PURA Syndrome: A Spesific Phenotype

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INTRODUCTION

PURA syndrome is caused by heterozygous pathogenic sequence variants in PURA gene has become an increasingly diagnosed disorder after the introduction of the new generation sequencing methods. It is assumed to have a specific phenotype. (Figure 1) It is an autosomal dominant inherited genetic neurodevelopmental disorder with cognitive impairment, delay in motor skills such as walking and speech. Expressive language skills are often more severely affected than receptive language skills, and a large proportion of affected individuals are unable to speak. Delayed walking is evident in patients and consequently most of them are unable to walk. Dysphagia, hypersomnolence, hypothermia, apnea attacks, hypoventilation, hypotonia, feeding difficulties are common and more prominent in the infantile period. Respiratory problems usually disappear after infancy.

OBJECTIVES

We aimed to present 3 patients whose diagnosed PURA syndrome by whole exome sequencing (WES).

MATERIALS & METHODS

Whole exome sequencing applied in 3 cases from different families who were followed up due to severe speech and cognitive impairment, hypotonia and dysmorphic findings and were not clinically diagnosed.

Genetic Analyses	c.692T>G (p..Phe231Cys) heterozygous variant in PURA gene	c.159dupG heterozygous variant in PURA gene	c.677_678del TG heterozygous variant in PURA gene
Age, Gender	3y, male	13y, male	4y, female
Growth			
Short Stature	No	Yes 115 cm (-4,92 SD)	Yes 87 cm (-4,64 SD)
Neonatal Problems			
Hypotonia	Yes	Yes	Yes
Feeding Difficulties	Yes	Yes	Yes
Reflux	Yes	Yes	No
Breathing Problems	Yes	No	Yes
Hypersomnolence	No	No	Yes
Hypothermia	No	No	No
Neurological Abnormalities			
Cognitive Impairment	Yes	Yes	Yes
Hypotonia	Yes	Yes	Yes
Stereotypy	No	No	No
Epilepsy	No	Yes	No
Movement Disorder	No	No	No
Brain Abnormalities	No	No	thin corpus callosum
Skeletal Abnormality			
Scoliosis	No	Yes	No
Hip Dysplasia	No	No	Yes
Hyperlaxity	No	Yes	No
Cardiac Abnormality	No	No	No
Urogenital Abnormality	No	No	No
Ophthalmological Abnormality	No	Yes	No

Table 1. : Clinical features of three patients with PURA syndrome

RESULTS

Pathogenic variations were found in the PURA gene as a result of WES. The clinical features of three cases are given in Table 1.

CONCLUSIONS

It is emphasized that the clinical phenotype of PURA syndrome is specific and this syndrome can be suspected clinically before genetic examination. The characteristics of our cases support this specific phenotype.

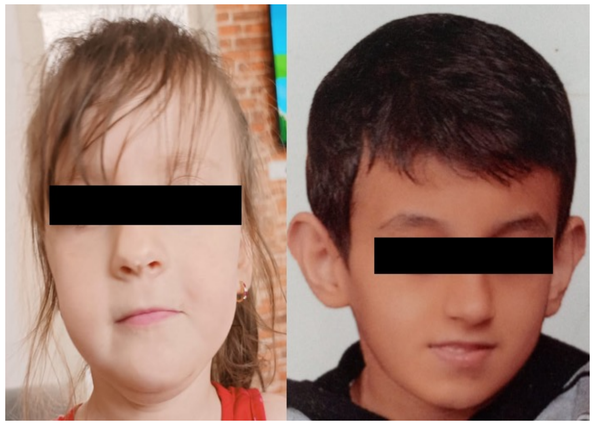

Figure 1. PURA syndrome phenotype

KEYWORDS

Hypotonia, speech impairment, PURA syndrome