Disorder	Causative Gene or Region (OMIM#)
Imprinting and Trinucleotide Repeat Disorders	
Angelman syndrome (AS)	15q11.2-q13 (*SNRPN* promoter, SNURF) (105830)
Beckwith-Wiedemann syndrome (BWS)	11p15 (IC1 and IC2) (130650)
Diabetes mellitus, transient neonatal 1 (TNDM1)	6q24 (*PLAG1*) (601410)
Fragile X syndrome (FXS)	*FMR1* promoter (300624)
Kagami-Ogata syndrome (KOS)	14q32 (*MEG3* promoter) (608149)
Mulchandani-Bhoj-Conlin syndrome (MBCS)	20q11-q13 (*GNAS*) (617352)
Multi-locus imprinting disturbances (MLID)	All EpiSign™ imprinting regions.
Prader-Willi syndrome (PWS)	15q11.2 (*SNRPN* promoter, SNURF) (176270)
Pseudohypoparathyroidism, Type Ia, Ib (PHP1A, PHP1B)	20q13.32 (*GNAS*) (139320, 603233)
Silver-Russel syndrome 1 (SRS1)	11p15 (IC1 and IC2) (180860)
Silver-Russel syndrome 2 (SRS2)	7p13-q32 (618905)
Temple syndrome	14q32 (*MEG3* promoter) (616222)
Episignature Disorders	
Alpha-thalassemia/Impaired intellectual development syndrome, X-linked (ATRX) and Intellectual disability-hypotonic facies syndrome, X-linked, 1 (MRXHF1)[1]*	*ATRX* (301040, 309580)
Ankylosing spondylitis[46]*	*IL12B* (161561)
Arboleda-Tham syndrome (ARTHS)[2]	*KAT6A* (616268)
ARID1A duplication-related syndrome[2,3]*	*ARID1A* (603024)
Au-Kline syndrome (AUKS)[46]*	*HNRNPK* (616580)
BAFopathies: Coffin-Siris (CSS1-5, 7) & Nicolaides-Baraitser (NCBRS) syndromes[4]*	*ARID1B, ARID1A, SMARCB1, SMARCA4, SMARCE1, DPF2 SMARCA2* (135900, 614607, 614608, 614609, 616938, 618027, 601358)
BAFopathies: Coffin-Siris syndrome 1 & 2 (CSS1, CSS2)[5]	*ARID1B, ARID1A* (135900, 614607)
Beck-Fahrner syndrome (BEFAHRS)[6,7]	*TET3* (618798)
Beck-Fahrner syndrome (BEFAHRS_biallelic)	*TET3* (618798)
Blepharophimosis-impaired intellectual development syndrome (BIS)	*SMARCA2* (619293)
Börjeson-Forssman-Lehmann syndrome (BFLS)[1,2,8]	*PHF6* (301900)
Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome (BCAHH) [2,9]	*KMT2D* (620186)
Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant (ADCADN)[2]	*DNMT1* (604121)
CHARGE syndrome	*CHD7* (214800)
CHD6-related syndrome*	*CHD6* (616114)
CHD6-related syndrome (CHD6_p986_987)[55]*	*CHD6* (616114)
Chr1p36 deletion syndrome [2,10]	Chr1p36 deletion (607872)
Chromosome 19p13.13 deletion syndrome[11]	Chr19p13.13del (613638)
Chromosome Xp11.22 duplication syndrome[1,2,12]	ChrXp11.22 duplication (300705)
Chromosome 8p inverted duplication/deletion syndrome[47]	Chr8p invdupdel

