754)	Locus
Chr11p15.5dup(mat)	Silver-Russell syndrome 1 (180860)	Imprinting
Chr11p15.5dup(pat)	Beckwith-Wiedemann syndrome (130650)	Imprinting
Chr11q14.1q14.3del	Cohen-Gibson syndrome (617561)	Family
Chr11q14.1q14.3del	Cohen-Gibson syndrome (617561)	Locus, Secondary
Chr11q23.3del	Wiedemann-Steiner syndrome (605130)	Locus

Chr12q12q13.11del	Coffin-Siris syndrome 6 (617808)	Locus
Chr12q24.31del	Neurodevelopmental disorder with congenital cardiac defects and variable renal and ocular abnormalities (621474)	Locus
Chr12q24.31del	Intellectual developmental disorder with seizures and language delay (619000)	Locus
Chr14q11.2del	Intellectual developmental disorder with autism and macrocephaly (615032)	Locus
Chr14q32del(mat)	Kagami-Ogata syndrome (608149)	Imprinting
Chr14q32del(pat)	Temple syndrome (616222)	Imprinting
Chr14q32.2del	Gabriele-de Vries syndrome (617557)	Locus
Chr15q11.2q13del(mat)	Angelman syndrome (105830)	Imprinting
Chr15q11.2q13del(pat)	Prader-Willi syndrome (176270)	Imprinting
Chr15q21.2q21.3del	Craniosynostosis 3 (615314)	Family
Chr15q24.2q25.1del	Witteveen-Kolk syndrome (613406)	Locus
Chr16p13.2del	Hao-Fountain syndrome (616863)	Locus
Chr16p13.3del	Rubinstein-Taybi syndrome 1 (180849)	Family
Chr16p13.3del	Rubinstein-Taybi syndrome 1 (180849)	Locus, Secondary
Chr16q24.3del	KBG syndrome (148050)	Family, Secondary
Chr17p11.2del	Smith-Magenis syndrome (182290)	Locus
Chr17p11.2dup	Potocki-Lupski syndrome (610883)	Locus
Chr17q11.2del	Imagawa-Matsumoto syndrome (618786)	Family, Secondary
Chr17q11.2del	PHF12-related syndrome (618645)	Locus
Chr17q21.31del	Koolen-De Vries syndrome (610443)	Locus
Chr17q22del	Neurodevelopmental disorder with dysmorphic facies and behavioral abnormalities (620489)	Locus
Chr18q12.3q21.1del	Intellectual developmental disorder, autosomal dominant 29 (616078)	Variant Class
Chr18q21.2q21.32del	Pitt-Hopkins syndrome (610954)	Family
Chr19p13.13del	Chromosome 19p13.13 deletion syndrome (613638)	Locus
Chr19q13.33del	Coffin-Siris syndrome 12 (619325)	Locus
Chr20p13del	Okur-Chung neurodevelopmental syndrome (617062)	Family
Chr20p13del	Okur-Chung neurodevelopmental syndrome (617062)	Locus
Chr20q13del(mat)	Pseudohypoparathyroidism IB (603233)	Imprinting

