Aromatic L-Amino Acid Decarboxylase Deficiency Is a Cause of Long-Fasting Hypoglycemia

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Résumé en anglais	Objective/Context: Long-fasting hypoglycemia in children may be induced by neurotransmitter disorders. Case Report: A 5-year-old girl with a medical history of chronic diarrhea presented three episodes of severe hypoglycemia (20 mg/dL) between ages 3 and 5 years. She became pale and sweaty with hypothermia (33.5°C), bradycardia (45 bpm), and acidosis and presented a generalized seizure. During the 17-hour fast test performed to determine the etiology of her hypoglycemia, insulin and C-peptide were appropriately low, and human GH, IGF-I, cortisol, amino acids, and acylcarnitines were in the usual range for fasting duration. However, the presence of vanillactic and vanilpyruvic acids in urine led us to investigate the metabolism of dopamine and serotonin in the cerebrospinal fluid. Indeed, these results indicated an aromatic L-amino acid decarboxylase deficiency that impairs the synthesis of serotonin, dopamine, and catecholamines. The diagnosis was confirmed by the low aromatic L-amino acid decarboxylase (AADC) enzyme activity in plasma (5 pmol/min/mL; reference value, 20–130) and the presence of two heterozygous mutations, c.97G>C (p.V33L, inherited from her father) and c.1385G>C (p.R462P, inherited from her mother) in the DCC gene. She was supplemented with pyridoxine and raw cornstarch (1 g/kg) at evening dinner to reduce the night fast. The episodes of hypoglycemia and the chronic diarrhea were suppressed. Conclusion: Here is the first case report of long-fasting hypoglycemia due to a nontypical AADC deficiency. Hypoglycemia was severe, but the other neurological clinical hallmarks present in AADC-deficient patients were mild to moderate. Thus, neurotransmitter disorders should be considered in any patients presenting hypoglycemia with urine excretion of vanillactic acid.
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