

DAFTAR PUSTAKA

- Akter, U.T., Sogir, S.B., Basak, T. 2023. On the Basics of *Pedigree* Visualization and Feature Extraction for the Autosomal Recessive Inheritance Pattern. *American Journal of Laboratory Medicine*. 8(1): 4-12.
- Aldalaqan, S., Dalglish, C., Luzzi, S., Siachisumo, C., Reynard, L. N., Ehrmann, I., and Elliott, D. J. 2022. Cryptic Splicing: Common Pathological Mechanisms Involved in Male Infertility and Neuronal Diseases. *Cell Cycle*. 21(3):219-227.
- Aledort, L., Mannucci, P. M., Schramm, W., and Tarantino, M. 2019. Factor VIII Replacement is still The Standard of Care in Haemophilia A. *Blood Transfusion*. 17(6):479-486.
- Alkarrash, M. S., Badawi, R., Sallah, H., Shashaa, M. N., Argilo, J., and Alkhoury, R. 2021. Hemophilia A and C in a Female: The First Case Report in Literature. *Annals of Medicine and Surgery*. 68:1-3.
- Anna, A and Monika G. 2018. Splicing Mutations in Human Genetic Disorders: Examples, Detection, and Confirmation. *J Appl Genet*. 59(3):253-268.
- Baig, M. A., and Swamy, K. B. 2021. Comparative Analysis of Chromogenic vs Clot Based One Stage APTT Assay for Determination of Factor VIII Level. *Indian Journal of Pathology & Microbiology*. 64(1):123-127.
- Balestra D, Maestri I, Branchini A, Ferrarese M, Bernardi F, and Pinotti M. 2019. An Altered Splicing Registry Explains the Differential ExSpeU1-Mediated Rescue of Splicing Mutations Causing Haemophilia A. *Front Genet*. 10:1-9.
- Barralle F.E., Singh R.N. and Stamm S. 2019. RNA Structure and Splicing Regulation. *Biochim Biophys Acta Gene Regul Mech*. 1862:11-12.
- Berntorp, E., Fischer, K., Hart, D.P. (2021). Haemophilia. *Nature Reviews Disease Primers*. 7(45):1-19.
- Bogdanova, N., Markoff, A., Eisert, R., Wermes, C., Pollmann, H., Todorova, A., Chlystun, M., Nowak-Göttl, U., and Horst, J. 2007. Spectrum of Molecular Defects and Mutation Detection Rate in Patients with Mild and Moderate Hemophilia A. *Human Mutation*. 28(1):54-60.

- Bordbar, M., Beigipour, R., Tahami, M., Zekavat, O. R., Haghpanah, S., and Moshfeghinia, R. 2023. Skeletal Complications in Patients with Hemophilia: A single-center Experience. *Journal of Orthopaedic Surgery and Research*. 18(907):1-6.
- Brummel-Ziedins, K. E., Whelihan, M. F., Gissel, M., Mann, K. G., and Rivard, G. E. 2009. Thrombin Generation and Bleeding in Haemophilia A. *Haemophilia*. 15(5):1118-1125.
- Cafe, A., Carvalho, M., and Crato, M. 2019. Haemophilia A: Health and Economic Burden of a Rare Disease in Portugal. *Orphanet Journal of Rare Diseases*. 14(211):1-11.
- Caminsky N., Mucaki E.J., and Rogan P.K. 2014. Interpretation of mRNA Splicing Mutations in Genetic Disease: Review of The Literature and Guidelines for Information-Theoretical Analysis. *F1000 Research*. 3(282):1-30.
- Castaman, G. and Matino, D. 2019. Hemophilia A and B: Molecular and Clinical Similarities and Differences. *Haematologica*. 104(9):1702-1709.
- Castaman, G., Giacomelli, S. H., Mancuso, M. E., Sanna, S., Santagostino, E., and Rodeghiero, F. 2010. F8 mRNA Studies in Haemophilia A Patients with different Splice Site Mutations. *Haemophilia*. 16(5):786-790.
- Choi, S. J., Jang, K. J., Lim, J. A., and Kim, H. S. 2015. Human Coagulation Factor VIII Domain Specific Recombinant Polypeptide Expression. *Blood Research*. 50(2):103-108.
- Cutler, J. A., Mitchell, M. J., Smith, M. P., and Savidge, G. F. 2002. The Identification and Classification of 41 Novel Mutations in The Factor VIII Gene (F8C). *Human Mutation*. 19(3):274-278.
- Dardik, R., Janczar, S., Lalezari, S., Avishai, E., Levy-Mendelovich, S., Barg, A. A., Martinowitz, U., Babol-Pokora, K., Mlynarski, W., and Kenet, G. 2023. Four Decades of Carrier Detection and Prenatal Diagnosis in Hemophilia A: Historical Overview, State of the Art and Future Directions. *International Journal of Molecular Sciences*. 24(14):1-13.
- Doncel. S. S., Mosquera, D. G. A., Cortes, J. M., Rico, A. C., Cadavid, M. F. J., and Pelaez. R. G. 2023. Haemophilia A: A Review of Clinical Manifestations Treatment Mutations and The Development of Inhibitors. *Hematology Reports*. 15(1):130-150.