Chung-Jansen syndrome (CHUJANS), White-Kernohan syndrome (WHIKERS) and Börjeson-Forssman-Lehmann syndrome (BFLS)	*PHIP, DDB1, PHF6* (617991, 619426, 301900)
Chung-Jansen syndrome (CHUJANS)[2,8Error! Bookmark not defined.]	*PHIP* (617991)
Clark-Baraitser syndrome (CLABARS)	*TRIP12* (617752)
Coffin-Siris syndrome 1 (CSS1)[2,13]	*ARID1B* (135900)
Coffin-Siris syndrome 2 (CSS2)[2,13]	*ARID1A* (614607)
Coffin-Siris syndrome 3 (CSS3)[2,13]	*SMARCB1* (614608)
Coffin-Siris syndrome 4 (CSS4)[2,13]	*SMARCA4* (614609)
Coffin-Siris syndrome 4 (CSS4)[2,13,14]	*SMARCA4* c.2656 (614609)
Coffin-Siris syndrome 5 (CSS5)[13]*	*SMARCE1* (616938)
Coffin-Siris syndrome 6 (CSS6)	*ARID2* (617808)
Coffin-Siris syndrome 7 (CSS7)*	*DPF2* (618027)
Coffin-Siris syndrome 12 (CSS12)[2]†	*BICRA* (619325)
Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder (CHDFIDD)[2]	*CDK13, CCNK* (617360)
Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder (CDK13_truncating)[45]	*CDK13* (617360)
Cornelia de Lange syndromes 1-4 (CDLS1, CDLS2, CDLS3, CDLS4)[15]	*NIPBL, SMC1A, SMC3, RAD21* (122470, 300590, 610759, 614701)
Cornelia de Lange syndrome 1 (CDLS1)[53]	*NIPBL* (122470)
Cornelia de Lange syndrome 2 (CDLS2)[53]	*SMC1A* (300590)
Cornelia de Lange syndrome 3 (CDLS3)[51,53]	*SMC3* (610759)
Cornelia de Lange syndrome 4 (CDLS4)[53]	*RAD21* (614701)
DEGCAGS syndrome[16]	*ZNF699* (619488)
Desanto-Shinawi syndrome (DESSH)†	*WAC* (616708)
Developmental and epileptic encephalopathy 54 (DEE54)	*HNRNPU* (617391)
Developmental and epileptic encephalopathy 94 (DEE94)	*CHD2* (615369)
Developmental delay, dysmorphic facies, and brain anomalies (DEVDFB)*	*U2AF2* (620535)
Developmental delay with or without dysmorphic facies and autism (DEDDFA)[17]†	*TRRAP* (618454)
Developmental delay with variable intellectual disability and dysmorphic facies (DIDDF)	*JARID2* (601594)
Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy (DIGFAN)*	*MORC2* (619090)
Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities (DEHMBA)[50]*	*SRCAP* (619595)
Diamond-Blackfan anemia types 1, 4, 5-7, and 10 (DBA1, DBA4, DBA5-7, DBA10)[49]*	*RPS19, RPS17, RPL35A, RPL5, RPL11, RPS26* (105650, 612527, 612528, 612561, 612562, 613309)
Diamond-Blackfan anemia 1 (DBA1)[18]†	*RPS19* (105650)
Diamond-Blackfan anemia 5 (DBA5)[18]†	*RPL35A* (612528)
Diamond-Blackfan anemia types 6 & 7 (DBA6, DBA7)*	*RPL5, RPL11* (612528, 612561)
Diamond-Blackfan anemia 10 (DBA10)*	*RPS26* (613309)
Diets-Jongmans syndrome (DIJOS)	*KDM3B* (618846)
DMAP1-associated syndrome*	*DMAP1* (605077)

