Unusual association : Allgrove syndrome and hypopituitarism

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Introduction	Results
Allgrove syndrome is a genetic disorder of autosomal recessive	This is the case of a boy who comes from a consanguineous marriage, with family history of hypogonadotropic hypogonadism in one sister. Its history goes back to the age of 2 years by the discovery of a micropenis associated with bilateral cryptorchidism, where the diagnosis of

complete form, esophageal achalasia, alacrymia and adrenal insufficiency. This is generally a resistant adreno-corticotropic hormone (ACTH). In this context, we report the case of a patient followed in the endocrinology department. hypogonadotropic hypogonadism was retained and the patient underwent orchidopexy and hormone replacement therapy and an alacrymia for which he was put under artificial tears. At the age of 22, he developed an adrenal insufficiency confirmed by a low level of serum cortisol contrasting with ACTH raised to 90 pg / ml. For a few years, he has been reporting the notion of dysphagia in association with an alacrymia and Addison's disease. The diagnosis of Allgrove was strongly suspected and the patient was subjected to a genetic investigation. The result of molecular analysis revealed the majority mutation in the homozygous state (IVS14 + 1G \mapsto A) by targeted molecular analysis of the AAAS gene that confirms the diagnosis of Allgrove in our patient.



Triple A syndrome is an autosomal recessive disorder characterized by adrenal insufficiency, achalasia of the esophageal cardia, and alacrima. Several other associated features have also been described in patients including progressive central, peripheral and autonomic nervous system abnormalities, palmo-plantar and punctate hyperkeratosis, developmental delays, and microcephaly. Using linkage analysis, Allgrove syndrome was mapped to chromosome 12q13 and mutations were subsequently identified in the AAAS gene (achalasia-adrenal insufficiency-alacrima syndrome gene).

The AAAS gene consists of 16 exons and encodes a WD-repeat protein, ALADIN (ALacrima Achalasia aDrenal Insufficiency Neurologic disorder) of 546 amino acids residues. ALADIN protein is located in the nuclear pore complexes, large multiprotein assemblies that are the sole site of nucleocytoplasmic transport (1,2). ALADIN plays a cell type-specific role in regulating nucleocytoplasmic transport and this function is essential for the proper maintenance and/or development of certain tissues such as adrenal gland, esophagus, cerebellum, pituitary gland, and pancreas (1).

CONCLUSION

Allgrove syndrome is a rare pediatric disorder, associating alacrymia and achalasia that are constant and early, and a less constant adrenal insufficiency. These disorders are at the origin of an alteration of the quality of life of the patients imposing a multidisciplinary care and especially a genetic advice in the siblings.

References

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