

Allan-Herndon-Dudley Syndrome

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Abstract

Allan-Herndon-Dudley syndrome (AHDS) is an X-linked mental retardation syndrome. The frequency of AHDS among the general population with mental retardation (MR) is hard to accurately estimate. At present, three large families with AHDS have been studied which would indicate it might be as frequent as 1/10,000 males with MR. Although the responsible gene has been localized to the proximal long arm of the X chromosome (Xq13-q21), the cause of AHDS is not known at present. Affected males present in early childhood with hypotonia and a generalized paucity of skeletal muscle mass. The facies do not have any distinctive features; however, there is some elongation of the face, abnormal folding of the ears and bitemporal narrowing. Spastic paraplegia and joint contractures are evident in adults. Carrier females have normal intelligence and do not manifest any of the phenotypic features associated with AHDS.

Keywords

Allan-Herndon-Dudley syndrome, X-linked mental retardation syndrome, Xq13-q21

Definition

Allan-Herndon-Dudley syndrome (AHDS) is an X-linked mental retardation syndrome (MIM 309600). Linkage analysis in three families has placed the gene between DXS983 (Xq13) and DXS995 (Xq21). AHDS has hypotonia in childhood, ataxia, dysarthria, athetosis and spastic paraplegia.

Main symptoms

Affected males present in early childhood with hypotonia and a generalized paucity of skeletal muscle mass. The facies does not have any distinctive features; however, there is some elongation of the face, abnormal folding of the ears and bitemporal narrowing. Spastic paraplegia and joint contractures are evident in

adults. Dysarthria is also present as are hyperactive deep tendon reflexes. Affected males are severely cognitively impaired and most fail to develop speech or independent ambulation. Other development appears normal (stature, head circumference, genitalia).

Etiology

The cause of AHDS is not known at present. Although the responsible gene has been localized to the proximal long arm of the X chromosome (Xq13-q21), thus far, candidate gene testing has proved unsuccessful. Carrier females have normal intelligence and do not manifest any of the phenotypic features associated with AHDS.

Frequency

The frequency of AHDS in the general population with mental retardation (MR) is hard to accurately estimate. At present, three large families with AHDS have been studied which would indicate it might be as frequent as 1/10,000 males with MR.

Management of disease

Since the gene responsible for AHDS is not known, there exists no cure or treatment for the disorder. As most males are severely cognitively impaired, structured care outside the home is usually necessary. There appears to be no increase in health risks and affected males experience a relatively long life span. At present, there are no data that suggest early intervention would significantly improve the outcome for the males.

References

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