

1. Linn Dara Child and Adolescent Mental Health - Intellectual Disability Services.

INTRODUCTION

Xia-Gibbs syndrome is a neurological disorder arising from de novo mutations in the AHDC 1 gene on chromosome one identified by whole-exome sequencing. AHDC 1 gene provides instructions for making a protein with an unknown function.

Xia-Gibbs syndrome has an autosomal dominant pattern of inheritance.

Since its first description in 2014, fewer than 50 patients with Xia-Gibbs syndrome have been noticed in the literature, most of them children.

Core features of Xia-Gibbs syndrome includes hypotonia, mild to severe intellectual disability and delayed development amongst other symptoms.

AIMS & OBJECTIVES

An understanding of the possible behavioural phenotypes of Xia – Gibbs syndrome.

METHODS

Twelve-year-old girl with a background diagnosis of Moderate Intellectual disability, Attention Deficit Hyperactivity Disorder (ADHD) and Global Developmental Delay. She has an early childhood history of a chromosome sixteen mutation, bilateral cataracts, spinal bifida occulta, mild left hemiplegia, posterior fossa cysts and asthma. There is an unresolved absence seizure query.

She is the first known case of Xia - Gibbs syndrome in Ireland, formally diagnosed in December 2019.

She was referred to Child Psychiatry – Intellectual Disability services for challenging risk behaviours primarily at home but subsequently progressed to the school and respite settings.

These behaviours are in the form of hitting out, hair pulling, scratching and biting others. They are primarily triggered by her demands not being met and has resulted in significant physical harm.

There are reports of emotional lability, inattention and distractibility. She has difficulties maintaining sleep and seeks out food on a constant basis. She seeks out engagement with adults routinely and constantly looks for reassurance from attachment figures. She struggles to work independently and functions best with one to one adult attention and scaffolding. She has limited awareness of social boundaries and requires regular supervision.

She is the first live birth of her parents after seven previous miscarriages.

EVALUATION (RESULTS)

Our clinical impression is that of a young girl with an anxious attachment behavioural pattern in a background of Xia - Gibbs syndrome, Moderate Intellectual disability, ADHD, a complex medical background and a significant risk history.

DISCUSSION

This case illustrates certain known behavioural patterns in children with neuro-developmental disorders particularly those functioning in the moderate - profound degree. However, Xia-Gibbs syndrome is a rare genetic disorder with well documented physical features but may also have specific behavioural phenotypes.

This pioneer case in Ireland will hopefully inform future research on the clinical presentation and management of Xia-Gibbs syndrome particularly in children with impairing behavioural difficulties.

OTHER INFORMATION

No sponsorship or any conflict of Interest to declare.

Written informed consent obtained from child parents.

REFERENCES

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