Abstract

Hypoglycemia and endocrine effects of adults' inborn errors of metabolism

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BACKGROUND: Inborn errors of metabolism (IEM) are rare diseases, most often inherited as an autosomal recessive disorder. They may be associated with endocrine dysfunction, the most frequent of them being disorders of carbohydrate metabolism (hypoglycemia, diabetes). The endocrinologist might be led to screen these complications in a patient whose diagnosis has been done during childhood. In some rare cases, he should evoke the diagnosis in front of an endocrine disorder most often associated to a multisystemic involvement.

DISCUSSION: This spreading field is new, not yet very well known in adulthood. Long-term consequences of IEM on fertility and bone metabolism are still poorly understood. Diagnosis orientation relies on a few specific lab investigations encompassing blood lactate, free fatty acids and 3-hydroxy-butyrate, ammoniemia, carnitine and acylcarnitines, aminoacid and urinary organic chromatography. Hyperinsulinism, glycogenosis, fatty acid ss-oxydation, carnitine cycle and glycosylation (CDG syndrome) disorders, fructose intolerance, tyrosinemia, organic aciduria may explain hypoglycemia. These diagnosis should be evoked in front of unexplained adult hypoglycemia. Diabetes is related to iron overload, mitochondriopathy and thiamine sensitive diabetes. Clinical spectrum of some forms of IEM switch from hypoglycemia in childhood to diabetes in adulthood. Mitochondriopathies can be associated to all types of endocrine disorders, the most frequent being diabetes and dysthyroidism. Hypothyroidism is encountered in mitochondriopathies, cystinosis and primary hyperoxaluria. Hypogonadism is almost constant in galactosemia, frequent in CDG syndromes, cystinosis and iron overload. Most of the time, a specialized advice is required, which is one of the mission of reference centres.

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