Purine and Pyrimidine analysis

Jörgen Bierau, Ph.D. – ERNDIM Scientific Advisor for Purines and Pyrimidines

The past decade, hyphenated tandem mass spectrometry have become the standard mode of analysis in many laboratories providing screening and diagnostic follow-up for inborn errors of metabolism. Thanks to whole exome sequencing and the efforts of dedicated scientist many new defects in purine and pyrimidine metabolism have been identified over the past decade as well. Perhaps with the exception of dihydro-orotic acid and N-carbamyl aspartatic acid no new biomarkers have been reported, yet. It is to be expected that sooner or later intermediates of purine de novo synthesis will be identified as biomarkers for defects in purine de novo synthesis that still have to be found. The new metabolomics and multi-omics approaches will certainly accelerate this.

Good and reliable sources for the analysis of purines and pyrimidines, and clinical and diagnostic information are given below. The previous version of the advisory document contains a lot of biochemical background generic methods for sample collection and HPLC-UV analysis and be downloaded.

Sources for purine and pyrimidine analysis using tandem mass spectrometry:

Monostori P, Klinke G, Hauke J, Richter S, Bierau J, Garbade SF, Hoffmann GF, Langhans CD, Haas D, Okun JG (2019) . Extended diagnosis of purine and pyrimidine disorders from urine: LC MS/MS assay development and clinical validation. PLoS One. 2019 14(2):e0212458. doi: 10.1371

Hartmann S, Okun JG, Schmidt C, Langhans CD, Garbade SF, Burgard P, Haas D, Sass JO, Nyhan WL, Hoffmann GF. Comprehensive detection of disorders of purine and pyrimidine metabolism by HPLC with electrospray ionization tandem mass spectrometry. Clin Chem. 2006 52(6):1127-37.

van Kuilenburg ABP, van Cruchten A, Abeling NGGM (2008) Screening for disorders of purine and pyrimidine metabolism using HPLC-electrospray tandem mass spectrometry. In: Blau N, Duran M, Gibson KM (eds) Laboratory guide to the methods in biochemical genetics. Springer, Berlin

Sources for clinical and diagnostic information:

Balasubramaniam S, Duley JA, Christodoulou J. Inborn errors of purine metabolism: clinical update and therapies. J Inherit Metab Dis. 2014 37(5):669-86. doi: 10.1007/s10545-014-9731-6

Balasubramaniam S, Duley JA, Christodoulou J. Inborn errors of pyrimidine metabolism: clinical update and therapies. J Inherit Metab Dis. 2014 37(5):687-98. doi: 10.1007/s10545-014-9742-3

Bierau J, Šebesta I (2014) Purine and pyrimidine disorders. In: Blau N, Duran M, Gibson KM, Dionisi-Vici C (eds) Physician's guide to the diagnosis, treatment, and follow-up of inherited metabolic diseases. Springer, Berlin.

Do not hesitate to contact me if you have any question regarding purines and pyrimidines.

Jörgen Bierau

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