- Elmahmoudi, H., Ben-lakhal, F., and Elborji, W. 2012. Identification of Genetic Defects Underlying FVII Deficiency in 10 Patients Belonging to Eight Unrelated Families of the North Provinces from Tunisia. *Diagnostic Pathology*. 7(92):1-5.
- Fan, G., Shen, Y., Cai, Y., Zhao, J. H., and Wu, Y. 2022. Uncontrollable Bleeding after Tooth Extraction from Asymptomatic Mild Hemophilia Patients: Two Case Reports. *BMC Oral Health*. 22(69):1-8.
- Fang, H., Wang, L., and Wang, H. 2007. The Protein Structure and Effect of Factor VIII. *Thrombosis Research*. 119:1-13.
- Feng, Y., Li, Q., Shi, P., Liu, N., Kong, X., and Guo, R. 2021. Mutation Analysis in the F8 Gene in 485 Families with Haemophilia A and Prenatal Diagnosis in China. *Haemophilia*. 27(1):88-92.
- Ferreira, C.N., Sousa, M.D., Dusse, L.M., and Carvalho, M.D. 2010. A Cell-Based Model of Coagulation and Its Implications. *Revista Brasileira De Hematologia e Hemoterapia*. 32 (5):416-421.
- Franchini, M. and Mannucci, P. M. 2013. Hemophilia A in The Third Millennium. *Blood Reviews*. 27(4):179-184.
- Garibyan, L., & Avashia, N. 2013. Polymerase Chain Reaction. *The Journal of Investigative Dermatology*. 133(3):1-4.
- Goodeve A. C. 2015. Hemophilia B: Molecular Pathogenesis and Mutation Analysis. *Journal of Thrombosis and Haemostasis*. 13(7):1184-1195.
- Gooding, R., Thachil, J., Alamelu, J., Motwani, J., and Chowdary, P. 2021. Asymptomatic Joint Bleeding and Joint Health in Hemophilia: A Review of Variables Methods and Biomarkers. *Journal of Blood Medicine*. 12:209-220.
- Graw, J., Brackmann, H. H., Oldenburg, J., Schneppenheim, R., Spannagl, M., and Schwaab, R. 2005. Haemophilia A: from Mutation Analysis to new Therapies. *Nature Reviews Genetics*. 6(6):488-501.
- Guo, Z., Yang, L., Qin, X., Liu, X., & Zhang, Y. 2018. Spectrum of Molecular Defects in 216 Chinese Families with Hemophilia A: Identification of non Inversion Mutation Hot Spots and 42 Novel Mutations. *Journal of the International Academy of Clinical and Applied Thrombosis*. 24(1):70-78.

