

Importância do sequenciamento de nova geração para o diagnóstico e tratamento dos erros inatos da imunidade

VASCONCELOS, Dewton

Referências:

[1] McKusick VA. Mendelian Inheritance in Man and its online version, OMIM. Am J Hum Genet. 2007 Apr;80(4):588-604.

[2] Tangye S et al. Human Inborn Errors of Immunity: 2019 Update on the Classification From the International Union of Immunological Societies Expert Committee. J Clin Immunol. 2020 Jan;40(1):24-64. doi: 10.1007/s10875-019-00737-x. Epub 2020 Jan 17.

[3] Bonilla FA. Laboratory evaluation of the immune system. UpToDate v. 19.3, January 2012 (www.uptodate.com).

[4] Brodin P. New approaches to the study of immune responses in humans. Human Genetics 2020. <https://doi.org/10.1007/s00439-020-02129-3>.

[5] Notarangelo LD. Primary immunodeficiencies. J Allergy Clin Immunol. 2010 Feb;125(2 Suppl 2):S182-94.

[6] Platt C, Geha RS, Chou J. Gene hunting in the genomic era: Approaches to diagnostic dilemmas in patients with primary immunodeficiencies. J Allergy Clin Immunol. 2013 Oct 4

[7] Brown L, Xu-Bayford J, Allwood Z, Slatter M, Cant A, Davies EG, et al. Neonatal diagnosis of severe combined immunodeficiency leads to significantly improved survival outcome: the case for newborn screening. Blood. 2011 Mar 17;117(11):3243-6.

[8] Nijman IJ, van Montfrans JM, Hoogstraat M, Boes ML, van de Corput L, Renner ED, et al. Targeted next-generation sequencing: A novel diagnostic tool for primary immunodeficiencies. J Allergy Clin Immunol. 2013 Oct 15.

- [9] Immunodeficiency resource. [HTTP://bioinf.uta.fi/index.shtml](http://bioinf.uta.fi/index.shtml)
- [10] Moens L, Meyts I. Recent human genetic errors of innate immunity leading to increased susceptibility to infection *Current Opinion in Immunology* 2020, 62:79–90.
- [11] Ansorge WJ. Next-generation DNA sequencing techniques. *N Biotechnol.* 2009 Apr; 25(4):195-203.
- [12] Badolato R, Prandini A, Caracciolo S, Colombo F, Tabellini G, Giacomelli M, et al. Exome sequencing reveals a pallidin mutation in a Hermansky-Pudlak-like primary immunodeficiency syndrome. *Blood.* 2012 Mar 29;119(13):3185-7.
- [13] de Greef JC, Wang J, Balog J, den Dunnen JT, Frants RR, Straasheijm KR, et al. Mutations in ZBTB24 are associated with immunodeficiency, centromeric instability, and facial anomalies syndrome type 2. *Am J Hum Genet.* 2011 Jun 10;88(6):796-804.
- [14] Moshous D, Martin E, Carpentier W, Lim A, Callebaut I, Canioni D, et al. Whole-exome sequencing identifies Coronin-1A deficiency in 3 siblings with immunodeficiency and EBV-associated B-cell lymphoproliferation. *J Allergy Clin Immunol.* 2013 Jun;131(6):1594-603.
- [15] Meyts I, Bosch B, Bolze A, et al. Exome and genome sequencing for inborn errors of immunity. *J Allergy Clin Immunol.* 2016;138(4):957-969. doi:10.1016/j.jaci.2016.08.003
- [16] Arts P, Simons A, AlZahrani MS, et al. Exome sequencing in routine diagnostics: a generic test for 254 patients with primary immunodeficiencies. *Genome Med.* 2019;11(1):38. Published 2019 Jun 17. doi:10.1186/s13073-019-0649-3
- [17] Telenti A, di Iulio J. Regulatory genome variants in human susceptibility to infection. *Hum Genet.* 2020;139(6-7):759-768. doi:10.1007/s00439-019-02091-9
- [18] Chou J, Ohsumi TK, Geha RS. Use of whole exome and genome sequencing in the identification of genetic causes of primary immunodeficiencies. *Curr Opin Allergy Clin Immunol.* 2012 Dec;12(6):623-8.

[19] Wang Z, Liu X, Yang BZ, Gelernter J. The role and challenges of exome sequencing in studies of human diseases. *Front Genet.* 2013;4:160.

[20] Bamshad MJ, Ng SB, Bigham AW, Tabor HK, Emond MJ, Nickerson DA, et al. Exome sequencing as a tool for Mendelian disease gene discovery. *Nat Rev Genet.* 2011 Nov;12(11):745-55.

[21] Clark MJ, Chen R, Lam HY, Karczewski KJ, Euskirchen G, Butte AJ, et al. Performance comparison of exome DNA sequencing technologies. *Nat Biotechnol.* 2011 Oct;29(10):908-14.

[22] Biesecker LG. Exome sequencing makes medical genomics a reality. *Nat Genet.* 2010 Jan;42(1):13-4.

[23] Ng SB, Turner EH, Robertson PD, Flygare SD, Bigham AW, Lee C, et al. Targeted capture and massively parallel sequencing of 12 human exomes. *Nature.* 2009 Sep 10;461(7261):272-6.

[24] Ng SB, Buckingham KJ, Lee C, Bigham AW, Tabor HK, Dent KM, et al. Exome sequencing identifies the cause of a mendelian disorder. *Nat Genet.* 2010a Jan;42(1):30-5.

[25] Joshi AY, Iyer VN, Hagan JB, St Sauver JL, Boyce TG. Incidence and temporal trends of primary immunodeficiency: a population-based cohort study. *Mayo Clin Proc.* 2009;84(1):16-22.

[26] Delmonte OM, Castagnoli R, Calzoni E, Notarangelo LD. Inborn Errors of Immunity With Immune Dysregulation: From Bench to Bedside. *Frontiers in Pediatrics*, 2019; 7: 353. DOI=10.3389/fped.2019.00353