A new case of pcsk1 pathogenic variant with congenital proprotein convertase 1/3 deficiency and literature review

Titre	A new case of pcsk1 pathogenic variant with congenital proprotein convertase 1/3 deficiency and literature review
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Résumé en anglais	Issue: To report a homozygous pathogenic variant in PCSK1 in a boy affected with proprotein convertase 1/3 (PC1/3) deficiency. Case description and literature review: A male infant born to consanguineous Turkish parents presented in the first week of life with transient central diabetes insipidus, watery diarrhea, micropenis due to hypogonadotropic hypogonadism and GH deficiency, and transient asymptomatic hypoglycemia. Further endocrine defects gradually appeared, including central hypothyroidism and mild central hypocortisolism (at 1 yr), central diabetes insipidus that reappeared progressively (at 2.5 yr), and obesity (at 2 yr). Whole exome sequencing revealed a homozygous nonsense pathogenic variant (NM_000439.4) c. 595 C>T in exon 5 of PCSK1, not yet reported in cases of proprotein convertase 1/3 (PC1/3) deficiency. To date, 26 cases of PC1/3 deficiency have been reported in the literature. All individuals had early and severe malabsorptive diarrhea and 83% had polyuria-polydipsia syndrome (before 5 yr). Most (79%) had early-onset obesity. Various endocrine disorders were present, including growth hormone deficiency (44%), mild central hypothyroidism (56%), central hypogonadism (44%), central hypocortisolism (57%), and postprandial hypoglycemia (52%). When described (n=15), proinsulin levels were consistently high: between 8 and 154 times the upper limit of normal (mean 74). Conclusion: We described a homozygous nonsense pathogenic variant (NM_000439.4) c. 595 C>T in exon 5 of PCSK1 in a boy with congenital proprotein convertase 1/3 deficiency. Elevated proinsulin could be useful in the diagnosis of this condition.
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