Four novel p.N385K, p.V36A, c.1033-1034insT and c.1417-1418delCT mutations in the sphingomyelin Phosphodiesterase 1 (SMPD1) gene in patients with types A and B Niemann-Pick disease (NPD)

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#	Paper	IF	Citations
11	Alleged Detrimental Mutations in the SMPD1 Gene in Patients with Niemann-Pick Disease. *International Journal of Molecular Sciences*, **2015**, 16, 13649-52	6.3	8
10	SMPD1 Mutation Update: Database and Comprehensive Analysis of Published and Novel Variants. *Human Mutation*, **2016**, 37, 139-47	4.7	44
9	Regulation of sphingomyelin metabolism. *Pharmacological Reports*, **2016**, 68, 570-81	3.9	85
8	SMPD1 variants in Chinese Han patients with sporadic Parkinson's disease. *Parkinsonism and Related Disorders*, **2017**, 34, 59-61	3.6	20
7	Deep sequencing of SMPD1 gene revealed a heterozygous frameshift mutation (p.Ser192Alafs) in a Palestinian infant with Niemann-Pick disease type A: a case report. *Journal of Medical Case Reports*, **2018**, 12, 272	1.2	2
6	Multimodal imaging including optical coherence tomography angiography in patients with type B Niemann-Pick disease. *International Ophthalmology*, **2019**, 39, 2545-2552	2.2	3
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3	Characterization of Niemann-Pick diseases genes mutation spectrum in Iran and identification of a novel mutation in gene. *Medical Journal of the Islamic Republic of Iran*, **2019**, 33, 126	1.1	
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1	SMPD1 gene variants in patients with Thalassemia major. **2023**, 50, 3355-3363		0