



Rare Inborn Errors of Metabolism in Children in Latvia

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Condition: New. Publisher/Verlag: LAP Lambert Academic Publishing | Inborn errors of metabolism (IEM) comprise a large class of genetic diseases involving disorders of metabolism. In most of the disorders, problems arise due to accumulation of substances which are toxic and it can cause acute or chronic intoxication, hypoglycaemia, or other metabolic disturbances. Clinical presentation in most cases are unspecific and in the neonatal period or during infancy could be misdiagnosed as manifestation of sepsis, birth trauma, encephalitis, sudden death syndrome or other disease. IEM individually are rare but collectively are common, and numbers of them are rising as there are improved diagnostic techniques. The aim of the study was to evaluate situation in Latvia in field of inborn errors of metabolism in childhood that could be detectable by selective screening. During study for 108 patients had confirmed diagnosis of rare inborn error of metabolism. 18 diseases were first time found in Latvia. The clinical and laboratory data were compared with data from other countries, calculated the prevalence of diagnosed IEM in Latvia and evaluated the problems in the diagnostic of rare inborn errors of metabolism in the state. | Format: Paperback | Language/Sprache: english | 56 pp.



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