- Inaba, H., Shinozawa, K., Seita, I., Otaki, M., Suzuki, T., Hagiwara, T., Amano, K., and Fukutake, K. 2013. Genotypic and Phenotypic Features of Japanese Patients with Mild to Moderate Hemophilia A. *International Journal of Hematology*. 97(6):758–764.
- Jackson I.J. 1991. A Reappraisal of non-consensus mRNA Splice Sites. *Nucleic Acids Res.* 19(14):3795-8.
- Jaoudeh, M. B. 2023. Eradication of FVIII Inhibitors in Patients with Hemophilia A. *Disertasi*. Paris: Sorbonne University.
- Jayakrishnan, T., Shah, D., and Mewawalla, P. 2019. Hemophilia C: A Case Report with Updates on Diagnosis and Management of a Rare Bleeding Disorder. *Journal of Hematology*. 8(3):144-147.
- Kapustin, Y., Chan, E., Sarkar, R., Wong, F., Vorechovsky, I., Winston, R.M., Tatusova, T., and Dibb N.J. 2011. Cryptic Splice Sites and Split Genes. *Nucleic Acids Res.* 39(14):5837-44.
- Kawashima, S., Hattori, A., and Suzuki, E. 2021. Methylation Status of Genes Escaping from X-Chromosome Inactivation in Patients with X-Chromosome Rearrangements. *Clin Epigenet.* 13 (134):1-11
- Khare, P., Raj, V., Chandra, S., and Agarwal, S. 2014. Quantitative and Qualitative Assessment of DNA Extracted from Saliva for Its Use in Forensic Identification. *Journal of Forensic Dental Sciences*. 6(2):81-85.
- Knobe, K., and Berntorp, E. (2011). Haemophilia and Joint Disease: Pathophysiology Evaluation and Management. *Journal of Comorbidity*. 1:51–59.
- Lannoy, N., Abinet, I., Bosmans, A., Lambert, C., Vermeylen, C., and Hermans, C. 2012. Computational and Molecular Approaches for Predicting Unreported causal Missense Mutations in Belgian Patients with Haemophilia A. *Hemophilia*. 18(3):331-339.
- Lee, Y., Gamazon, E. R., Rebman, E., Lee, Y., Lee, S., Dolan, M. E., Cox, N. J., and Lussier, Y. A. 2012. Variants Affecting Exon Skipping Contribute to Complex Traits. *PLoS Genetics*. 8(10):1-12.
- Liu, Y., Li, D., Yu, D., Liang, Q., Chen, G., Li, F., Gao, L., Li, Z., Xie, T., Wu, L., Mao, A., Wu, L., and Liang, D. 2023. Comprehensive Analysis of Hemophilia A (CAHEA): Towards Full Characterization of the F8 Gene Variants by Long-Read Sequencing. *Thrombosis and Haemostasis*. 123(12):1151-1164.

- Lucena-Aguilar G., Sanchez-Lopez A. M., Barberan-Aceituno C., Carrillo-Avila J. A., Lopez-Guerrero J. A., and Aguilar-Quesada R. 2016. DNA Source Selection for Downstream Applications based on DNA Quality Indicators Analysis. *Biopreserv Biobank*. 14(4):264-270.
- Luo, L., Zheng, Q., Chen, Z., Huang, M., Fu, L., Hu, J., Shi, Q., and Chen, Y. 2022. Hemophilia A Patients with Inhibitors: Mechanistic Insights and Novel Therapeutic Implications. *Frontiers in Immunology*. 13:1-15.
- Mahlangu, J., Dolan, G., Dougal, A., Goddard, Hernandez, E. D. P., N, J., Pierce G.F., Ragni. M. V., Rayner, B., Windyga, J., and Srivastava, A. 2020. WFH Guidelines for the Management of Hemophilia, 3rd edition Chapter 7: Treatment of Specific Hemorrhages. *Hemorrhages*. 1-158.
- Mandal, S., Gami, S., and Shah, S. 2020. A Case Report on an Extremely Rare Disease: Factor XI Deficiency. *Cureus*. 12(10):1-4.
- Marian, A. J. 2020. Clinical Interpretation and Management of Genetic Variants. *JACC Basic Transl*. 5(10):1029-1042.
- McVey, J. H., Rallapalli, P. M., Cook, G. K., Hampshire, D.J., Blaizot, M. G., Gomez, K., Perkins, S.J and Ludlam, C.A. 2020. The European Association for Haemophilia and Allied Disorders (EAHAD) Coagulation Factor Variant Databases. *Haemophilia*. 26: 306-313.
- Mazurkiewicz-Pisarek, A., Plucienniczak, G., Ciach, T., & Plucienniczak, A. (2016). The factor VIII protein and its function. *Acta biochimica Polonica*. 63(1):11–16.
- Mehta, P. and Reddivari, A. K. R. 2024. Hemophilia. StartPearls Publishing LLC
- Miller, C. H. 2021. The Clinical Genetics of Hemophilia B (Factor IX Deficiency). *The Application of Clinical Genetics*. 14: 445-454.
- Miller, C. H., Benson, J., Ellingsen, D., Driggers, J., Payne, A., Kelly, F. M., Soucie, J. M., and Craig Hooper, W. 2012. F8 and F9 Mutations in US Haemophilia Patients: Correlation with History of Inhibitor and Race/Ethnicity. *Haemophilia*. 18(3):375-382.
- Morfini, M., Coppola, A., Franchini, M., and Minno, D. G. 2013. Clinical Use of Factor VIII and Factor IX Concentrates. *Blood Transfusion*. 11:55-63.