Down syndrome	Chr21 trisomy (190685)
Dystonia 28, Childhood-onset (DYT28)	*KMT2B* (617284)
Euchromatic Histone-Lysine N-methyltransferase 2 disorder*	*EHMT2* (604599)
EHMT1 duplication-related syndrome*	*EHMT1* (607001)
Facioscapulohumeral muscular dystrophy 2, digenic (FSHD2)*	*SMCHD1* (158901)
Fanconi anemia (FANCA, FANCC, FANCD2, FANCG, FANCI, FANCL)[16,19]*	*FANCA, FANCC, FANCD2, FANCG, FANCI, FANCL* (227650, 613899, 613984, 602956, 611360, 608111)
Fetal valproate syndrome (FVS)[20]*	Not applicable (609442)
Floating-Harbor syndrome (FLHS)	*SRCAP* (136140)
Gabriele-de Vries syndrome (GADEVS)[2]	*YY1* (617557)
Genitopatellar syndrome (see also Ohdo syndrome) (GTPTS)[2,21]	*KAT6B* (606170)
Hao-Fountain syndrome (HAFOUS)	*USP7* (616863)
Helsmoortel-van der Aa syndrome (HVDAS)[22]	*ADNP* (615873)
Helsmoortel-van der Aa syndrome (HVDAS), Sifrim-Hitz-Weiss syndrome (SIHIWES)[54]*	*ADNP, CHD4* (615873, 617159)
Hunter-McAlpine craniosynostosis syndrome[23]	Chr5q35 duplication including *NSD1* (601379)
Hypercholesterolemia, familial, 1 (FHCL1)[24]†	*LDLR* (143890)
Hypermethioninemia with S-adenosylhomocysteine hydrolase deficiency*†	*AHCY* (613752)
Immunodeficiency-centromeric instability-facial anomalies syndromes 1-4 (ICF1, ICF2, ICF3, ICF4)[2,25]	*DNMT3B, CDCA7, ZBTB24, HELLS* (242860, 614069, 616910, 616911)
Immunodeficiency 31C, chronic mucocutaneous candidiasis, autosomal dominant (IMD31C)*	*STAT1* (614162)
Intellectual developmental disorder with autism and macrocephaly (IDDAM)[2]	*CHD8* (615032)
Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities (IDDFBA)†	*FBXO11* (618089)
Intellectual developmental disorder with microcephaly and with or without ocular malformations or hypogonadotropic hypogonadism (IDDMOH)[2]	*SOX11* (615866)
Intellectual developmental disorder with seizures and language delay (IDDSELD)	*SETD1B* (619000)
Intellectual developmental disorder with speech delay, dysmorphic facies, and T-cell abnormalities (IDDSFTA)[2]	*BCL11B* (618092)
Intellectual developmental disorder, autosomal dominant 7 (MRD7)*	*DYRK1A* (600855)
Intellectual developmental disorder, autosomal dominant 21 (MRD21)*	*CTCF* (615502)
Intellectual developmental disorder, autosomal dominant 23 (MRD23)[2,26]	*SETD5* (615761)
Intellectual developmental disorder, autosomal dominant 23 (MRD23); KBGS syndrome (KBGS)[2]*	*SETD5, ANKRD11* (615761, 148050)
Intellectual developmental disorder, autosomal dominant 29 (MRD29)*	*SETBP1* (616078)
Intellectual developmental disorder, autosomal dominant 51 (MRD51)[2,6]	*KMT5B* (617788)
Intellectual developmental disorder, autosomal dominant 57 (MRD57)†	*TLK2* (618050)