Chr20q13del(pat)	Mulchandani-Bhoj-Conlin syndrome (617352)	Imprinting
Chr21 trisomy	Down syndrome	Locus
Chr22q11.2del	Velocardiofacial syndrome (192430)	Locus
Chr22q13.2dup	Rubinstein-Taybi syndrome 2 (613684)	Family
Chr22q13.2dup	Rubinstein-Taybi syndrome 2 (613684)	Locus, Secondary
Chr22q13.3del	Phelan-McDermid syndrome (606232)	Locus
ChrXp11.22dup	Chromosome Xp11.22 duplication syndrome (300705)	Locus
ChrXp11.3dup	Kabuki syndrome 2 (300867)	Family
ChrXp11.3dup	Kabuki syndrome 2 (300867)	Locus
ChrXq28dup	Intellectual developmental disorder, X-linked syndromic, Lubs type (300260)	Variant Class
CREBBP	Rubinstein-Taybi syndrome 1 (180849)	Family
CREBBP	Rubinstein-Taybi syndrome 1 (180849)	Locus, Secondary
CREBBP	Menke-Hennekam syndrome 1 (618332)	Region
CSNK2A1	Okur-Chung neurodevelopmental syndrome (617062)	Family
CSNK2A1	Okur-Chung neurodevelopmental syndrome (617062)	Locus
CSNK2B	Poirier-Bienvenu neurodevelopmental syndrome (618732)	Family
CTCF	Intellectual developmental disorder, autosomal dominant 21 (615502)	Locus
CUL3	Neurodevelopmental disorder with or without autism or seizures (619239)	Locus
DDB1	White-Kernohan syndrome (619426)	Family
DDB1	White-Kernohan syndrome (619426)	Locus, Secondary
DMAP1	DMAP1-related syndrome (605077)	Locus
DNMT1	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant (604121)	Locus
DNMT3A	Tatton-Brown-Rahman syndrome (615879)	Locus
DNMT3B	Immunodeficiency-centromeric instability-facial anomalies syndrome 1 (242860)	Locus
DOHH	Neurodevelopmental disorder with microcephaly, cerebral atrophy, and visual impairment (620066)	Locus
DPF2	Coffin-Siris syndrome 7 (618027)	Family
DPF2	Coffin-Siris syndrome 7 (618027)	Locus, Secondary
DYRK1A	Intellectual developmental disorder,	Locus

	autosomal dominant 7 (614104)	
EED	Cohen-Gibson syndrome (617561)	Family
EED	Cohen-Gibson syndrome (617561)	Locus, Secondary
EHMT1	Kleefstra syndrome 1 (610253)	Locus
EHMT2	EHMT2-related syndrome (604599)	Locus
EP300	Rubinstein-Taybi syndrome 2 (613684)	Family
EP300	Rubinstein-Taybi syndrome 2 (613684)	Locus, Secondary
EP300	Menke-Hennekam syndrome 2 (618333)	Region
EZH2	Weaver syndrome (277590)	Family
EZH2	Weaver syndrome (277590)	Locus, Secondary
FAM50A	Intellectual developmental disorder, X-linked, syndromic, Armfield type (300261)	Locus
FANCA	Fanconi anemia, complementation group A (227650)	Family
FANCC	Fanconi anemia, complementation group C (227645)	Family
FANCD2	Fanconi anemia, complementation group D2 (227646)	Family
FANCG	Fanconi anemia, complementation group G (614082)	Family
FANCI	Fanconi anemia, complementation group I (609053)	Family
FANCL	Fanconi anemia, complementation group L (614083)	Family
FBXO11	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities (618089)	Locus
FMR1	Fragile X syndrome (300624)	Promoter
H1-4	Rahman syndrome (617537)	Locus
H4C3	Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 1 (619758)	Family
H4C4	H4C4-related syndrome (602823)	Family
H4C5	Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 3 (619950)	Family
H4C6	H4C6-related syndrome (602824)	Family
H4C9	Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 4 (619951)	Family
H4C11	Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 2 (619759)	Family
HELLS	Immunodeficiency-centromeric instability-facial anomalies syndrome 4 (616911)	Family

