

Chromatinopathia (Chromatinopathy) génpanel v:2025-05-21

Panel tartalma 131 gén:

ACTB, ACTL6A, ACTL6B, ADNP, ALG13, ARID1A, ARID1B, ARID2, ASH1L, ASXL1, ASXL2, ASXL3, ATRX, AUTS2, BCOR, BCORL1, BPTF, BRD4, BRPF1, BRWD3, CDK9, CHD1, CHD2, CHD3, CHD4, CHD7, CHD8, CREBBP, CTBP1, CTCF, CTNNB1, CUL4B, DDB2, DDX11, DNMT3A, DNMT3B, DPF2, EED, EHMT1, EP300, ERCC6, EZH2, FMR1, GATAD2B, GPX4, H1-4, H4C3, HCFC1, HDAC4, HDAC8, HLCS, HNRNPU, HUWE1, KANSL1, KAT6A, KAT6B, KDM1A, KDM3B, KDM5B, KDM5C, KDM6A, KDM6B, KMT2A, KMT2B, KMT2C, KMT2D, KMT2E, KMT5B, KPNA7, MAP3K7, MBD5, MCM3AP, MECP2, MORC2, MSL3, NCOR1, NIPBL, NSD1, NSD2, OGT, ORC1, PAK1, PCGF2, PHF21A, PHF6, PHF8, PHIP, PPM1D, PRMT7, RAI1, RBPJ, RCBTB1, RERE, SATB2, SET, SETD1A, SETD1B, SETD2, SETD5, SIN3A, SKI, SMAD4, SMARCA2, SMARCA4, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SOX2, SRCAP, SUPT16H, SUZ12, TAF1, TAF6, TBL1XR1, TCF20, TET3, TLK2, TRIM37, TRIP12, TRRAP, TWIST1, UBE2A, USP7, VRK1, WAC, YY1, ZMIZ1, ZMYND11, ZNF335, ZNF462

Forrás:

Ciptasari U, van Bokhoven H. The phenomenal epigenome in neurodevelopmental disorders. *Hum Mol Genet.* 2020;29(R1):R42-R50. doi: 10.1093/hmg/ddaa175. Supplementary_Table_3_ddaa175.