Intellectual developmental disorder, autosomal dominant 58 (MRD58)*	*SET* (618106)
Intellectual developmental disorder, autosomal recessive 65 (MRT65), KDM5B-related neurodevelopmental disorder*	*KDM5B* (618109)
Intellectual developmental disorder, X-linked 93 (XLID93)[2,6,44]	*BRWD3* (300659)
Intellectual developmental disorder, X-linked 97 (XLID97)[2,27]	*ZNF711* (300803)
Intellectual developmental disorder, X-linked 112 (XLID112)[2]†	*ZMYM3* (301111)
Intellectual developmental disorder, X-linked syndromic, Lubs type (MRXSL)*	*MECP2* (300260)
Intellectual developmental disorder, X-linked syndromic, Nascimento type (MRXSN)[1,2]	*UBE2A* (300860)
Intellectual developmental disorder, X-linked, syndromic, Armfield type (MRXSA)[1,2]	*FAM50A* (300261)
Intellectual developmental disorder, X-linked, syndromic, Claes-Jensen type (MRXSCJ)[6,28]	*KDM5C* (300534)
Intellectual developmental disorder, X-linked syndromic, Siderius type (MRXSSD)[1]†	*PHF8* (300263)
Intellectual developmental disorder, X-linked, syndromic, Snyder-Robinson type (MRXSSR)[1,2]	*SMS* (309583)
Jacobs syndrome (XYY)*	ChrY duplication
Kabuki syndrome 1 (KABUK1)[29]	*KMT2D* (147920)
Kabuki syndrome 1 and 2 (KABUK1, KABUK2)	*KMT2D, KDM6A* (147920, 300867)
Kabuki syndrome 2 (KABUK2)[29]	*KDM6A* (300867)
Karayol-Borroto-Haghshenas neurodevelopmental syndrome (KBHS)[2]*	*MSL2* (620985)
KAT6-related syndromes*	*KAT6A, KAT6B* (601408, 605880)
KBG syndrome (KBGS)[26]	*ANKRD11* (148050)
KDM2B-related syndrome	*KDM2B* (609078)
Kleefstra syndrome 1 (KLEFS1)	*EHMT1* (610253)
Klinefelter Syndrome[30]	ChrX duplication; 47,XXY
KMT2C-related syndrome†*	*KMT2C* (606833)
Koolen-De Vries syndrome (KDVS)	*KANSL1* (610443)
Luscan-Lumish syndrome (LLS)	*SETD2* (616831)
MACROH2A1-associated syndrome*	*MACROH2A1* (610054)
MAU2-associated syndrome*	*MAU2* (614560)
Menke-Hennekam syndrome 1 and 2 (MKHK1, MKHK2)[31]	*CREBBP, EP300* (618332, 618333)
Mowat-Wilson Syndrome (MOWS)	*ZEB2* (235730)
MSL2 associated syndrome[2]	*MSL2* (614802)
Myhre syndrome (MYHRS)*	*SMAD4* (139210)
Neurodevelopmental-craniofacial syndrome with variable renal and cardiac abnormalities (NECRC)†	*ZMYM2* (619522)
Neurodevelopmental disorder with or without autism or seizures (NEDAUS)[2]*	*CUL3* (619239)
Neurodevelopmental disorder with dysmorphic facies and behavioral abnormalities (NEDFBA)	*SRSF1* (620489)

Neurodevelopmental disorder with hypotonia, stereotypic hand movements, and impaired language (NEDHSIL)	*MEF2C* (613443)
Neurodevelopmental disorder with impaired speech and hyperkinetic movements (NEDISHM)[46]*	*ZNF142* (618425)
Neurodevelopmental disorder with microcephaly, cerebral atrophy, and visual impairment (NEDMVIC)*	*DOHH* (620066)
Neurodevelopmental disorder with neonatal respiratory insufficiency, hypotonia, and feeding difficulties (NEDRIHF)[46]*	*PURA* (616158)
Neurofibromatosis, type 1 (NF1)[2]†	*NF1* (162200)
Neuroocular syndrome[2,32]*	*PRR12* (619539)
Nicolaides-Baraitser syndrome (NCBRS)[13]	*SMARCA2* (601358)
NOTCH1-associated syndrome[2]†*	*NOTCH1* (190198)
NSD2 duplication-related syndrome[33]	*NSD2* (602952)
Ohdo syndrome, SBBYSS variant (see also Genitopatellar syndrome) (SBBYSS)[2,21]	*KAT6B* (603736)
Okur-Chung neurodevelopmental syndrome (OCNDS) & Poirier-Bienvenu neurodevelopmental syndrome (POBINDS)[48]*	*CSNK2A1, CSNK2B* (617062, 618732)
Phelan-McDermid syndrome (PHMDS)[34]	Chr22q13.3 deletion (606232)
PHF12-related syndrome†	*PHF12* (618645)
Pitt-Hopkins Syndrome (PTHS)	*TCF4* (610954)
Pitt-Hopkins syndrome (PTHS); Craniosynostosis 3 (CRS3)*	*TCF4, TCF12* (610954, 615314)
Potocki-Lupski syndrome (PTLS)[35]	Chr17p11.2 duplication (610883)
PTBP1-associated syndrome (PTBP1)*	*PTBP1* (600693)
Rahman syndrome (RMNS)	*HIST1H1E* (617537)
Rauch-Steindl syndrome (RAUST)*	*NSD2* (619695)
Recurrent constellations of embryonic malformations[36]†	Not applicable
Renpenning syndrome (RENS1)[1,2]	*PQBP1* (309500)
ReNU syndrome (RENU)*	*RNU4-2* (620851)
RNU2-2 associated syndrome (RNU2-2)[46]*	*RNU2-2* (621238)
Rubinstein-Taybi syndrome 1 (RSTS1)[37]	*CREBBP* (180849)
Rubinstein-Taybi syndrome 1 and 2 (RSTS1, RSTS2)	*CREBBP, EP300* (180849, 613684)
Rubinstein-Taybi syndrome 2 (RSTS2)[37]	*EP300* (613684)
Schinzel-Giedion midface retraction syndrome (SGMRS)*	*SETBP1* (269150)
Schuurs-Hoeijmakers syndrome (SHMS)†	*PACS1* (615009)
SETD1A-related syndrome†*	*SETD1A* (611052)
Sex Chromosome Aneupoidy: Trisomy X, Klinefelter syndrome, XYY syndrome/Jacobs syndrome*	ChrX, ChrY
Sifrim-Hitz-Weiss syndrome (SIHIWES)[52]	*CHD4* (617159)
SMARCA5-associated syndrome (SMARCA5)*	*SMARCA5* (603375)
SMARCA5-related syndrome; Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies (SMARCA5_NEDDFL)*	*SMARCA5, BPTF* (603375, 617755)
Smith-Kingsmore syndrome (SKS)*	*MTOR* (616638)
Smith-Magenis syndrome (SMS)[38]	Chr17p11.2 deletion (182290)

