

Publication list Chromatin - Net

Maternal transmission of a mild Coffin-Siris syndrome phenotype caused by a SOX11 missense variant

European Journal of Human Genetics. 2021 Mar 31. >>[PubMed-Link](#)<<
Hanker B, Gillissen-Kaesbach G, Huning I, Ludecke HJ, Wieczorek D.

Unique signatures of stress-induced senescent human astrocytes

Experimental Neurology 2020 Dec;334:113466 >>[PubMed-Link](#)<<
Simmnacher K, Krach F, Schneider Y, Alecu JE, Mautner L, Klein P, Roybon L, Prots I, Xiang W, Winner B.

Intracellular A53T Mutant alpha-Synuclein Impairs Adult Hippocampal Newborn Neuron Integration

Frontiers in Cell and Developmental Biology 2020 Nov 11;8:561963 >>[PubMed-Link](#)<<
Regensburger M, Stemick J, Masliah E, Kohl Z, Winner B.

Janus-faced spatacsin (SPG I I) involvement in neurodevelopment and multisystem neurodegeneration

Brain 2020 Aug 1;143(8):2369-2379 >>[PubMed-Link](#)<<
Pozner T, Regensburger E, Engelhorn T, Winkler J, Winner B.

CRISPR/Cas9 mediated generation of human ARID1B heterozygous knockout hESC lines to model Coffin-Siris syndrome

Stem Cell Research 2020 Jun 29;47:101889 >>[PubMed-Link](#)<<
Boerstler T, Wend H, Krumbiegel M, Kavyanifar A, Reis A, Lie DC, Winner B, Turan S.

Polyhydroxyphenylvalerate/polycaprolactone nanofibers improve the life-span and mechanoreponse of human iPSC-derived cortical neuronal cells

Materials Science & Engineering C-Materials for Biological Applications 2020 Jun;111:110832 >>[PubMed-Link](#)<<
Cerrone F, Pozner T, Siddiqui A, Ceppi P, Winner B, Rajendiran M, Babu R, Ibrahim HS, Rodriguez BJ, Winkler J, Murphy KJ, O'Connor KE

Evaluating Face2Gene as a Tool to Identify Cornelia de Lange Syndrome by Facial Phenotypes

International Journal of Molecular Sciences 2020 Feb 4;21(3):1042 >>[PubMed-Link](#)<<
Latorre-Pellicer A, Ascaso A, Trujillano L, Gil-Salvador M, Arnedo M, Lucia-Campos C, Antonanzas-Perez R, Marcos-Alcalde I, Parenti I, Bueno-Lozano G, Musio A, Puisac B, Kaiser FJ, Ramos FJ, Gomez-Puertas P, Pie J.

Prenatal diagnosis of HNF1B-associated renal cysts: Is there a need to differentiate intragenic variants from 17q12 microdeletion syndrome?

Prenatal Diagnosis 2019 Nov;39(12):1136-1147.
Vasileiou G, Hoyer J, Thiel CT, Schaefer J, Zapke M, Krumbiegel M, Kraus C, Zweier M, Uebe S, Ekici AB, Schneider M, Wiesener M, Rauch A, Faschingbauer F, Reis A, Zweier C, Popp B.

Genome-Wide Analysis of the Nucleosome Landscape in Individuals with Coffin-Siris Syndrome

Cytogenetic and Genome Research 2019; 159(1): 1-11 [Epub 2019 Oct 26]
Kalmbach A, Schroder C, Klein-Hitpass L, Nordstrom K, Ulz P, Heitzer E, Speicher MR, Rahmann S, Wieczorek D, Horsthemke B, Bramswig NC.

Disease Modeling of Neuropsychiatric Brain Disorders Using Human Stem Cell-Based Neural Models

Curr Top Behav Neurosci. 2019 Aug 13. doi: 10.1007/7854_2019_111. [Epub ahead of print]
Kaindl J, Winner B

Mutations in SMARCB1 and in other Coffin-Siris syndrome genes lead to various brain midline defects

Nat Commun. 2019 Jul 4;10(1):2966. doi: 10.1038/s41467-019-10849-y

Filatova A, Rey LK, Lechler MB, Schaper J, Hempel M, Posmyk R, Szczaluba K, Santen GWE, Wiczorek D, Nuber U

[DETAILS](#)

Dissecting TSC2-mutated renal and hepatic angiomyolipomas in an individual with ARID1B-associated intellectual disability

Bmc Cancer 2019 May 19; 19(1):435.

