

Hochdurchsatz-Sequenzierung – Anwendung in der Thrombozytendiagnostik

1. International Human Genome Sequencing Consortium. Initial sequencing and analysis of the human genome. *Nature*. 2001;409:860-921.
2. Hua T, Chitnis N, Monos D, Dinh A. Next-generation sequencing technologies: An overview. *Human Immunology*. 2021;82:801–811.
3. Richards S, Aziz N, Bale S, Bick D, Das S, Gastier-Foster J, Grody WW, Hegde M, Lyon E, Spector E, Voelkerding K, Rehm HL; ACMG Laboratory Quality Assurance Committee. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. *Genet Med*. 2015;17:405-424.
4. White JG. Electron microscopy methods for studying platelet structure and function. *Methods Mol Biol*. 2004;272:47-63.
5. Lentaigne C, Freson K, Laffan MA, Turro E, Ouwehand WH; BRIDGE-BPD Consortium and the ThromboGenomics Consortium. Inherited platelet disorders: toward DNA-based diagnosis. *Blood*. 2016;127:2814-2823.
6. Sivapalaratnam S, Collins J, Gomez K. Diagnosis of inherited bleeding disorders in the genomic era. *Br J Haematol*. 2017;179:363-376.
7. Knöfler R, Eberl W, Schulze H, Bakchoul T, Bergmann F, Gehrlich S, Geisen C, Gottstein S, Halimeh S, Harbrecht U, Kappert G, Kirchmaier C, Kehrel B, Lösche W, Krause M, Mahnel R, Meyer O, Pilgrimm AK, Pillitteri D, Rott H, Santoso S, Siegemund A, Schambeck C, Scheer M, Schmutz M, Scholl T, Strauss G, Zieger B, Zotz R, Hermann M, Streif W. Diagnosis of inherited diseases of platelet function. Interdisciplinary S2K guideline of the Permanent Paediatric Committee of the Society of Thrombosis and Haemostasis Research (GTH e.V.). *Hamostaseologie*. 2014;34:201-212.
8. S2k Leitlinie-Thrombozytopathien; Version 2.1; AWMF-Register Nr. 086-003; update 2/2018. <https://www.awmf.org/leitlinien/detail/II/086-003.html>
9. Simeoni I, Stephens JC, Hu F, Deevi SV, Megy K, Bariana TK, Lentaigne C, Schulman S, Sivapalaratnam S, Vries MJ, Westbury SK, Greene D, Papadia S, Alessi MC, Attwood AP, Ballmaier M, Baynam G, Bermejo E, Bertoli M, Bray PF, Bury L, Cattaneo M, Collins P, Daugherty LC, Favier R, French DL, Furie B, Gattens M, Germeshausen M, Ghevaert C, Goodeve AC, Guerrero JA, Hampshire DJ, Hart DP, Heemskerk JW, Henskens YM, Hill M, Hogg N, Jolley JD, Kahr WH, Kelly AM, Kerr R, Kostadima M, Kunishima S, Lambert MP, Liesner R, López JA, Mapeta RP, Mathias M, Millar CM, Nathwani A, Neerman-Arbez M, Nurden AT, Nurden P, Othman M, Peerlinck K, Perry DJ, Poudel P, Reitsma P, Rondina MT, Smethurst PA, Stevenson W, Szkotak A, Tuna S, van Geet C, Whitehorn D, Wilcox DA, Zhang B, Revel-Vilk S, Gresele P, Bellissimo DB, Penkett CJ, Laffan MA, Mumford AD, Rendon A, Gomez K, Freson K, Ouwehand WH, Turro E. A high-throughput sequencing test for diagnosing inherited bleeding, thrombotic, and platelet disorders. *Blood*. 2016;127:2791-2803.
10. Downes K, Megy K, Duarte D, Vries M, Gebhart J, Hofer S, Shamardina O, Deevi SV, Stephens J, Mapeta R, Tuna S, Al Hasso N, Besser MW, Cooper N, Daugherty L, Gleadall N, Greene D, Haimel M, Martin H, Papadia S, Revel-Vilk S, Sivapalaratnam S, Symington E, Thomas W, Thys C, Tolios A, Penkett CJ; NIHR BioResource, Ouwehand WH, Abbs S, Laffan MA, Turro E, Simeoni I, Mumford AD, Henskens YMC, Pabinger I, Gomez K, Freson K. Diagnostic high-throughput sequencing of 2396 patients with bleeding, thrombotic, and platelet disorders. *Blood*. 2019;134:2082-2091.
11. Andres O, König EM, Althaus K, Bakchoul T, Bugert P, Eber S, Knöfler R, Kunstmann E, Manukjan G, Meyer O, Strauß G, Streif W, Thiele T, Wiegering V, Klopocki E, Schulze H; THROMKIDplus Study Group of the Society of Paediatric Oncology Haematology (Gesellschaft für Pädiatrische Onkologie und Hämatologie, GPOH) and the Society of Thrombosis Haemostasis Research (Gesellschaft für Thrombose- und Hämostaseforschung, GTH). Use of Targeted High-Throughput Sequencing for Genetic Classification of Patients with Bleeding Diathesis and Suspected Platelet Disorder. *TH Open*. 2018;2:e445-e454.
12. Bastida JM, Lozano ML, Benito R, Janusz K, Palma-Barqueros V, Del Rey M, Hernández-Sánchez JM, Riesco S, Bermejo N, González-García H, Rodríguez-Alén A, Aguilar C, Sevivas T, López-Fernández MF, Marneth AE, van der Reijden BA, Morgan NV, Watson SP, Vicente V, Hernández-Rivas JM, Rivera J, González-Porrás JR. Introducing high-throughput sequencing into mainstream genetic diagnosis practice in inherited platelet disorders. *Haematologica*. 2018;103:148-162.
13. Megy K, Downes K, Simeoni I, Bury L, Morales J, Mapeta R, Bellissimo DB, Bray PF, Goodeve AC, Gresele P, Lambert M, Reitsma P, Ouwehand WH, Freson K; Subcommittee on Genomics in Thrombosis and Hemostasis. Curated disease-causing genes for bleeding, thrombotic, and platelet disorders: Communication from the SSC of the ISTH. *J Thromb Haemost*. 2019;17:1253-1260.
14. Saes JL, Simons A, de Munnik SA, Nijziel MR, Blijlevens NMA, Jongmans MC, van der Reijden BA, Smit Y, Brons PP, van Heerde WL, Schols SEM. Whole exome sequencing in the diagnostic workup of patients with a bleeding diathesis. *Haemophilia*. 2019;25:127-135.
15. Khan AO, Stapley RJ, Pike JA, Wijesinghe SN, Reyat JS, Almazni I, Machlus KR, Morgan NV; UK GAPP Study Group. Novel gene variants in patients with platelet-based bleeding using combined exome sequencing and RNAseq murine expression data. *J Thromb Haemost*. 2021;19:262-268.