



Publications s'appuyant sur les données du PFMG2025

Septembre 2025

- Courdier C et al. The phenotypic spectrum of CEP250 gene variants. *Genet.* 2024 Nov 28;1-8. Helbling-Leclerc A et al. Biallelic germline BRCA1 frameshift mutations associated with isolated diminished ovarian reserve. *Int J Mol Sci.* 2024 Nov 20;25(22):12460.
- Racine C et al. De novo balanced translocations disrupting the FBN1 gene diagnosed by genome sequencing: an uncommon cause of Marfan Syndrome modifying genetic counseling. *Am J Med Genet A.* 2024 Nov 25:e63923.
- Favier Met al. Fetal presentation of MYRF-related cardiac urogenital syndrome: an emerging and challenging prenatal diagnosis. *J.Prenat Diagn.* 2024 Dec;44(13):1647-1658.
- Kraft F et al. Brain malformations and seizures by impaired chaperonin function of TRiC. *Science.* 2024 Nov;386(6721):516-525.
- Tan S et al. Mol Psychiatry. Monoallelic loss-of-function variants in GSK3B lead to autism and developmental delay. 2024 Oct 29. Online ahead of print.
- Gong M et al. MARK2 variants cause autism spectrum disorder via the downregulation of WNT/ β -catenin signaling pathway. *Am J Hum Genet.* 2024 Oct 11:S0002-9297(24)00366-5
- Stehr AM et al. Consolidating the role of mutated ATP2B2 in neurodevelopmental and cerebellar pathologies. *Clin Genet.* 2024 Oct 5.
- Symonds JD et al. POLR3B is associated with a developmental and epileptic encephalopathy with myoclonic atonic seizures and ataxia. *Epilepsia.* 2024 Sep 30
- Pingault V et al. Chromatin assembly factor subunit CHAF1A as a monogenic cause for oculo-auriculo-vertebral spectrum. *Eur J Hum Genet.* 2024 Sep 27.
- Ham H et al. Germline mutations in a G protein identify signaling cross-talk in T cells. *Science.* 2024 Sep 20;385(6715):eadd8947.
- Maraval J et al. Expanding MNS1 heterotaxy phenotype. *Am J Med Genet A.* 2024 Sep 5:e63862.
- Jury J et al. Multiple congenital anomalies in two fetuses with glutathione-synthetase deficit (GSS). *Clin Genet.* 2024 Sep 2.
- Bernard E, et al. A Novel De Novo missense mutation in KIF1A associated with young-onset upper-limb Amyotrophic Lateral Sclerosis. *Int J Mol Sci.* 2024 Jul 26;25(15):8170.
- Jury J et al. Prenatal diagnosis of Myhre syndrome in two cases: further delineation of the cardiac and external phenotype. *Prenat Diagn.* 2024 Aug 8.
- Kayal D et al. Unveiling atypical diagnoses: when whole-genome analysis performed for refractory infantile hypomagnesemia reveals primary hyperoxaluria. *Pediatr Nephrol.* 2024 Aug 1.
- Schneider V et al. Compound heterozygous WARS2 variants including a hypomorphic allele cause a milder phenotype of complex dopa responsive dystonia: case report and review of the literature. *Cerebellum.* 2024 Jul 29.