Popp B, Agaimy A, Kraus C, Knaup KX, Ekici AB, Uebe S, Reis A, Wiesener M, Zweier C.

Dissecting TSC2-mutated renal and hepatic angiomyolipomas in an individual with ARID1B-associated intellectual disability

BMC Cancer. 2019 May 10;19(1):435

Popp B, Agaimy A, Kraus C, Knaup KX, Ekici AB, Uebe S, Reis A, Wiesener M, Zweier C.(2019)

Human SPG11 cerebral organoids reveal cortical neurogenesis impairment

Human Molecular Genetics 2019 Mar 15; 28(6): 961-971

Perez-Branguli F, Buchsbaum IY, Pozner T, Regensburger M, Fan WQ, Schray A, Borstler T, Mishra H, Graf D, Kohl Z, Winkler J, Berninger B, Cappello S, Winner B.

Zooming in on Cryopreservation of hiPSCs and Neural Derivatives: A Dual-Center Study Using Adherent Vitrification

Stem Cells Translational Medicine 2019 Mar; 8(3): 247-259

Kaindl J, Meiser I, Majer J, Sommer A, Krach F, Katsen-Globa A, Winkler J, Zimmermann H, Neubauer JC, Winner B.

Pathogenic variants in USP cause a neurodevelopmental disorder with speech delays, altered behavior, and neurologic anomalies

Genet Med. 2019 doi: 10.1038/s41436-019-0433-1. [Epub ahead of print]

Fountain MD, Oleson DS, Rech ME, Segebrecht L, Hunter JV, McCarthy JM, Lupo PJ, Holtgrewe M, Moran R, Rosenfeld JA, Isidor B, Le Caignec C, Saenz MS, Pedersen RC, Morgan TM, Pfothenhauer JP, Xia F, Bi W, Kang SL, Patel A, Krantz ID, Raible SE, Smith W, Cristian I, Torti E, Juusola J, Millan F, Wentzensen IM, Person RE, Küry S, Bézieau S, Uguen K, Férec C, Munnich A, van Haelst M, Lichtenbelt KD, van Gassen K, Hagelstrom T, Chawla A, Perry DL, Taft RJ, Jones M, Masser-Frye D, Dymant D, Venkateswaran S, Li C, Escobar LF, Horn D, Spillmann RC, Peña L, Wierzba J, Strom TM, Parenti I, Kaiser FJ, Ehmke N, Schaaf CP (2019)

[DETAILS](#)

Pain relief in a neuropathy patient by lacosamide: Proof of principle of clinical translation from patient-specific iPSC cell-derived nociceptors

EBioMedicine. 2019 Jan;39:401-408. doi: 10.1016/j.ebiom.2018.11.042. Epub 2018 Nov 28.

Namer B, Schmidt D, Eberhardt E, Maroni M, Dorfmeister E, Kleggetveit IP, Kaluza L, Meents J, Gerlach A, Lin Z, Winterpacht A, Dragicevic E, Kohl Z, Schüttler J, Kurth I, Warncke T, Jorum E, Winner B, Lampert A.

Tideglusib Rescues Neurite Pathology of SPG11 iPSC Derived Cortical Neurons

Front Neurosci. 2018 Dec 6;12:914. doi: 10.3389/fnins.2018.00914. eCollection 2018

Pozner T, Schray A, Regensburger M, Lie DC, Schlötzer-Schrehardt U, Winkler J, Turan S, Winner B.

Need for high-resolution Genetic Analysis in iPSC: Results and Lessons from the ForIPS Consortium

Sci Rep. 2018 Nov 21;8(1):17201. doi: 10.1038/s41598-018-35506-0.