HNRNPU	Developmental and epileptic encephalopathy 54 (617391)	Locus
ID(6)mat-loss	Diabetes mellitus, transient neonatal 1 (601410)	Imprinting
ID(11)mat-loss	Beckwith-Wiedemann syndrome (130650)	Imprinting
ID(11)pat-loss	Silver-Russell syndrome 1 (180860)	Imprinting
ID(14)mat-loss	Kagami-Ogata syndrome (608149)	Imprinting
ID(14)pat-loss	Temple syndrome (616222)	Imprinting
ID(15)mat-loss	Angelman syndrome (105830)	Imprinting
ID(15)pat-loss	Prader-Willi syndrome (176270)	Imprinting
ID(20)mat-loss	Pseudohypoparathyroidism IB (603233)	Imprinting
ID(20)pat-loss	Mulchandani-Bhoj-Conlin syndrome (617352)	Imprinting
JARID2	Developmental delay with variable intellectual disability and dysmorphic facies (601594)	Locus
KANSL1	Koolen-De Vries syndrome (610443)	Locus
KAT6A	Arboleda-Tham syndrome (616268)	Family, Secondary
KAT6B	Genitopatellar syndrome (606170)	Family, Secondary
KAT6B	Ohdo syndrome, SBBYSS variant (603736)	Family, Secondary
KDM2B	Neurodevelopmental disorder with congenital cardiac defects and variable renal and ocular abnormalities (621474)	Locus
KDM3B	Diets-Jongmans syndrome (618846)	Locus
KDM5B	Intellectual developmental disorder, autosomal recessive 65 (618109)	Locus
KDM5B	KDM5B-related neurodevelopmental disorder (605393)	Locus
KDM5C	Intellectual developmental disorder, X-linked, syndromic, Claes-Jensen type (300534)	Locus
KDM6A	Kabuki syndrome 2 (300867)	Family
KDM6A	Kabuki syndrome 2 (300867)	Locus
KMT2A	Wiedemann-Steiner syndrome (605130)	Locus
KMT2B	Dystonia 28, childhood-onset (617284)	Locus
KMT2B	Intellectual developmental disorder, autosomal dominant 68 (619934)	Locus
KMT2C	KMT2C-related syndrome (606833)	Locus
KMT2D	Kabuki syndrome 1 (147920)	Family
KMT2D	Kabuki syndrome 1 (147920)	Locus, Secondary

KMT2D	Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome (620186)	Region
KMT5B	Intellectual developmental disorder, autosomal dominant 51 (617788)	Locus
MACROH2A1	MACROH2A1-related syndrome (610054)	Locus
MACROH2A1	MACROH2A1-related syndrome (610054)	Variant Class
MAU2	Cornelia de Lange syndrome 7 (621570)	Family
MAU2	Cornelia de Lange syndrome 7 (621570)	Locus
MECP2	Intellectual developmental disorder, X-linked syndromic, Lubs type (300260)	Variant Class
MEF2C	Neurodevelopmental disorder with hypotonia, stereotypic hand movements, and impaired language (613443)	Locus
MORC2	MORC2-related syndrome (616661)	Locus
MSL2	Karayol-Borroto-Haghshenas neurodevelopmental syndrome (620985)	Locus
MTOR	Smith-Kingsmore syndrome (616638)	Locus
Multiple imprinted loci	Multi-locus imprinting disturbances	Imprinting
Multiple imprinted loci	Genome-wide uniparental disomy	Imprinting
NIPBL	Cornelia de Lange syndrome 1 (122470)	Family
NIPBL	Cornelia de Lange syndrome 1 (122470)	Locus, Secondary
NOTCH1	NOTCH1-related syndrome (190198)	Locus
NSD1	Sotos syndrome (117550)	Locus
NSD2	Rauch-Steindl syndrome (619695)	Family
NSD2	Rauch-Steindl syndrome (619695)	Locus
Other	Craniofacial microsomia 1 (164210)	Phenotype
Other	Valproate embryopathy, susceptibility to (609442)	Environmental
Other	VATER/VACTERL association (192350)	Phenotype
Other	Recurrent constellations of embryonic malformations	Phenotype
PACS1	Schuurs-Hoeijmakers syndrome (615009)	Locus
PDS5B	PDS5B-related syndrome (605333)	Family
PHF6	Börjeson-Forssman-Lehmann syndrome (301900)	Family, Secondary
PHF8	Intellectual developmental disorder, X-linked syndromic, Siderius type (300263)	Locus
PHF12	PHF12-related syndrome (618645)	Locus