- Desjardins C et al. A novel pattern of dystonia in DYT-VPS16 : speaking in tongues. *Neurol Genet.* 2024 Jul 8;10(4):e200154.
- Huang Y et al. Loss-of-function in RBBP5 results in a syndromic neurodevelopmental disorder associated with microcephaly. *Genet Med.* 2024 Jul19;26(11):101218.
- Marelli C et al. Phenotypic variability related to dominant UCHL1 mutations: about three families with optic atrophy and ataxia. *J Neurol.* 2024 Sep;271(9):6038-6044.
- Rots D et al. Pathogenic variants in KMT2C result in a neurodevelopmental disorder distinct from Kleefstra and Kabuki syndromes. *Am J Hum Genet.* 2024 Aug 8;111(8):1626-1642.
- Dorval G et al. Targeted RNAseq from patients' urinary cells to validate pathogenic noncoding variants in autosomal dominant polycystic kidney disease genes: a proof of concept. *Kidney Int.* 2024 Sep;106(3):532-535.
- Theuriet J et al. A previously unreported NARS1 variant causes dominant distal hereditary motor neuropathy in a French family. *J Peripher Nerv Syst.* 2024 Jun;29(2):275-278.
- Mouren A et al. A de novo germline pathogenic BRCA1 variant identified following an osteosarcoma pangenomic molecular analysis. *Fam Cancer.* 2024 May 19
- Watts LM et al. The phenotype of MEGF8-related Carpenter syndrome (CRPT2) is refined through the identification of eight new patients. *Eur J Hum Genet.* 2024 Jul;32(7):864-870.
- Chaussenot A, et al. Loss of heterozygosity in CCM2 cDNA revealing a structural variant causing multiple cerebral cavernous malformations. *Eur J Hum Genet.* 2024 Jul;32(7):876-878.
- Nguyen A et al. Late-onset refractory hemolytic anemia in siblings treated for methionine synthase reductase deficiency: A rare complication possibly prevented by hydroxocobalamin dose escalation? *JIMD Rep.* 2024 Apr15;65(3):163-170.
- Legrand C et al. Germline POT1 mutation and neuroblastoma: a mere coincidence or true association. *Pediatr Blood Cancer.* 2024 Jul;71(7):e31054.
- Layo-Carris DE et al. Expanded phenotypic spectrum of neurodevelopmental and neurodegenerative disorder Bryant-Li-Bhoj syndrome with 38 additional individuals. *Eur J Hum Genet.* 2024 Aug;32(8):928-937. doi:10.1038/s41431-024-01610-1.
- Billon C et al. Genome-wide analysis identifies MYH11 compound heterozygous variants leading to visceral myopathy corresponding to late-onset form of megacystis-microcolon-intestinal hypoperistalsis syndrome. *Mol Genet Genomics.* 2024 Apr 16;299(1):44.
- Dohrn MF et al. Recurrent ATP1A1 variant Gly903Arg causes developmental delay, intellectual disability, and autism. *Ann Clin Transl Neurol.* 2024 Apr;11(4):1075-1079.
- Plaisancié J et al. Structural variant disrupting the expression of the remote FOXC Gene in a Patient with Syndromic Complex Microphthalmia. *Int J Mol Sci.* 2024 Feb 25;25(5):2669.
- Previdi A et al. Novel variant in LRP6 associated with unusual and severe clinical presentation: Case report. *Clin Genet.* 2024 Jun;105(6):666-670. Szot JO et al. A metabolic signature for NADSYN1-dependent congenital NAD deficiency disorder. *J Clin Invest.* 2024 Feb15;134(4):e174824.
- Tusseau M et al. Genome sequencing identify chromosome 9 inversions disrupting ENG in 2 unrelated HHT families. *Eur J Med Genet.* 2024 Apr;68:104919

- Nicolle R et al. Expanding the phenotypic spectrum of LIG4 pathogenic variations: neuro-histopathological description of 4 fetuses with stenosis of the aqueduct. *Eur J Hum Genet.* 2024 May;32(5):545-549.
- Papadopoulos T et al. New description of an MRPS2 homozygous patient: Further features to help expand the phenotype. *Eur J Med Genet.* 2024 Feb;67:104889.
- Durin Z et al. Efficacy of oral manganese and D-galactose therapy in a patient bearing a novel TMEM165 variant. *Transl Res.* 2024 Apr;266:57-67.
- Li D et al. Spliceosome malfunction causes neurodevelopmental disorders with overlapping features. *J Clin Invest.* 2024 Jan 2;134(1):e171235.
- Sabbagh Q et al. Clinico-biological refinement of BCL11B-related disorder and identification of an epismutation: A series of 20 unreported individuals. *Genet Med.* 2024 Jan;26(1):101007.
- Chevrollier A et al. Homozygous MFN2 variants causing severe antenatal encephalopathy with clumped mitochondria. *Brain.* 2024 Jan 4;147(1):91-99.
- Sperelakis-Beedham B et al. Expanding the phenotype of GTF2E2-associated trichothiodystrophy. *J Eur Acad Dermatol Venereol.* 2024 Mar;38(3):e222-e226.
- Poggio E et al. ATP2B2 de novo variants as a cause of variable neurodevelopmental disorders that feature dystonia, ataxia, intellectual disability, behavioral symptoms, and seizures. *Genet Med.* 2023 Dec;25(12):100971.
- Limousin W et al. Molecular-based targeted therapies in patients with hepatocellular carcinoma and hepato cholangiocarcinoma refractory to atezolizumab/bevacizumab. *J Hepatol.* 2023 Dec;79(6):1450-1458.
- Parra A et al. Clinical heterogeneity and different phenotypes in patients with SETD2 variants: 18 new patients and review of the literature. *Genes (Basel).* 2023 May 29;14(6):1179.
- Rive Le Gouard N et al. First reports of fetal SMARCC1 related hydrocephalus. *Eur J Med Genet.* 2023 Aug;66(8):104797.
- Duployez N et al. UBTF tandem duplications define a distinct subtype of adult de novo acute myeloid leukemia. *Leukemia.* 2023 Jun;37(6):1245-1253
- Frost FG et al. Bi-allelic SNAPC4 variants dysregulate global alternative splicing and lead to neuroregression and progressive spastic paraparesis. *Am J Hum Genet.* 2023 Apr;110(4):663-680.
- Aubert-Mucca M et al. Clinical heterogeneity of NADSYN1-associated VCRL syndrome. *Clin Genet.* 2023 Jul;104(1):114-120.
- Hamdan D et al. Olaparib in the setting of radiotherapy-associated sarcoma: what can precision medicine offer for rare cancers? *JCO Precis Oncol.* 2023 Feb;7:e2200582
- Khatri D et al. Deficiency of the minor spliceosome component U4atac snRNA secondarily results in ciliary defects in human and zebrafish. *Proc Natl Acad Sci U S A.* 2023 Feb 28;120(9):e2102569120.
- Evin C et al. Adenoid cystic carcinoma of Bartholin's gland, a case report with genomic data and literature review. *Cancer Radiother.* 2023 Jun;27(4):328-336.
- Dirix M et al. Overcoming the challenges associated with identification of deep intronic variants by whole genome sequencing. *Clin Genet.* 2023 Jun;103(6):693-698.

