

Selected peer-reviewed publications:

1. Luder A, Mandel H, Khayat M, Gurevich I, Frankel P, Rivlin J, **Falik-Zaccai TC**. Chronic lung disease and cystic fibrosis phenotype in prolidase deficiency: a newly recognized association: *J Pediatr*. 2007; 150: 656-658.
2. **Falik-Zaccai TC**, Kfir N, Frankel P, Cohen C, Tanus M, Mandel H, Shihab S, Morkos S, Aaref S, Summar ML, Khayat M. Population screening in a Druze community: the challenge and the reward: *Genet Med*. 2008; 10(12): 903-909.
3. **Falik-Zaccai TC**, Laskar M, Kfir N, Nasser W, Slor H, Khayat M. Cockayne syndrome type II in a Druze isolate in northern Israel in association with an insertion mutation in ERCC6: *Am J Med Genet*. 2008; 146 A(11): 1423-1429.
4. Khayat M, Korman SH, Frankel P, Weintraub Z, Hershckowitz S, Flaisher Sheffer V, Ben Elisha M, Wevers RA, **Falik-Zaccai TC**. PNPO deficiency: an under diagnosed inborn error of pyridoxine metabolism: *Mol. Genet Metab*. 2008; 94(4): 431-434.
5. Keren Z, **Falik-Zaccai TC**. Cerebrotendinous xanthomatosis (CTX): a treatable lipid storage disease: *Pediatr Endocrinol Rev*. 2009; 7: 6-11.
6. Berginer VM, Gross B, Morad K, Kfir N, Morkos S, Aaref S, **Falik-Zaccai TC**. Chronic Diarrhea and Juvenile Cataracts: Think Cerebrotendinous Xanthomatosis and Treat: *Pediatrics*. 2009; 123(1): 143-147.
7. **Falik-Zaccai TC**, Keren Z, Slor H. The Versatile DNA Nucleotide Excision Repair (NER) and its Medical Significance: *Pediatr Endocrinol Rev*. 2009; 7:117-122.
8. Suriu C, Khayat M, Weiler M, Kfir N, Cohen C, Zinger A, Aslanidis C, Schmitz G, **Falik-Zaccai TC**. Skoura-a genetic island for congenital insensitivity to pain and anhidrosis among Moroccan Jews, as determined by a novel mutation in the NTRK1 Gene: *Clinical Genet*. 2009; 75 (3): 230-236.
9. Klar A, Navon-Elkan P, Rubinow A, Branski D, Hurvitz H, Christensen E, Khayat M, **Falik-Zaccai TC**. Prolidase deficiency: it looks like systemic lupus erythematosus but it is not: *Eur J Pediatr*. 2010; 169(6): 727-732.
10. **Falik-Zaccai TC**, Khayat M, Luder A, Frenkel P, Magen D, Brik R, Gershoni Baruch R, Mandel H. A broad spectrum of developmental delay in a large cohort of prolidase deficiency patients demonstrates marked interfamilial and

intrafamilial phenotypic variability: *Am J Med. Genet. B. Neuropsychiatr Genet.* 2010; 153B(1): 46-56.

11. Genzer-Nir M, Khayat M, Kogan L, Cohen HI, Hershkowitz M, Geiger D, **Falik-Zaccai TC**. Mammary-digital-nail (MDN) syndrome: a novel phenotype maps to human chromosome 22q12.3-13.1: *Eur J Hum Genet.* 2010 Jun ;18(6): 662-7.
12. Gunay- Aygun M, Zivony-Elboun Y, Gumruk F, Geiger D, Cetin M, Khayat M, Kleta R, Kfir N, Anikster Y, Chezar J, Arcos-Bugos M, Shalata A, Stanescu H, Manaster J, Arat M, Edwards H, Freiberg AS, Hart PS, Riney LC, Patzel K, Tanpaiboon P, Markello T, Huizing M, Maric I, Horne M, Kehrel BE0, Jurk K0, Hansen NF, Cherukuri PF, Jones M, Cruz P, Mullikin JC, Nurden A, White JG, Gahl WA, **Falik-Zaccai TC**. Gray Platelet Syndrome: Natural history of a large patient cohort and locus assignment to chromosome 3p: *Blood.* 2010 Dec 2;116(23):4990-5001.
13. Gunay- Aygun M*, **Falik-Zaccai TC***, Vilboux T, Zivony-Elboun Y, Gumruk F, Cetin M, Khayat M, Boerkoel CF, Kfir N, Huang Y, Maynard D, Dorward H, Berger K, Kleta R, Anikster Y, Arat M, Freiberg AS, Kehrel BE, Jurk K, Cruz P, Mullikin JC, White JG, Huizing M, Gahl WA. *NBEAL2* is mutated in Gray Platelet Syndrome and required for biogenesis of platelet alpha-granules. *Nat Genet.* 2011 Jul 17;43(8):732-4. ***These authors contributed equally.**
14. Zivony-Elboun Y, Westbroek W, Kfir N, Savitzki D, Shoal Y, Bloom A, Rod R, Khayat M, Gross B, Samri W, Cohen H, Sonkin V, Freidman T, Geiger D, Fattal-Valevski A, Anikster Y, Waters AM, Kleta R, **Falik-Zaccai TC**. [A founder mutation in Vps37A causes autosomal recessive complex hereditary spastic paraparesis.](#) *J Med Genet.* 2012 Jul;49(7):462-72
15. **Falik-Zaccai TC**, Erel-Segal R, Horev L, Bitterman-Deutsch O, Koka S, Chaim S, Keren Z, Kalfon L, Gross B, Segal Z, Orgal S, Shoal Y, Slor H, Spivak G, Hanawalt PC. [A novel XPD mutation in a compound heterozygote; the mutation in the second allele is present in three homozygous patients with mild sun sensitivity.](#) *Environ Mol Mutagen.* 2012 Aug;53(7):505-14