Popp B, Krumbiegel M, Grosch J, Sommer A, Uebe S, Kohl Z, Plötz S, Farrell M, Trautmann U, Kraus C, Ekici AB, Asadollahi R, Regensburger M, Günther K, Rauch A, Edenhofer F, Winkler J, Winner B, Reis A.

Zooming in on Cryopreservation of hiPSCs and Neural Derivatives: A Dual-Center Study Using Adherent Vitrification

Stem Cells Transl Med. 2018 Nov 19. doi: 10.1002/sctm.18-0121.

Kaindl J, Meiser I, Majer J, Sommer A, Krach F, Katsen-Globa A, Winkler J, Zimmermann H, Neubauer JC, Winner B.

FoxO Function Is Essential for Maintenance of Autophagic Flux and Neuronal Morphogenesis in Adult Neurogenesis

Neuron. 99:1188-1203

Schäffner I, Minakaki G, Khan MA, Balta EA, Schlötzer-Schrehardt U, Schwarz TJ, Beckervordersandforth R, Winner B, Webb AE, DePinho RA, Paik J, Wurst W, Klucken J, Lie DC

alpha-Synuclein oligomers induce early axonal dysfunction in human iPSC-based models of synucleinopathies

Proceedings of the National Academy of Sciences of the United States of America 2018 Jul 24; 115(30): 7813-7818;

Prots I, Grosch J, Brazdis RM, Simmnacher K, Veber V, Havlicek S, Hannappel C, Krach F, Krumbiegel M, Schutz O, Reis A, Wrasidlo W, Galasko DR, Groemer TW, Masliah E, Schlötzer-Schrehardt U, Xiang W, Winkler J, Winner B.

A monocyte gene expression signature in the early clinical course of Parkinson's disease

Sci Rep. 8:10757

Schlachetzki JCM, Prots I, Tao J, Chun HB, Saijo K, Gosselin D, Winner B, Glass CK, Winkler J

Diagnosis and management of Cornelia de Lange syndrome: first international consensus statement

Nat Rev Genet. 2018 Jul 11. doi: 10.1038/s41576-018-0031-0.

Kline AD, Moss JF, Selicorni A, Bisgaard AM, Deardorff MA, Gillett PM, Ishman SL, Kerr LM, Levin AV, Mulder PA, Ramos FJ, Wierzbica J, Ajmone PF, Axtell D, Blagowidow N, Cereda A, Costantino A, Cormier-Daire V, FitzPatrick D, Grados M, Groves L, Guthrie W, Huisman S, Kaiser FJ, Koekkoek G, Levis M, Mariani M, McCleery JP, Menke LA, Metrena A, O'Connor J, Oliver C, Pie J, Piening S, Potter CJ, Quaglio AL, Redeker E, Richman D, Rigamonti C, Shi A, Tümer Z, Van Balkom IDC, Hennekam RC

α-Synuclein oligomers induce early axonal dysfunction in human iPSC-based models of synucleinopathies

Proc Natl Acad Sci USA. 115:7813–7818.

Prots I, Grosch J, Brazdis RM, Simmnacher K, Veber V, Havlicek S, Hannappel C, Krach F, Krumbiegel M, Schütz O, Reis A, Wrasidlo W, Galasko DR, Groemer TW, Masliah E, Schlötzer-Schrehardt U, Xiang W, Winkler J, Winner B

Sporadic Parkinson's disease derived neuronal cells show disease-specific mRNA and small RNA signatures with abundant deregulation of piRNAs

Acta Neuropathol Commun 2018

Schulze M, Sommer A, Plötz S, Farrell M, Winner B, Grosch J, Winkler J, Riemenschneider MJ

Th17 Lymphocytes Induce Neuronal Cell Death in a Human iPSC-Based Model of Parkinson's Disease

Cell Stem Cell. 23:123-131.