- Ravindran E et al. Monoallelic CRMP1 gene variants cause neurodevelopmental disorder. *Elife*. 2022 Dec 13;11:e80793.
- Reis LM et al. ARHGAP35 is a novel factor disrupted in human developmental eye phenotypes. *Eur J Hum Genet*. 2023 Mar;31(3):363-367.
- Happ HC et al. Neurodevelopmental and epilepsy phenotypes in individuals with missense variants in the voltage sensing and pore domains of KCNH5. *Neurology*. 2023 Feb 7;100(6):e603-e615.
- Pacot L et al. Contribution of whole genome sequencing in the molecular diagnosis of mosaic partial deletion of the NF1 gene in neurofibromatosis type 1. *Hum Genet*. 2023 Jan;142(1):1-9.
- Riou MC et al. Oral phenotype of Singleton-Merten syndrome: a systematic review illustrated with a case report. *Front Genet*. 2022 Jun 9;13:875490.
- Guerrini R et al. Phenotypic and genetic spectrum of ATP6V1A encephalopathy: a disorder of lysosomal homeostasis. *Brain*. 2022 Aug 27;145(8):2687-2703.
- Lacombe D et al. Hemidystonia with polymicrogyria is part of ATP1A3-related disorders. *Brain Dev*. 2022 Sep;44(8):567-570.
- Christensen MB et al. Biallelic variants in ZNF142 lead to a syndromic neurodevelopmental disorder. *Clin Genet*. 2022 Aug;102(2):98-109.
- Sabbagh Q et al. A second individual with rhizomelic spondyloepimetaphyseal dysplasia and homozygous variant in GNPAT1. *Eur J Med Genet*. 2022 Jun;65(6):104495.
- Rajan DS et al. Autosomal recessive cerebellar atrophy and spastic ataxia in patients with pathogenic biallelic variants in GEMIN5. *Front Cell Dev Biol*. 2022 Feb 28;10:783762.
- Münch J et al. Biallelic pathogenic variants in roundabout guidance receptor 1 associate with syndromic congenital anomalies of the kidney and urinary tract. *Kidney Int*. 2022 May;101(5):1039-1053.
- Kumble S et al. The clinical and molecular spectrum of QRIC1 associated neurodevelopmental disorder. *Hum Mutat*. 2022 Feb;43(2):266-282.
- Rucheton B et al. Adult cerebellar ataxia, axonal neuropathy, and sensory impairments caused by biallelic SCO2 variants. *Neurol Genet*. 2021 Nov 3;7(6):e630.
- Zech M et al. Biallelic AOPEP loss-of-function variants cause progressive dystonia with prominent limb involvement. *Mov Disord*. 2022 Jan;37(1):137-147.
- von der Lippe C et al. Heterozygous variants in ZBTB7A cause a neurodevelopmental disorder associated with symptomatic overgrowth of pharyngeal lymphoid tissue, macrocephaly, and elevated fetal hemoglobin. *Am J Med Genet A*. 2022 Jan;188(1):272-282.
- Lunati A et al. VPS4A mutation in syndromic congenital hemolytic anemia without obvious signs of dyserythropoiesis. *Am J Hematol*. 2021 Apr1;96(4):E121-E123.