Publikationsverzeichnis
Dr. rer. nat. Anna Benet-Pagès

MGZ
Medizinisch Genetisches Zentrum

1.

Abicht A, Scharf F, Kleinle S, et al. Mitochondrial and nuclear disease panel (Mito-aND-Panel): Combined sequencing of mitochondrial and nuclear DNA by a cost-effective and sensitive NGS-based method. *Mol Genet Genomic Med*. 2018;6(6):1188-1198. doi:10.1002/mgg3.500

2.

Morak M, Kasbauer S, Kerscher M, et al. Loss of MSH2 and MSH6 due to heterozygous germline defects in MSH3 and MSH6. *Fam Cancer*. 2017;16(4):491-500. doi:10.1007/s10689-017-9975-z

3.

Rump A, Benet-Pages A, Schubert S, et al. Identification and Functional Testing of ERCC2 Mutations in a Multi-national Cohort of Patients with Familial Breast- and Ovarian Cancer. *PLoS Genet*. 2016;12(8):e1006248. doi:10.1371/journal.pgen.1006248

4.

Di Donato N, Neuhann T, Kahlert A-K, et al. Mutations in EXOSC2 are associated with a novel syndrome characterised by retinitis pigmentosa, progressive hearing loss, premature ageing, short stature, mild intellectual disability and distinctive gestalt. *J Med Genet*. 2016;53(6):419-425. doi:10.1136/jmedgenet-2015-103511

5.

Kohler C, Heyer C, Hoffjan S, et al. Early-onset leukoencephalopathy due to a homozygous missense mutation in the DARS2 gene. *Mol Cell Probes*. 2015;29(5):319-322. doi:10.1016/j.mcp.2015.06.005

6.

Anna Benet-Pagès Ina Vogl, et al. Application and data analysis of next-generation sequencing. *LaboratoriumsMedizin*. 2013;37:305-315.

7.

Jansen S, Aigner B, Benet-Pagès A, et al. Assessment of the genomic variation in a cattle population by re-sequencing of key animals at low to medium coverage. *BMC Genomics*. 2013;14:446.

8.

Stogmann E, Reinthaler E, Eltawil S, et al. Autosomal recessive cortical myoclonic tremor and epilepsy: association with a mutation in the potassium channel associated gene CNTN2. *Brain*. 2013;136(Pt 4):1155-1160. doi:10.1093/brain/awt068

9.

Sabrautzki S, Rubio-Aliaga I, Benet-Pagès A, et al. New mouse models for metabolic bone diseases generated by genome-wide ENU mutagenesis. *Mamm Genome*. 2012:648.

10.

Ina Vogl, Anna Benet-Pagès, Sebastian H. Eck,, et al. Diagnostic applications of next generation sequencing: working towards quality standards. *J Lab Med*. 2012:227-239.

11.

Zimprich A, Benet-Pages A, Struhal W, et al. A mutation in VPS35, encoding a subunit of the retromer complex, causes late-onset Parkinson disease. *Am J Hum Genet*. 2011;89(1):168-175. doi:10.1016/j.ajhg.2011.06.008

12.

Greif PA, Eck SH, Konstandin NP, et al. Identification of recurring tumor-specific somatic mutations in acute myeloid leukemia by transcriptome sequencing. *Leukemia*. 2011;25(5):821-827. doi:10.1038/leu.2011.19

13.

Bergbauer M, Kalla M, Schmeinck A, et al. CpG-methylation regulates a class of Epstein-Barr virus promoters. *PLoS Pathog*. 2010;6(9):e1001114. doi:10.1371/journal.ppat.1001114

14.

Lorenz-Depiereux B, Bastepe M, Benet-Pages A, et al. DMP1 mutations in autosomal recessive hypophosphatemia implicate a bone matrix protein in the regulation of phosphate homeostasis. *Nat Genet*. 2006;38(11):1248-1250. doi:10.1038/ng1868

15.

Kato K, Jeanneau C, Tarp MA, et al. Polypeptide GalNAc-transferase T3 and familial tumoral calcinosis. Secretion of fibroblast growth factor 23 requires O-glycosylation. *J Biol Chem*. 2006;281(27):18370-18377. doi:10.1074/jbc.M602469200

16.

Lorenz-Depiereux B, Benet-Pages A, Eckstein G, et al. Hereditary hypophosphatemic rickets with hypercalciuria is caused by mutations in the sodium-phosphate cotransporter gene SLC34A3. *Am J Hum Genet*. 2006;78(2):193-201. doi:10.1086/499410

17.

Benet-Pages A, Orlik P, Strom TM, Lorenz-Depiereux B. An FGF23 missense mutation causes familial tumoral calcinosis with hyperphosphatemia. *Hum Mol Genet*. 2005;14(3):385-390. doi:10.1093/hmg/ddi034

18.

Lorenz-Depiereux B. B-PA, Strom T. Molekulargenetik der hypophosphatämischen Rachitis. In: *Hyperphosphatämische Rachitis. Diagnose Und Betreuung von Kindern Mit Spezifischen Kleinwuchsformen.* Berlin: AWB-Verlag; 2005.

19.

Giehl KA, Eckstein GN, Benet-Pages A, et al. A gene locus responsible for the familial hair shaft abnormality pili annulati maps to chromosome 12q24.32-24.33. *J Invest Dermatol*. 2004;123(6):1073-1077. doi:10.1111/j.0022-202X.2004.23423.x

20.

Benet-Pages A, Lorenz-Depiereux B, Zischka H, White KE, Econs MJ, Strom TM. FGF23 is processed by proprotein convertases but not by PHEX. *Bone*. 2004;35(2):455-462. doi:10.1016/j.bone.2004.04.002

21.

White KE, Carn G, Lorenz-Depiereux B, Benet-Pages A, Strom TM, Econs MJ. Autosomal-dominant hypophosphatemic rickets (ADHR) mutations stabilize FGF-23. *Kidney Int*. 2001;60(6):2079-2086. doi:10.1046/j.1523-1755.2001.00064.x