Sotos syndrome (SOTOS)	*NSD1* (117550)
Tatton-Brown-Rahman syndrome (TBRS)[2]	*DNMT3A* (615879)
Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 1, 3 and 4 (TEBIVANED1, TEBIVANED3, TEBIVANED4)†	*H4C3, H4C4, H4C5, H4C9* (619758, 602823, 619950, 619951)
Trisomy X (XXX)*	ChrX triplication
Turner Syndrome	ChrX deletion; 45,X; Monosomy X
Uniparental disomy of chromosome 16 (UPD16)*	Chr16
Velocardiofacial syndrome (VCFS)[39]	Chr22q11.2 deletion (192430)
WAPL-associated syndrome (WAPL)*	*WAPL* (610754)
WAPL-associated syndrome, PDS5A-associated syndrome, PDS5B-associated syndrome (WAPL-PDS5A-PDS5B)[52]*	*WAPL, PDS5A, PDS5B* (610754, 613200, 605333)
Weaver syndrome; Cohen-Gibson syndrome; Imagawa-Matsumoto syndrome (PRC2)[36]*	*EZH2, EED, SUZ12* (277590, 617561, 618786)
White-Kernohan syndrome[8]	*DDB1* (619426)
White-Sutton syndrome (WHSUS)	*POGZ* (616364)
Wieacker-Wolff Syndrome (WRWF)[1,2]	*ZC4H2* (314580)
Wiedemann-Steiner syndrome (WDSTS)	*KMT2A* (605130)
Williams-Beuren region duplication syndrome[40]	Chr7q11.23 duplication (609757)
Williams-Beuren syndrome (WBS)[41]	Chr7q11.23 deletion (194050)
Witteveen-Kolk syndrome (WITKOS)[2]	*SIN3A* (613406)
Wolf-Hirschhorn syndrome (WHS) & Rauch-Steindl Syndrome (RAUST)[42]	Chr4p16.13 deletion, *NSD2* (194190, 602952)
Xia-Gibbs syndrome (XIGIS)[52]*	*AHDC1* (615829)

**These disorders are new for V6 of the EpiSign™ classifier.*

†These episignatures are available for EpiSign™ Single requests only, not available through EpiSign™ METRIC.

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[1] Defined with male cases only. Heterozygotes have been shown to not match the episignature.

[2] Reduced sensitivity may be observed.

[3] The range of validated coordinates is 1p36.11(26,964,202-27,099,490). CNVs overlapping or expanding this region may also be detected.

[4] Patients with other BAFopathy genes may be detected, but not confirmed in our experiments.

[5] Only for variants near c.6200. No separate episignature due small cohort size, however these samples cluster separately from other BAFopathy/CSS1&2 samples.

[6] Healthy carriers and those with incomplete penetrance are detectable.

[7] Patients with biallelic variants are distinguishable from those with monoallelic variants.

[8] This is a secondary episignature; sample must also be positive for CHU_BFL_WHI.

