

Disorders of Transcriptional Regulation

Kabuki syndrome, type 1	147920	<i>KMT2D</i>
Kabuki syndrome, type 2	602113	<i>KDM6A</i>
CHARGE syndrome	214800	<i>CHD7</i>
Rubinstein-Taybi, type 1	180849	<i>CREBBP</i>
Rubinstein-Taybi, type 2	613684	<i>EP300</i>
Wolf-Hirschhorn syndrome	194190	<i>WHSC2 (NELF)</i>
ICF syndrome type 1	242860	<i>DNMT3B</i>
ICF syndrome type 2	614069	<i>ZBTB24</i>
Wiedemann Steiner syndrome	605130	<i>KMT2A</i>
Kleefstra syndrome type 1	610253	<i>EHMT1</i>
Kleefstra syndrome type 2	617768	<i>KMT2C</i>
Koolen-De Vries syndrome	610443	<i>KANSL1</i>
Ohdo syndrome, SBBYS	603736	<i>KAT6B</i>
Soto syndrome type 1	117550	<i>NSD1</i>
Weaver syndrome	277590	<i>EZH2</i>
Soto-like / Luscan-Lumish syndrome	616831	<i>SETD2</i>
Tatton-Brown-Rahman syndrome	615879	<i>DNMT3A</i>
Hereditary Sensory Neuropathy type 1E (HSN1E)	614116	<i>DNMT1 (exon 20)</i>
Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant (ADCADN)	604121	<i>DNMT1 (exon 21)</i>
Floating Harbor syndrome	136140	<i>SRCAP</i>
ATR-X syndrome (MENTAL RETARDATION-HYPOTONIC FACIES SYNDROME, X-LINKED)	309580	<i>ATRX</i>
Coffin-Siris syndrome-type 2 (MRD14)	614607	<i>ARID1A</i>
Coffin-Siris syndrome- type 4 (MRD16)	614609	<i>SMARCA4</i>
Rhabdoid tumor predisposition syndrome-1 (RTPS1)	609322	<i>SMARCB1</i>
Rhabdoid tumor predisposition syndrome-2 (RTPS2)	613325	<i>SMARCA4</i>
Schwannomatosis, type 1	162091	<i>SMARCB1</i>
Coffin-Siris syndrome type 3	614608	<i>SMARCB1</i>
Nicolaidis-Baraitser syndrome (NCBRS) Intellectual disability-sparse hair-brachydactyly syndrome	601358	<i>SMARCA2</i>
Childhood-onset epileptic encephalopathy (EEOC)	615369	<i>CHD2</i>
CHD8 disorder / Autism spectrum disorder-18	615032	<i>CHD8</i>
Rett syndrome	312750	<i>MECP2</i>
MRX93 (X-linked mental retardation (XLMR) associated with macrocephaly)	300659	<i>BRWD3</i>
Bohring-Opitz syndrome (c-like syndrome)	605039	<i>ASXL1</i>
Boerjeson-Forssman-Lehmann syndrome (BFLS)	301900	<i>PHF6</i>
MBD5-associated neurodevelopmental disorder (MAND)	156200	<i>MBD5</i>

Smith-Magenis syndrome	182290	<i>RAI1</i>
BDMR syndrome	600430	<i>HDAC4</i>
Cornelia de Lange syndrome type 5	300882	<i>HDAC8</i>
Siderius XLMR syndrome	300263	<i>PHF8</i>
Townes-Brocks syndrome-1	107480	<i>SALL1</i>
Claes - Jensen syndrome	300534	<i>KDM5C</i>
Cornelia de Lange syndrome 1	122470	<i>NIPBL</i>
Cornelia de Lange syndrome 2	300590	<i>SMC1A</i>
Cornelia de Lange syndrome 3	610759	<i>SMC3</i>
Cornelia de Lange syndrome 4	614701	<i>RAD21</i>
Cornelia de Lange syndrome 5	300882	<i>HDAC8</i>