

- 1: Taha I, Foroni S, Valli R, Frattini A, Rocchia P, Porta G, Zecca M, Bergami E, Cipolli M, Pasquali F, Danesino C, Scotti C, Minelli A. Case Report: Heterozygous Germline Variant in EIF6 Additional to Biallelic SBDS Pathogenic Variants in a Patient With Ribosomopathy Shwachman-Diamond Syndrome. *Front Genet.* 2022 Aug 12;13:896749. doi: 10.3389/fgene.2022.896749. PMID: 36035165; PMCID: PMC9411639.
- 2: Pérez-Juárez J, Tapia-Vieyra JV, Gutiérrez-Magdaleno G, Sánchez-Puig N. Altered Conformational Landscape upon Sensing Guanine Nucleotides in a Disease Mutant of Elongation Factor-like 1 (EFL1) GTPase. *Biomolecules.* 2022 Aug 19;12(8):1141. doi: 10.3390/biom12081141. PMID: 36009035; PMCID: PMC9405973.
- 3: Tang Q, Ye XM, Yang YC, Wen XL. Shwachman-Diamond syndrome: A case report. *Asian J Surg.* 2022 Aug 10;S1015-9584(22)01007-7. doi: 10.1016/j.asjsur.2022.07.061. Epub ahead of print. PMID: 35963699.
- 4: Scheers I, Berardis S. Congenital etiologies of exocrine pancreatic insufficiency. *Front Pediatr.* 2022 Jul 22;10:909925. doi: 10.3389/fped.2022.909925. PMID: 35935370; PMCID: PMC9354839.
- 5: Taha I, De Paoli F, Foroni S, Zucca S, Limongelli I, Cipolli M, Danesino C, Ramenghi U, Minelli A. Phenotypic Variation in Two Siblings Affected with Shwachman-Diamond Syndrome: The Use of Expert Variant Interpreter (eVai) Suggests Clinical Relevance of a Variant in the KMT2A Gene. *Genes (Basel).* 2022 Jul 23;13(8):1314. doi: 10.3390/genes13081314. PMID: 35893049; PMCID: PMC9394309.
- 6: Spinetti E, Delre P, Saviano M, Siliqi D, Lattanzi G, Mangiatordi GF. A Comparative Molecular Dynamics Study of Selected Point Mutations in the Shwachman-Bodian-Diamond Syndrome Protein SBDS. *Int J Mol Sci.* 2022 Jul 19;23(14):7938. doi: 10.3390/ijms23147938. PMID: 35887285; PMCID: PMC9320453.
- 7: Cesaro S, Donadieu J, Cipolli M, Dalle JH, Styczynski J, Masetti R, Strahm B, Mauro M, Alseraihy A, Aljurf M, Dufour C, de la Tour RP. Stem Cell Transplantation in Patients Affected by Shwachman-Diamond Syndrome: Expert Consensus and Recommendations From the EBMT Severe Aplastic Anaemia Working Party. *Transplant Cell Ther.* 2022 Jul 20;S2666-6367(22)01472-5. doi: 10.1016/j.jtct.2022.07.010. Epub ahead of print. PMID: 35870777.
- 8: Farooqui SM, Ward R, Aziz M. Shwachman-Diamond Syndrome. 2022 Jul 19. In: *StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2022 Jan-. PMID: 29939643.*
- 9: Moore CA, Krishnan K. Bone Marrow Failure. 2022 Jul 11. In: *StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2022 Jan-. PMID: 29083589.*
- 10: Frontiers Production Office. Erratum: Novel Biallelic Variants in *DNAJC21* Causing an Inherited Bone Marrow Failure Spectrum Phenotype: An Odyssey to Diagnosis. *Front Genet.* 2022 May 25;13:930132. doi: 10.3389/fgene.2022.930132. Erratum for: *Front Genet.* 2022 Apr 08;13:870233. PMID: 35692840; PMCID: PMC9174891.