[9] Only for variants within the amino acid range of 3400-3700.

[10] The range of validated coordinates is 1p36.33p36.32(1,019,753-2,867,961). CNVs overlapping or expanding this region may also be detected.

[11] The range of validated coordinates is 19p13.13p13.2(13,201,983-13,213,144). CNVs overlapping or expanding this region may also be detected. Only for copy number variants. NFIX sequence variants have been shown to not match the episignature.

[12] The range of validated coordinates is Xp11.22(53,559,057-53,654,518). CNVs overlapping or expanding this region may also be detected.

[13] This is a secondary episignature; sample must also be positive for BAFopathy.

[14] Only for variants near c.2656. No separate episignature due small cohort size however these samples cluster separately from other BAFopathy/CSS4 samples.

[15] Male CdLS5 patients (HDAC8 mutations) may be detected, but not confirmed in our experiments.

[16] Heterozygotes have been shown to not match the episignature.
[17] Only for variants within the amino acid range of 960-1159.
[18] Reduced sensitivity against other Diamon-Blackfan anemia disorders may be observed.
[19] Patients with other FANC genes may be detected, but not confirmed in our experiments.
[20] Available as a targeted request only.
[21] GTPTS and SBBYSS are both caused by KAT6B mutations. We will report both regardless of which one is requested.
[22] ADNP consists of two distinct episignatures dependent on variant location. HVDAS_T includes variants within the N- and C-terminus while HVDAS_C includes variants within the central region (approximately c.2054-2340).
[23] The range of validated coordinates is 5q35.2q35.3(175,839,681-176,904,798). CNVs overlapping or expanding this region may also be detected.
[24] Sensitivity against other hereditary hypercholesterolemia disorders has not been evaluated. Both monoallelic and biallelic cases are detected.
[25] ICF1 exhibits a unique episignature while ICF 2, 3 and 4 exhibit a distinct, shared episignature
[26] This is a secondary episignature; sample must also be positive for KBGS_MRD23.
[27] Heterozygotes have been shown to match the episignature.
[28] Heterozygotes have a distinct profile from hemizygotes.
[29] This is a secondary signature; sample must also be positive for combined Kabuki signature.
[30] XXX and XYY cases may also be detected.
[31] Only for domain ID4. MKHK1/2 exhibit a shared ID4 domain episignature and therefore cannot distinguish between MKHK1 and MKHK2. Other domains of MKHK1/2 are not available for assessment.
[32] Healthy carriers and those with incomplete penetrance are detectable. Reduced sensitivity may be observed.
[33] The range of validated coordinates is 4p16.3(1,832,733-1,975,031). CNVs overlapping or expanding this region may also be detected.
[34] 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.
[35] 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.
[36] Includes cases with phenotypic presentation of OAV, OAVV, VACTERL and VATER.
[37] This is a secondary signature; sample must also be positive for combined RSTS signature.
[38] 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.
[39] The range of validated coordinates is 22q11.21(19,510,547-20,285,090). CNVs overlapping or expanding these regions may be detected.
[40] The range of validated coordinates is 7q11.23(73,953,518-74,138,459). CNVs overlapping or expanding this region may also be detected.
[41] CNVs overlapping or expanding 7q11.23 may also be detected.
[42] 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.
[44] Defined with female cases only.
[45] There is a robust missense episignature for the condition. This is a new truncating episignature that is not as robust.
[46] Episignature developed using synthetic data.
[47] Episignature defined with 12 samples, 11 invdupdel and one del only. The del only does not match the identified pattern Deletion only cases do not match the episignature.
[48] Signature detects variants in casein kinase II (CSNK2) subunits (CSNK2A1 and CSNK2B).
[49] Episignature may detect other Diamond-Blackfan anemia types.
[50] Truncating mutations outside of the FLHS region.
[51] This episignature detects loss of function variants.
[52] This episignature detects truncating variants.
[53] This is a secondary episignature; sample must also be positive for CdLS. Reduced sensitivity may be observed.
[54] Signature can detect HVDAS variants outside of the coding nucleotide range of c.2054-2340 and SIHIWES variants.
[55] Signature defined with missense variants at p.986 and p.987