**CV Johannes A. MAYR (Principal Investigator MEET, WP3, T-3.2)**
born October 6th, 1968, Austrian citizenship

*Education*
2013 Venia docendi in Pathobiochemistry and Molecular Genetics, PMU Salzburg
1997 PhD in Biochemistry, Institute of Biochemistry, TU Graz
1992 Diploma in Technical Chemistry, Institute of Biochemistry, TU Graz

*Present position*
Head of the diagnostic laboratory for "Mitochondrial Disorders" at the Department of Paediatrics, Paracelsus Medical University (PMU) Salzburg, Austria.

*Research interests*
Since 1998 Johannes Mayr has specialised in the diagnosis of inherited and acquired genetic defects of metabolism, with a special focus on the mitochondrial energy metabolism (MEM). More than 1700 samples from patients with suspected defects in the MEM have been investigated, a biochemical defect could be identified in approximately 300 of them. In 60% of them a genetic diagnosis could be made, including several novel genetic diseases. In addition to inherited defects, the relevance of the MEM in tumours is investigated, which lead to the identification of complex I deficiency and somatic mutations of the mitochondrial DNA in various oncocytic tumours, e.g. of the kidney and thyroid.

*Research institution*
The Department of Paediatrics of the PMU Salzburg is directed by Prof. Dr. Wolfgang Sperl, who is a pioneer of inherited metabolic diseases in Austria, especially in the field of mitochondrial diseases. The research laboratory at the Department of Paediatrics is equipped with all standard molecular biology tools including Sanger sequencing, HPLC, real-time PCR, high resolution respirometry, spectrophotometer and a multimode plate reader.

*Selected peer reviewed publications*
Lack of the mitochondrial protein acylglycerol kinase causes Sengers syndrome.
Mayr JA, Haack TB, Graf E, Zimmermann FA, Wieland T, Haberberger B, Superti-Furga A, Kirschner J, Steinmann B, Baumgartner MR, Moroni I, Lamantea E, Zeviani M, Rodenburg RJ, Smeitink J, Strom TM, Meitinger T, Sperl W, Prokisch H.
Am J Hum Genet. 2012;90:314-20.
Thiamine pyrophosphokinase deficiency in encephalopathic children with defects in the pyruvate oxidation pathway.
Mayr JA, Freisinger P, Schlachter K, Rolinski B, Zimmermann FA, Scheffner T, Haack TB, Koch J, Ahting U, Prokisch H, Sperl W.
Am J Hum Genet. 2011;89:806-12.
Lipoic acid synthetase deficiency causes neonatal-onset epilepsy, defective mitochondrial energy metabolism, and glycine elevation.
Mayr JA, Zimmermann FA, Fauth C, Bergheim C, Meierhofer D, Radmayr D, Zschocke J, Koch J, Sperl W.
Am J Hum Genet. 2011;89:792-7.
Mitochondrial ATP synthase deficiency due to a mutation in the ATP5E gene for the F1 epsilon subunit.
Mayr JA, Havlícková V, Zimmermann F, Magler I, Kaplanová V, Jesina P, Pecinová A, Nusková H, Koch J, Sperl W, Houstek J.
Hum Mol Genet. 2010;19:3430-9
Mitochondrial phosphate-carrier deficiency: a novel disorder of oxidative phosphorylation.
TMEM70 mutations cause isolated ATP synthase deficiency and neonatal mitochondrial encephalocardiomyopathy.
Cízková A, Stránecký V, Mayr JA, Tesarová M, Havlícková V, Paul J, Ivánek R, Kuss AW, Hansíková H, Kaplanová V, Vrbacký M, Hartmannová H, Nosková L, Honzík T, Drahota Z, Magner M, Hejzlarová K, Sperl W, Zeman J, Houstek J, Kmoch S.
Nat Genet. 2008;40:1288-90.
Mitochondrial phosphate-carrier deficiency: a novel disorder of oxidative phosphorylation.
Mayr JA, Merkel O, Kohlwein SD, Gebhardt BR, Böhles H, Fötschl U, Koch J, Jaksch M, Lochmüller H, Horváth R, Freisinger P, Sperl W.
Am J Hum Genet. 2007;80:478-84.