

Publications

Epigenetic Immune Cell Quantification Technology Description

Comparison with flow cytometry; dried blood spot analysis; detection of SCID/XLA in newborns

1. Baron U, Werner J, Schildknecht K, Schulze JJ, Mulu A, Liebert UG, Sack U, Speckmann C, Gossen M, Wong RJ, Stevenson DK, Babel N, Schürmann D, Baldinger T, Bacchetta R, Grützkau A, Borte S, Olek S. Epigenetic immune cell counting in human blood samples for immunodiagnosics. *Sci Transl Med*. 2018 Aug 1;10(452). pii: ean3508. doi: 10.1126/scitranslmed.aan3508. [PubMed PMID: 30068569](#)

Comparison with flow cytometry; cord vs. adult blood

2. Nettenstrom L, Alderson K, Raschke EE, Evans MD, Sondel PM, Olek S, Seroogy CM. An optimized multi-parameter flow cytometry protocol for human T regulatory cell analysis on fresh and viably frozen cells, correlation with epigenetic analysis, and comparison of cord and adult blood. *J Immunol Methods*. 2013 Jan 31;387(1-2):81-8. doi: 10.1016/j.jim.2012.09.014. Epub 2012 Oct 9. PubMed PMID: 23058673; [PubMed Central PMCID: PMC3529842](#)
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DNA (de)methylation markers for detection of specific immune cell subtypes

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6. Wiczorek G, Asemissen A, Model F, Turbachova I, Floess S, Liebenberg V, Baron U, Stauch D, Kotsch K, Pratschke J, Hamann A, Loddenkemper C, Stein H, Volk HD, Hoffmüller U, Grützkau A, Mustea A, Huehn J, Scheibenbogen C, Olek S. Quantitative DNA methylation analysis of FOXP3 as a new method for counting regulatory T cells in peripheral blood and solid tissue. *Cancer Res*. 2009 Jan 15;69(2):599-608. doi: 10.1158/0008-5472.CAN-08-2361. [PubMed PMID: 19147574](#)
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8. Baron U, Floess S, Wiczorek G, Baumann K, Grützkau A, Dong J, Thiel A, Boeld TJ, Hoffmann P, Edinger M, Türbachova I, Hamann A, Olek S, Huehn J. DNA demethylation in the human FOXP3

locus discriminates regulatory T cells from activated FOXP3(+) conventional T cells. *Eur J Immunol.* 2007 Sep;37(9):2378-89. [PubMed PMID: 17694575](#)

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Early Detection of Inborn Errors of Immunity in Newborns

10. Blom M, Bredius RGM, van der Burg M. Future Perspectives of Newborn Screening for Inborn Errors of Immunity. *Int J Neonatal Screen.* 2021 Nov 2;7(4):74. doi: 10.3390/ijns7040074. PMID: 34842618; [PMCID: PMC8628921](#).
11. Blom M, Pico-Knijnenburg I, Imholz S, Vissers L, Schulze J, Werner J, Bredius R, van der Burg M. Second Tier Testing to Reduce the Number of Non-actionable Secondary Findings and False-Positive Referrals in Newborn Screening for Severe Combined Immunodeficiency. *J Clin Immunol.* 2021 Nov;41(8):1762-1773. doi: 10.1007/s10875-021-01107-2. Epub 2021 Aug 9. PMID: 34370170; [PMCID: PMC8604867](#)

Epigenetic Quantification of Regulatory T-cells (Treg) for disease detection, prognosis and monitoring

Immune regulatory disorders

12. Cepika AM, Sato Y, Liu JM, Uyeda MJ, Bacchetta R, Roncarolo MG. Tregopathies: Monogenic diseases resulting in regulatory T-cell deficiency. *J Allergy Clin Immunol.* 2018 Dec;142(6):1679-1695. doi: 10.1016/j.jaci.2018.10.026. Review. PubMed PMID: 30527062
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Solid Tumors

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Cardiovascular Disease

23. Barth SD, Kaaks R, Johnson T, Katzke V, Gellhaus K, Schulze JJ, Olek S, Kühn T. The Ratio of Regulatory (FOXP3+) to Total (CD3+) T Cells Determined by Epigenetic Cell Counting and Cardiovascular Disease Risk: A Prospective Case-cohort Study in Non-diabetics. *EBioMedicine*. 2016 Sep;11:151-156. doi: 10.1016/j.ebiom.2016.07.035. Epub 2016 Jul 30. PubMed PMID: 27499494; PubMed Central PMCID: PMC5049920

Allergy, inflammation, autoimmunity

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Epigenetic Quantification of immune cells in therapy monitoring

Tumor vaccination

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Hematopoietic stem cell transplantation

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Solid organ transplantation

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Autoimmune hepatitis

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Regulatory T-cells as immunosuppressive therapy (adoptive transfer of Treg)

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DNA methylation analysis as quality control in regenerative medicine

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Epigenetic Data Analysis

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