Introduction: Long-chain polyunsaturated fatty acid (LCPUFA) can be provided by diet (fatty fish, eggs, viscera and human milk) or synthesised from essential fatty acids linoleic and -linolenic acids through the microsomal pathway. However, endogenous LCPUFA synthesis is rather low, especially for docosahexaenoic (DHA), and seems insufficient to achieve normal DHA values in individuals devoid of preformed dietary supply. Inborn errors of metabolism (IEMs) are therefore diseases with a special risk for LCPUFA deficient status. Aim: Our aim was to evaluate LCPUFA status in 132 patients with different IEMs. Methods: We performed a cross-sectional study of plasma and erythrocyte LCPUFA composition of 63 patients with IEMs treated with protein-restricted diets compared with data from 69 patients with IEMs on protein-unrestricted diets, and 43 own reference values. Results: Erythrocyte and plasma DHA and arachidonic acid concentrations were significantly decreased in patients treated with protein-restriction compared with those on protein-unrestricted diets and with our reference values (p < 0.0001). In the protein-restricted group, 45% of patients showed decreased erythrocyte and plasma DHA values (only 7% and 10%, respectively in the proteinunrestricted group) (p < 0.0001). Erythrocyte and plasma DHA values correlated with the natural protein intake in patients on protein-restriction (r = 0.257; p = 0.045; r = 0.313; p = 0.014, respectively). Conclusion: Plasma and erythrocyte DHA concentrations are decreased in patients with IEMs treated with protein restriction. DHA concentration correlates with the patients’ protein intake. Supplementation of patients with LCPUFA would have a beneficial influence on their nutritional status.

Keywords
Polyunsaturated fatty acids, Docosahexaenoic acid, Inborn errors of metabolism, Protein restricted diets. LCPUFA