- Nguyen, H., Das, U., Wang, B., & Xie, J. 2018. The Matrices and Constraints of GT/AG Splice Sites of more than 1000 Species/Lineages. *Gene*. 660:92-101.
- Nissen, S. K., Laursen, A. L., and Poulsen, L. H. 2018. Identification of a Novel Mutation in The Factor VIII Gene Causing Severe Haemophilia A. *BMC Hematol*. 18(17):1-4.
- Niu, J., Ning, L., Zhang, Q., Liu, Z., Ma, Y., Xu, X., Wu, Q., Hao, Y., Cui, Y., and Liu, C. 2022. Health-Related Quality of Life of Patients with Haemophilia: a Cross-Sectional Survey in The Northeast of China. *BMJ*. 12(2):1-9.
- Palta, S., Saroa, R., and Palta, A. 2014. Overview of the Coagulation System. *Indian Journal of Anaesthesia*. 58(5):515-523.
- Rogalska, M. E., Vivori, C., and Valcarcel, J. 2023. Regulation of pre-mRNA Splicing: Roles in Physiology and Disease and Therapeutic Prospects. *Genetics*. 24(4):251-269.
- Salen, P and Babiker, H.M. 2023. Hemophilia A. StatPearls Publishing LLC.
- Sasanakul, W., Chuansumrit, A., Sirachainan, N., and Kadegasem, P. 2022. Prominent Mutation of Intron 22 Inversion in Sporadic Hemophilia: Is It Worth The Antenatal Screening. *The Application of Clinical Genetics*. 15:49-54.
- Shen, B. W., Spiegel, P. C., Chang, C. H., Huh, J. W., Lee, J. S., Kim, J., Kim, Y. H., and Stoddard, B. L. 2008. The Tertiary Structure and Domain Organization of Coagulation Factor VIII. *Blood*. 111(3):1240-1247.
- Shih, M. Y., Wang, J. D., Yin, J. D., Tsan, Y. T., and Chan, W. C. 2020. Differences in Major Bleeding Events between Patients with Severe Hemophilia A and Hemophilia B: a Nationwide, Population-Based Cohort Study. *Clinical and Applied Thrombosis*. 25:1-7.
- Shinozawa, K., Amano, K., Hagiwara, T., Bingo, M., Chikasawa, Y., Inaba, H., Kinai, E., and Fukutake, K. 2021. Genetic Analysis of Carrier Status in Female Members of Japanese Hemophilia Families. *Journal of Thrombosis and Haemostasis*. 19(6):1493-1505.
- Sinclair A. 2002. Genetics 101: Detecting Mutations in Human Genes. *CMAJ*. 167(3):275-279.

- Singh, M and kaur, H. 2002. Assessment of the carrier status by *pedigree* analysis in some families from India. *Haemophilia*. 8: 680–684.
- Smith S. A. 2009. The Cell-Based Model of Coagulation. *Journal of Veterinary Emergency and Critical Care*. 19(1):3-10.
- Smith, S. A., Travers, R. J., and Morrissey, J. H. 2015. How It all Starts: Initiation of The Clotting Cascade. *Critical Reviews in Biochemistry and Molecular Biology*. 50(4):326-336.
- Sola, N. F., Morovvati, S., Sabetghadam Moghadam, M., and Entezari, M. 2020. Mutation Detection and Inhibitor Risk in Iranian Patients with Hemophilia A: Six Novel Mutations. *Clinical Case Reports*. 8(12):2976-2985.
- Srivastava, A., Santagostino, E., Dougall, A., Kitchen, S., Sutherland, M., Pipe, S. W., Carcao, M., Mahlangu, J., Ragni, M. V., Windyga, J., Llinas, A., Goddard, N. J., Mohan, R., Poonnoose, P. M., Feldman, B. M., Lewis, S. Z., van den Berg, H. M., and Pierce, G. F. 2020. WFH Guidelines for the Management of Hemophilia, 3rd Edition. *Haemophilia*. 1-158.
- Sun, J., Zhou, X., and Hu, N. 2021. Factor VIII Replacement Prophylaxis in Patients with Hemophilia A Transitioning to Adults. *Orphanet Journal of Rare Diseases*. 16(1):1-16.
- Thompson, A. 2003. Structure and Function of The Factor VIII Gene and Protein. *Seminars in Thrombosis and Hemostasis*. 29(1):11-22.
- Uen C, Oldenburg J, Schroder J, Brackmann H J, Schwaab R, Schneppenheim R, Graw, J. Mutation H. 2% Hamophilie-A-Patienten ohne Mutation im. 2003;1–5.
- Valikhani, A., Mirakhorly, M., Namvar, A., Rastegarlari, G., Toogeh, G., Shirayeh, F. V., and Ahmadinejad, M. 2021. Genetic Analysis of Non-Severe Hemophilia A Phenotype with a Discrepancy between One-Stage and Chromogenic Factor VIII Activity Assays. *Transfusion and Apheresis Science*. 60:1-8.
- Vencesla, A., Corral-Rodriguez, M. A., Baena, M., Cornet, M., Domenech, M., Baiget, M., Fuentes-Prior, P., and Tizzano, E. F. 2008. Identification of 31 Novel Mutations in the F8 Gene in Spanish Hemophilia A Patients: Structural Analysis of 20 Missense Mutations Suggests New Intermolecular Binding Sites. *Blood*. 111(7):3468-3478.

