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3L0Q2V - LANG RODNEY

Allgrove or triple A syndrome (AS or AAA) is a rare autosomal recessive syndrome with variable phenotype due to mutations in AAAS gene which encodes a protein called ALADIN. Generally, it’s characterized by of adrenal insufficiency in consequence of adrenocorticotropic hormone (ACTH) resistance, besides of achalasia, and alacrimia.

Many cases of Allgrove (AAA) syndrome present with classic symptoms of primary adrenal insufficiency, including hypoglycemic seizures and shock. Less frequently, a child may be evaluated initially for recurrent vomiting, dysphagia, and failure to thrive (achalasia) or for ocular symptoms associated with alacrima.

We report a young woman with the clinical picture of Allgrove syndrome in whom neurological symptoms are prominent. It usually presents in the first decade of life with a deficiency of tears, recurrent vomiting and dysphagia due to achalasia, severe hypoglycemic seizures and shock due to adrenal insufficiency.

Allgrove syndrome - Medigoo - Health Medical Tests ...

Allgrove syndrome can arise from mutations of the ADRACALIN (or AAAS) gene encoding the ALADIN protein of the NPC. [6 , 7] Surprisingly, among many logical candidate genes that have not been associated with the disorder are those coding for the ACTH receptor, vasoactive intestinal polypeptide (VIP), the vip-1 receptor, pituitary adenylate ...

Allgrove Syndrome - an overview | ScienceDirect Topics

Many of the neurological symptoms of triple A syndrome worsen over time. People with triple A syndrome frequently develop a thickening of the outer layer of skin (hyperkeratosis) on the palms of their hands and the soles of their feet. Other skin abnormalities may also be present in people with this condition.

Allgrove (AAA) Syndrome: Background, Pathophysiology ...

Allgrove (AAA) Syndrome Clinical Presentation: History ...

Allgrove Syndrome. Allgrove, or triple A, syndrome is a similar disorder to FGD, with additional features of alacrima and achalasia. Presenting in the first decade of life, it is frequently associated with progressive neurologic dysfunction, polyneuropathy, deafness, mental retardation, and hyperkeratosis of palms and soles (Houlden et al, 2002).

Triple-A syndrome - Wikipedia

Download Citation | Neurological manifestations of Allgrove syndrome | Allgrove syndrome is a rare syndrome with conspicuous neurological abnormalities.

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The cases presented in this study showed the features of Allgrove syndrome during first decade in life with the neurological symptoms being the earliest manifestation, with decreased muscle power, sensation and deep reflexes in both brothers (more progressive in the elderly sib). In addition, cerebellar signs were clear.

The pattern and severity of neurologic and autonomic dysfunction in Triple-A syndrome is quite variable, including hyperreflexia, impaired visual evoked potentials, optic nerve atrophy, anisocoria (unequal pupil size), abnormal sweating, postural (orthostatic [genedx.com]

Allgrove Syndrome with Neurological Symptoms - Centogene

Triple A syndrome | Genetic and Rare Diseases Information ...

Triple A syndrome - Genetics Home Reference - NIH

In most cases, there is no family history of it. The syndrome was first identified by Jeremy Allgrove and colleagues in 1978. The syndrome involves achalasia, addisonianism (adrenal insufficiency of primary type), and alacrima (insufficiency of tears). Alacrima is usually the earliest manifestation.

Triple A Syndrome (Allgrove Syndrome): Symptoms, Diagnosis ...

A broad range of symptoms in allgrove syndrome: single ...

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Neurological Manifestations Of Allgrove Syndrome

Allgrove syndrome with prominent neurological symptoms ...

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CiteSeerX — NEUROLOGICAL MANIFESTATIONS OF ALLGROVE SYNDROME

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The authors describe two families with two affected siblings and a further unrelated patient with typical clinical features of Allgrove’s syndrome, who exhibit signs of multisystem neurological disease including hyperreflexia, muscle wasting, dysarthria, ataxia, optic atrophy, and intellectual impairment.

Allgrove or 4 “A” syndrome: an autosomal recessive ...

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Allgrove syndrome and motor neuron disease

Allgrove syndrome (OMIM 231550) is a rare autosomal recessive disease characterized by non-CAH primary adrenal insufficiency (non-CAH PAI), alacrima, and achalasia. It is caused by mutations in the AAAS gene. The syndrome is also associated with variable progressive neurological impairment and dermatological abnormalities.

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Triple A syndrome | Genetic and Rare Diseases Information ...

Allgrove syndrome (AS), also known as Triple-A syndrome is a rare autosomal recessive disease that has been mapped to chromosome 12q13. General manifestation of this chromosome include isolated glucocorticoid failure (hypoglycemia, weakness, fatigue, anorexia, nausea, vomiting, constipation, abdominal pain, diarrhea, salt carving,...

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Web of Knowledge / Web of Science: Pub Med / Medline ...
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Allgrove syndrome symptoms, causes, diagnosis, and treatment information for Allgrove syndrome (Achalasia - Addisonianism - Alacrimia syndrome) with alternative diagnoses, full-text book chapters, misdiagnosis, research treatments, prevention, and prognosis.

Allgrove syndrome - RightDiagnosis.com
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