

**Cohen, L.**, Assady, S., Zaid, A. The Kidney as an Achilles heel in the Use of Cyclooxygenase-2 selective Inhibitors (COXIBs). Israeli Bilingual Journal of Hypertension 2004, 5, 4-13.

**Cohen L**, Salzberg A. Identifying direct target genes of the homeotic co- factor Homothorax. Mol Genet Genomics. 2008 May 15.

**Cohen L**, Tzur S, Goldenberg-Cohen N, Bormans C, Behar DM, Reinstein E. Exome sequencing identified a novel de novo OPA1 mutation in a consanguineous family presenting with optic atrophy. Genet Res (Camb). 2016 Jun 6;98:e10.

**Cohen L**, Orenstein N, Weisz-Hubshman M, Bazak L, Davidov B, Reinstein E, Tzur S, Behar D, Smirin-Yosef P, Salmon-Divon M, Gross A, Shohat M, Basel-Vanagaite L. Utilization of whole exome sequencing in diagnostics of genetic disease:Rabin medical centre's experience. Harefuah. 2017 Apr;156(4):212-216.

Weiss K , Terhal P, **Cohen L** , Brucolieri M, Irving M, Martinez AF, Rosenfeld JA, Machol K , Yang Y, Liu P, Walkiewicz M, Beuten J, Gomez-Ospina N, Haude K, Fong CT, M. Enns G, Bernstein JA, Fan J, Gotway G, Ghorbani M, DDD study, Gassen KV, Monroe G, Haaften GB, Basel-Vanagaite L, Yang XJ, Campeau PM, Muenke M. *De Novo* Mutations in *CHD4*, an ATP-Dependent Chromatin Remodeler Gene, Cause an Intellectual Disability Syndrome with Distinctive Dysmorphisms. AJMG 2016 [Accepted]

Masotti A, Uva P, Davis-Keppen L, Basel-Vanagaite L, **Cohen L**, Pisaneschi E, Celluzzi A, Ben-civenga P, Fang M, Tian M, Xu X, Cappa M, Dallapiccola B.. Keppen-Lubinsky syndrome is caused by mutations in the inwardly-rectifying K<sup>+</sup> channel encoded by KCNJ6. American Journal of Human Genetics Feb 2015.

Reinstein E, Orvin K, Tayeb-Fligelman E, Stiebel-Kalish H, Tzur S, Pimienta AL, Bazak L, Bengal T, **Cohen L**, Gaton DD, Bormans C, Landau M, Kornowski R, Shohat M, Behar DM. Mutations in *TAX3* cause Dilated Cardiomyopathy with Septo-Optic Dysplasia Hum Mutat. 2015 Jan 23.

Kurolap A, Orenstein N, Kedar I, Weisz Hubshman M, Tiosano D, Mory A, Levi Z, Marom D, **Cohen L**, Ekhilevich N, Douglas J, Nowak CB, Tan WH, Baris HN. Is one diagnosis the whole story? Patients with double diagnoses. Am J Med Genet A. 2016 Sep;170(9):2338-48

Smirin-Yosef P, Zuckerman-Levin N, Tzur S, Granot Y, **Cohen L**, Sachsenweger J, Borck G, Lagovsky I, Salmon-Divon M, Wiesmüller L, Basel-Vanagaite. A biallelic mutation in the homologous recombination repair gene SPIDR is associated with human gonadal dysgenesis. *J Clin Endocrinol Metab.* 2016 Dec 14.

Sharony R, Borochowitz Z, **Cohen L**, Storch A, Rosenfeld R, Modai S, Reinstein E. Prenatal course of metaphyseal anadysplasia associated with homozygous mutation in MMP9 identified by exome sequencing. *Clin Genet.* 2017 Mar

Straussberg R, Onoufriadiis A, Konen O , Zouabi Y, **Cohen L**, John Y. W. Lee, Chao-Kai Hsu, Michael A. Simpso, John A. McGrath. Novel homozygous missense mutation in *NT5C2* underlying hereditary spastic paraplegia SPG45. *Am J Med Genet A.* 2017 Nov

Montagne L, Derhourhi M, Piton A, Toussaint B, Durand E, Vaillant E, Thuillier D, Gaget S, De Graeve F, Rabearivelo I, Lansiaux A, Lenne B, Sukno S, Desailloud R, Cnop M, Niculescu R, **Cohen L**, Zagury JF, Amouyal M, Weill J, Muller J, Sand O, Delobel B, Froguel P, Bonnefond A. CoDE-seq, an augmented whole-exome sequencing, enables the accurate detection of CNVs and mutations in Mendelian obesity and intellectual disability. *Mol Metab.* 2018 May 16.

Shohet A, **Cohen L**, Haguel D, Mozer Y, Shomron N, Tzur S, Bazak L, Basel Salmon L, Krause I. Variant in SCYL1 gene causes aberrant splicing in a family with cerebellar ataxia, recurrent episodes of liver failure, and growth retardation. *Eur J Hum Genet.* 2018 Sep 26