Sommer A, Maxreiter F, Krach F, Fadler T, Grosch J, Maroni M, Graef D, Eberhardt E, Riemenschneider MJ, Yeo GW, Kohl Z, Xiang W, Gage FH, Winkler J, Prots I, Winner B

Impact of Swiprosin-1/Efh2 on adult hippocampal neurogenesis

Stem Cell Reports 10:347-355(2018)

Regensburger M, Prots I, Reimer D, Brachs S, Loskarn S, Lie DC, Mielenz D, Winner B

Mutations in the BAF-Complex Subunit DPF2 Are Associated with Coffin-Siris Syndrome

Am J Hum Genet. 2018 Mar 1;102(3):468-479. doi: 10.1016/j.ajhg.2018.01.014.

Vasileiou G, Vergarajauregui S, Endeles S, Popp B, Büttner C, Ekici AB, Gerard M, Bramswig NC, Albrecht B, Clayton-Smith J, Morton J, Tomkins S, Low K, Weber A, Wenzel M, Altmüller J, Li Y, Wollnik B, Hoganson G, Plona MR, Cho MT; Deciphering Developmental Disorders Study, Thiel CT, Lüdecke HJ, Strom TM, Calpena E, Wilkie AOM, Wiczorek D, Engel FB2, Reis A.

[DETAILS](#)

Regulation of the cohesin-loading factor NIPBL: Role of the lncRNA NIPBL-AS1 and identification of a distal enhancer element

PLoS Genet. 2017 Dec 20;13(12):e1007137. doi: 10.1371/journal.pgen.1007137. eCollection 2017 Dec

Zuin J, Casa V, Pozojevic J, Kolovos P, van den Hout MCGN, van Ijcken WFJ, Parenti I, Braunholz D, Baron Y, Watrin E, Kaiser FJ, Wendt KS.

Regulation of the cohesin-loading factor NIPBL: Role of the lncRNA NIPBL-AS1 and identification of a distal enhancer element

Plos Genetics 2017 Dec 20; 13(12): 24 >>[PubMed-Link](#)<<

Zuin J, Casali V, Pozojevic J, Kolovos P, van den Hout M, van Ijcken WFJ, Parenti I, Braunholz D, Baron Y, Watrin E, Kaiser FJ, Wendt KS.

Novel mosaic variants in two patients with Cornelia de Lange syndrome

Eur J Med Genet. 2017 Nov 15. pii: S1769-7212(17)30498-6. doi: 10.1016/j.ejmg.2017.11.004.

Pozojevic J, Parenti I, Graul-Neumann L, Ruiz Gil S, Watrin E, Wendt KS, Werner R, Strom TM, Gillissen-Kaesbach G, Kaiser FJ.

Oligomer-prone E57K-mutant alpha-synuclein exacerbates integration deficit of adult hippocampal newborn neurons in transgenic mice

Brain Struct Funct 223:1357-1368.

Regensburger M, Schreglmann SR, Stoll S, Rockenstein E, Loskarn S, Xiang W, Masliah E, Winner B

Haploinsufficiency of the Chromatin Remodeler BPTF Causes Syndromic Developmental and Speech Delay, Postnatal Microcephaly, and Dysmorphic Features.

Am J Hum Genet. 2017 Oct 5;101(4):503-515. doi: 10.1016/j.ajhg.2017.08.014. Epub 2017 Sep 21.

Stankiewicz P, Khan TN, Szafranski P, Slattery L, Streff H, Vetrini F, Bernstein JA, Brown CW, Rosenfeld JA, Rednam S, Scollon S, Bergstrom KL, Parsons DW, Plon SE, Vieira MW, Quaio CRDC, Baratela WAR, Acosta Guio JC, Armstrong R, Mehta SG, Rump P, Pfundt R, Lewandowski R, Fernandes EM, Shinde DN, Tang S, Hoyer J, Zweier C, Reis A, Bacino CA, Xiao R, Breman AM, Smith JL; Deciphering Developmental Disorders Study, Katsanis N, Bostwick B, Popp B, Davis EE, Yang Y.

Phenotypes and genotypes in individuals with SMC1A variants.