PHIP	Chung-Jansen syndrome (617991)	Family, Secondary
POGZ	White-Sutton syndrome (619426)	Family
PQBP1	Renpenning syndrome (309500)	Locus
PRR12	Neuroocular syndrome 1 (619539)	Locus
PTBP1	STAD syndrome (621495)	Locus
RAD21	Cornelia de Lange syndrome 4 (614701)	Family
RAD21	Cornelia de Lange syndrome 4 (614701)	Locus, Secondary
RNU4-2	ReNU syndrome (620851)	Family
RNU4-2	ReNU syndrome (620851)	Locus
RPL5	Diamond-Blackfan anemia 6 (612561)	Family
RPL11	Diamond-Blackfan anemia 7 (612562)	Family
RPL35A	Diamond-Blackfan anemia 5 (612528)	Family
RPS17	Diamond-Blackfan anemia 4 (612527)	Family
RPS19	Diamond-Blackfan anemia 1 (105650)	Family
RPS19	Diamond-Blackfan anemia 1 (105650)	Locus
RPS26	Diamond-Blackfan anemia 10 (613309)	Family
SET	Intellectual developmental disorder, autosomal dominant 58 (618106)	Locus
SETBP1	Schinzel-Giedion midface retraction syndrome (269150)	Region
SETBP1	Intellectual developmental disorder, autosomal dominant 29 (616078)	Variant Class
SETD1A	Neurodevelopmental disorder with speech impairment and dysmorphic facies (619056); Epilepsy, early-onset, 2, with or without developmental delay (618832)	Locus
SETD1B	Intellectual developmental disorder with seizures and language delay (619000)	Locus
SETD2	Luscan-Lumish syndrome (616831)	Locus
SETD5	Intellectual developmental disorder, autosomal dominant 23 (615761)	Family, Secondary
SIN3A	Witteveen-Kolk syndrome (613406)	Locus
SMAD4	Myhre syndrome (139210)	Locus
SMARCA2	Nicolaides-Baraitser syndrome (601358)	Family
SMARCA2	Nicolaides-Baraitser syndrome (601358)	Region, Secondary

SMARCA2	Blepharophimosis-impaired intellectual disability syndrome (619293)	Region
SMARCA4	Coffin-Siris syndrome 4 (614609)	Family
SMARCA4	Coffin-Siris syndrome 4 (614609)	Locus, Secondary
SMARCA4	Coffin-Siris syndrome 4 (614609)	Variant Consequence
SMARCA5	SMARCA5-related syndrome (603375)	Family
SMARCA5	SMARCA5-related syndrome (603375)	Locus
SMARCB1	Coffin-Siris syndrome 3 (614608)	Family
SMARCB1	Coffin-Siris syndrome 3 (614608)	Region
SMARCB1	Coffin-Siris syndrome 3 (614608)	Variant Consequence
SMARCB1	Coffin-Siris syndrome 3 (614608)	Locus, Secondary
SMARCE1	Coffin-Siris syndrome 5 (616938)	Family
SMARCE1	Coffin-Siris syndrome 5 (616938)	Locus, Secondary
SMC1A	Cornelia de Lange syndrome 2 (300590)	Family
SMC1A	Cornelia de Lange syndrome 2 (300590)	Locus, Secondary
SMC3	Cornelia de Lange syndrome 3 (610759)	Family
SMC3	Cornelia de Lange syndrome 3 (610759)	Locus, Secondary
SMCHD1	Facioscapulohumeral muscular dystrophy 2 (158901)	Locus
SMS	Intellectual developmental disorder, X-linked, syndromic, Snyder-Robinson type (309583)	Locus
SOX11	Intellectual developmental disorder with microcephaly and with or without ocular malformations or hypogonadotropic hypogonadism (615866)	Locus
SRCAP	Floating-Harbor syndrome (136140)	Family
SRCAP	Floating-Harbor syndrome (136140)	Region
SRCAP	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities (619595)	Family
SRCAP	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities (619595)	Region
SRSF1	Neurodevelopmental disorder with dysmorphic facies and behavioral abnormalities (620489)	Locus
STAT1	Immunodeficiency 31C, chronic mucocutaneous candidiasis, autosomal dominant (614162)	Locus
SUZ12	Imagawa-Matsumoto syndrome (618786)	Family, Secondary
TCF4	Pitt-Hopkins syndrome (610954)	Family

