CASE REPORT

Smith-Magneis syndrome: behavioural phenotype mimics ADHD

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SUMMARY

A mentally retarded 7-year-old male child presented with inattention and hyperactivity which was initially diagnosed as attention deficit hyperactivity disorder (ADHD). However, a careful evaluation of symptomatology along with clues provided by specific features of facial dysmorphism in this case along with genetic testing clinched the diagnosis of Smith-Magneis syndrome the behavioural phenotype of which closely resembles ADHD.

BACKGROUND

Smith-Magneis syndrome is a genetic disorder characterised by intellectual disability accompanied by sleep disorders, hyperactivity, attention deficit, self-harm and facial dysmorphic features including broad, flat middle part of the face, flat and broad nasal bridge, hypoplasia of the cheeks, telecanthus and prognathism. The associated features include ocular, hearing abnormalities and epilepsy.¹ This case intends to provide clinicians' knowledge regarding the behavioural phenotype of this rare genetic syndrome that might mimic psychiatric illnesses like attention deficit hyperactivity disorder (ADHD) and hence has definite diagnostic and management implications.

CASE PRESENTATION

A 7-year-old male child was brought to psychiatric outpatient department of our tertiary care centre presenting with inattention and hyperactivity. This was their first visit to a mental health physician. The parents reported that the child would be easily distracted moving from one task to another, would not be interested in any activity more than a brief time, would not listen to people talking to him more than a few minutes, would appear 'on the go', would be restless always moving from one place to another and unmanageable on most of the occasions. In addition, the child would engage in unusual behaviours including repeated self-hitting, self-biting as well as aggressive behaviour with frequent temper tantrums.

The parents reported that the child was born by normal delivery and there were no significant perinatal complications except difficulty in feeding and the baby appearing flabby. Though the child was possibly low birth weight at the time of birth (details unavailable), the child was currently in the normal weight range. They also reported of retarded developmental milestones in all the four domains: fine motor skills, gross motor skills, social and language milestones. Currently at the age of 7 years, the child was able to walk with support,

talk individual words (not in the form of even short sentences) and identify family members differentiating from strangers.

On examination, the child exhibited the following features of facial dysmorphism; square-shaped face, flat and broad nasal bridge, telecanthus and prognathism. The child also showed definite signs of hearing loss and on examination thought his vision appeared normal. The IQ of the child was assessed using Malin's intelligence scale for Indian children (MISIC).² The MISIC score was 47 indicating moderate mental retardation.

The indicators of facial dysmorphic features, specific behavioural abnormalities alerted the treating team regarding the possibility of rare genetic syndromes, possibly Smith-Magneis syndrome on careful review of literature. The other possibilities that we considered included velocardiofacial syndrome, Di George's syndrome, Kleeftstra syndrome, William's syndrome and Prader-Willi syndrome that share many of the features with Smith-Magneis syndrome.³

INVESTIGATIONS

Fluorescent in situ hybridisation (FISH) revealed deletion of chromosome 17p11.2 that clinched the diagnosis of Smith-Magneis syndrome.⁴ An otolaryngological and ophthalmological examination was ordered considering the high frequency of such abnormalities in Smith-Magneis syndrome. Renal ultrasound, echocardiogram, thyroid function tests, fasting lipid profile and spine radiographs were ordered that did not demonstrate any specific findings. Thyroid function tests revealed hypothyroidism and thyroxine supplement was started after endocrinology consultation. The parents were referred for genetic counselling.

DIFFERENTIAL DIAGNOSIS

Although an independent diagnosis of ADHD was considered, a thorough literature search indicated that ADHD symptoms were an integral part of the Smith-Magneis syndrome. The peculiarities of the case included sleep abnormalities, seizure disorder and features of autistic spectrum disorders that are very common and characteristic of Smith-Magneis syndrome.

TREATMENT

After discussion, it was decided to start the patient on low-dose risperidone (1 mg/day) for management of behavioural symptoms in the absence of empirical evidence favouring any particular drug.

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To cite: Gnanavel S. *BMJ Case Rep* Published online: [please include Day Month Year] doi:10.1136/bcr-2013-201766

A comprehensive management plan was formulated for the patient that included speech therapy, use of communication programme and behavioural management strategies.

OUTCOME AND FOLLOW-UP

However, the patient unfortunately was lost to follow-up after a few sessions due to logistics issues and the parents could not be contacted further.

DISCUSSION

Smith-Magenis syndrome is among the rarer genetic syndromes characterised by distinctive physical features, developmental delay, intellectual impairment and behavioural abnormalities.

Learning points

- ▶ Peculiar behavioural phenotypes of rare genetic disorders can mimic common psychiatric illnesses.
- ▶ Comprehensive management plan on biopsychosocial lines is integral for the management of children with such genetic syndromes.
- ▶ Significant psychiatric and physical comorbidity are associated with genetic syndromes.

Behavioural phenotypes of these rare syndromes are distinct and provide clues for diagnosis even in the absence of genetic testing facilities. Many of these behavioural symptoms can mimic psychiatric illnesses such as ADHD and psychosis thereby posing a diagnostic dilemma and issues in management. A comprehensive management plan composed of psychotropic medication, behavioural measures, specialised education, evaluation of possible medical comorbidity and management of the same can prove tough in resource-scarce settings in developing countries like ours though every effort should be made towards the same.

Contributors SG was involved in clinical care, conceptualisation and drafting of manuscript.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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