Huisman S, Mulder PA, Redeker E, Bader I, Bisgaard AM, Brooks A, Cereda A, Cinca C, Clark D, Cormier-Daire V, Deardorff MA, Diderich K, Elting M, van Essen A, Patrick DF, Gervasini C, Gillissen-Kaesbach G, Girisha KM, Hilhorst-Hofstee Y, Hopman S, Horn D, Isrie M, Jansen S, Jespersgaard C, Kaiser FJ, Kaur M, Kleefstra T, Krantz ID, Lakeman P, Landlust A, Lessel D, Michot C, Moss J, Noon SE, Oliver C, Parenti I, Pie J, Ramos FJ, Rieubland C, Russo S, Selicorni A, Tümer Z, Vorstenbosch R, Wenger TL, van Balkom I, Piening S, Wierzba J, Hennekam RC.

Am J Med Genet A. 2017 May 26. doi: 10.1002/ajmg.a.38279. [Epub ahead of print]

Mutations in chromatin regulators functionally link Cornelia de Lange syndrome and clinically overlapping phenotypes.

Hum Genet. 2017 Mar;136(3):307-320. doi: 10.1007/s00439-017-1758-y.

Parenti I, Teresa-Rodrigo ME, Pozojevic J, Ruiz Gil S, Bader I, Braunholz D, Bramswig NC, Gervasini C, Larizza L, Pfeiffer L, Ozkinay F, Ramos F, Reiz B, Rittinger O, Strom TM, Watrin E, Wendt K, Wiczorek D, Wollnik B, Baquero-Montoya C, Pié J, Deardorff MA, Gillissen-Kaesbach G, Kaiser FJ.

mRNA Quantification of NIPBL Isoforms A and B in Adult and Fetal Human Tissues, and a Potentially Pathological Variant Affecting Only Isoform A in Two Patients with Cornelia de Lange Syndrome

Int J Mol Sci. 2017 Feb 23;18(3). pii: E481. doi: 10.3390/ijms18030481

Puisac B, Teresa-Rodrigo ME, Hernández-Marcos M, Baquero-Montoya C, Gil-Rodríguez MC, Visnes T, Bot C, Gómez-Puertas P, Kaiser FJ, Ramos FJ, Ström L, Pié J

Heterozygosity for ARID2 loss-of-function mutations in individuals with a Coffin-Siris syndrome-like phenotype

Hum Genet. 136:297-305.

Bramswig NC, Caluseriu O, Lüdecke HJ, Bolduc FV, Noel NC, Wieland T, Surowy HM, Christen HJ, Engels H, Strom TM, Wieczorek D. (2017)

Epigenetic dynamics of monocyte-to-macrophage differentiation

Epigenetics Chromatin 9:33.

Wallner S, Schröder C, Leitão E, Berulava T, Haak C, Beißer D, Rahmann S, Richter AS, Manke T, Bönisch U, Arrigoni L, Fröhler S, Klironomos F, Chen W, Rajewsky N, Müller F, Ebert P, Lengauer T, Barann M, Rosenstiel P, Gasparoni G, Nordström K, Walter J, Brors B, Zipprich G, Felder B, Klein-Hitpass L, Attenberger C, Schmitz G, Horsthemke B

Rescue of GSK3/β-Catenin-dependent human neuronal precursor proliferation defects in spatacsin-linked motor neuron disease

Ann Neurol 79:826-840.(2016)

Mishra HK, Prots I, Havlicek S, Kohl Z, Perez-Branguli F, Boerstler T, Anneser L, Minakaki G, Wend H, Hampf M, Leone M, Brückner M, Klucken J, Reis A, Boyer L, Schuierer G, Behrens J, Lampert A, Engel FB, Gage FH, Winkler J, Winner B

GSK3ss-Dependent Dysregulation of Neurodevelopment in SPG11-Patient Induced Pluripotent Stem Cell Model

Annals of Neurology 2016; 79(5): 826-840; [>>>PubMed-Link<<](#)

Mishra HK, Prots I, Havlicek S, Kohl Z, Perez-Branguli F, Boerstler T, Anneser L, Minakaki G, Wend H, Hampf M, Leone M, Bruckner M, Klucken J, Reis A, Boyer L, Schuierer G, Behrens J, Lampert A, Engel FB, Gage FH, Winkler J, Winner B.