Intellectual disability (ID) affects 2.5% of population worldwide. 85% of the treatable conditions known to cause global developmental delay or intellectual disability in children. Misdiagnosis is common and these disorders can mimic conditions like cerebral palsy. Recommendations to investigate possible metabolic or genetic aetiologies of intellectual disability are based on conditions and on the yield of diagnostic methods, rather than availability of causal therapy. Inborn errors of metabolism are subgroup of rare genetic/metabolic conditions for which an increasing number of treatments are now available. Some common causes of disorders of ID include disorder of amino acids cholesterol and fatty acids, creatine, glucose and its transport, lysosomes, metals, mitochondria, neurotransmission, organic acids, peroxisomes, urea cycle, and vitamins/co-factors. All these disorders can be identified by metabolic screening tests in the blood (plasma amino acids, etc.) and urine (creatine metabolites, glycosaminoglycans, etc.). Further or secondary tests depend on the results of these primary tests. Therapeutic modalities include sick-day management, dietary interventions, co-factor/vitamin supplements, substrate inhibition, stem cell transplant and gene therapy. Early interventions can improve or stabilize child’s cognitive and behavioral development, control the seizures or other neurological manifestations.

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