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Xia-Gibbs Syndrome: A Psychiatric Perspective

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Dear Editor,

Xia-Gibbs syndrome is a neurological disorder arising from de novo mutations in the AHDC 1 gene on chromosome one[1]. These mutations can be identified by whole - exome sequencing. AHDC 1 gene provides instructions for making a protein with an unknown function[2]. 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[3], most of them children. Core features of Xia-Gibbs syndrome includes hypotonia, mild-severe intellectual disability and delayed development amongst other symptoms[4].

This letter discusses the psychiatric presentation of the only confirmed case of Xia-Gibbs syndrome in Ireland formally diagnosed in December 2019.

It is a case of a twelve-year-old girl with a background diagnosis of Moderate Intellectual disability, Attention Deficit Hyperactivity Disorder (ADHD) and a history of global developmental delay. She has a background medical history of a chromosome sixteen mutation, bilateral cataracts, spinal bifida occulta, mild left hemiplegia, posterior fossa cysts, asthma and an unresolved absence seizure query.

She was first referred to Child and Adolescent Psychiatry - Intellectual Disability services for challenging risk behaviours predominantly at home and had progressed to 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. There is a paternal history of similar chromosome sixteen mutation. She has a younger sibling who is of typical development.

She impressed as a young girl with anxious attachment behavioural patterns in a background of Xia-Gibbs syndrome, Moderate Intellectual disability, Attention Deficit Hyperactivity Disorder (ADHD), a significant risk history and with complex neurological difficulties.

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

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

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