- Verhagen, M. J. A., van Heerde, W. L., van der Bom, J. G., Beckers, E. A. M., Blijlevens, N. M. A., Coppens, M., Gouw, S. C., Jansen, J. H., Leebeek, F. W. G., van Vulpen, L. F. D., Meijer, D., and Schols, S. E. M. 2023. In Patients with Hemophilia, a Decreased Thrombin Generation Profile Is Associated with a Severe Bleeding Phenotype. *Research and Practice in Thrombosis and Haemostasis*. 7(2):1-12.
- Villarreal-Martinez, L., Ibarra-Ramirez, M., Calvo-Anguiano, G., Lugo-Trampe, J. J., Luna-Zaizar, H., Martinez-de-Villarreal, L. E., Melendez-Aranda, L., and Jaloma-Cruz, A. R. 2020. Molecular Genetic Diagnosis by Next Generation Sequencing in a Cohort of Mexican Patients with Haemophilia and Report of Novel Variants. *Blood Cells. Molecules and Diseases*. 83:1-6.
- Wang, J., Xiao, H., Gu, J., Chen, H., Wu, Q., Xiong, L., Qiao, B., Zhang, Y., and Tong, Y. 2022. A Novel Deletion Mutation of The F8 Gene for Hemophilia A. *Diagnostics*. 12:1-10.
- Wang, J., Ye, Z., Huang, T. H., Shi, H., and Jin, V. X. 2017. Computational Methods and Correlation of Exon-skipping events with Splicing, Transcription, and Epigenetic Factors. *Methods in Molecular Biology*. 1513: 163–170.
- WFH. 2021. Annual Report 2021.
- Wolberg A. S. 2007. Thrombin Generation and Fibrin Clot Structure. *Blood Reviews*. 21(3):131-142.
- Yu M., Cao Y., Ji Y. 2017. The Principle and Application of new PCR Technologies. *IOP Conference Series*. 1-4
- Zahari, M., Sulaiman, S. A., Othman, Z., Ayob, Y., Karim, F. A., and Jamal, R. 2018. Mutational Profiles of F8 and F9 in a Cohort of Haemophilia A and Haemophilia B Patients in the Multi-ethnic Malaysian Population. *Mediterranean Journal of Hematology and Infectious Diseases*. 10(1):1-13.
- Zarrilli, F., Coppola, A., Schiavulli, M., Cimino, E., Elce, A., Rescigno, G., Castaldo, G., and Amato, F. 2018. Haemophilia A: The Consequences of de Novo Mutations Two Case Reports. *Blood Transfusion*. 16(4):392-393.
- Zhang, H., Li, Y., Lv, X., Mao, Y., Sun, Y., and Xu, T. 2023. A Novel F8 Variant in A Chinese Hemophilia A Family and Involvement of X-chromosome Inactivation: A Case Report. *Medicine*. 102(18):1-6.



Zimmermann, M. A., Gehrig, A., Oldenburg, J., Muller, C. R., and Rost, S. 2013. Analysis of F8 mRNA in Haemophilia A Patients with Silent Mutations or Presumptive Splice Site Mutations. *Haemophilia*. 19(2):310-317.