TCF12	Craniosynostosis 3 (615314)	Family
TET3	Beck-Fahrner syndrome (618798)	Locus
TET3	Beck-Fahrner syndrome (618798)	Variant Zygosity
TLK2	Intellectual developmental disorder, autosomal dominant 57 (618050)	Locus
TRIP12	Clark-Baraitser syndrome (617752)	Locus
TRRAP	Developmental delay with or without dysmorphic facies and autism (618454)	Locus
U2AF2	Developmental delay, dysmorphic facies, and brain anomalies (620535)	Family
U2AF2	Developmental delay, dysmorphic facies, and brain anomalies (620535)	Locus
UBE2A	Intellectual developmental disorder, X-linked, syndromic, Nascimento type (300860)	Family
UBE2A	Intellectual developmental disorder, X-linked, syndromic, Nascimento type (300860)	Locus
UPD(6)pat	Diabetes mellitus, transient neonatal 1 (601410)	Imprinting
UPD(7)mat	Silver-Russell syndrome 2 (618905)	Imprinting
UPD(7)pat	Paternal UPD7	Imprinting
UPD(11)mat	Silver-Russell syndrome 1 (180860)	Imprinting
UPD(11)pat	Beckwith-Wiedemann syndrome (130650)	Imprinting
UPD(14)mat	Temple syndrome (616222)	Imprinting
UPD(14)pat	Kagami-Ogata syndrome (608149)	Imprinting
UPD(15)mat	Prader-Willi syndrome (176270)	Imprinting
UPD(15)pat	Angelman syndrome (105830)	Imprinting
UPD(16)mat	Maternal UPD16	Imprinting
UPD(16)pat	Paternal UPD16	Imprinting
UPD(20)mat	Mulchandani-Bhoj-Conlin syndrome (617352)	Imprinting
UPD(20)pat	Pseudohypoparathyroidism IB (603233)	Imprinting
USP7	Hao-Fountain syndrome (616863)	Locus
WAC	DeSanto-Shinawi syndrome (616708)	Family
WAC	DeSanto-Shinawi syndrome (616708)	Locus
WAPL	WAPL-related syndrome (610754)	Family
WAPL	WAPL-related syndrome (610754)	Locus

YY1	Gabriele-de Vries syndrome (617557)	Locus
ZBTB24	Immunodeficiency-centromeric instability-facial anomalies syndrome 2 (614069)	Family
ZC4H2	Wieacker-Wolff syndrome (314580)	Locus
ZEB2	Mowat-Wilson syndrome (235730)	Locus
ZMYM2	Neurodevelopmental-craniofacial syndrome with variable renal and cardiac abnormalities (619522)	Locus
ZMYM3	Intellectual developmental disorder, X-linked 112 (301111)	Locus
ZNF699	DEGCAGS syndrome (619488)	Locus
ZNF711	Intellectual developmental disorder, X-linked 97 (300803)	Locus

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Locus: Episignature aligned to a single locus or a gene's canonical profile.
Family: Episignature reflecting a shared signal across a gene family or pathway.
Region: Episignature associated with sub-gene regions, such as domains or hotspot segments.
Variant class: Episignature corresponding to a variant category, such as truncating or missense.
Variant consequence: Episignature reflecting a targeted functional consequence at a specific genomic position.
Variant zygosity: Episignature distinguishing zygosity-related methylation differences.
Environmental: Episignature influenced by non-genetic exposures, including prenatal factors.
Phenotype: Episignature associated with a clinical phenotype without a known gene-level explanation.
Secondary: Episignature used to help distinguish gene-specific cases within the broader signature family.