

The second case of Noonan Syndrome: The association with unique multiple Cardiac Defects

Aamir Jalal Al Mosawi

Advisor in Pediatrics and Pediatric Psychiatry Children Teaching Hospital of Baghdad Medical City, Head, Iraq Headquarter of Copernicus Scientists International Panel Baghdad, Iraq

Corresponding author: Aamir Jalal Al Mosawi, Advisor in Pediatrics and Pediatric Psychiatry Children Teaching Hospital of Baghdad Medical City, Head, Iraq Headquarter of Copernicus Scientists International Panel Baghdad, Iraq.

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Abstract

Background: Noonan syndrome is a heterogeneous congenital disorder that can occur sporadically or inherited as an autosomal dominant disorder. It is characterized by a wide spectrum of phenotypic abnormalities that vary greatly in range and severity, and two patients with Noonan syndrome may have two different characteristic features. In many patients the syndrome characterized by craniofacial abnormalities including low set ears, hypertelorism, congenital heart defect, short stature, and undescended testes. Although pulmonary stenosis is the commonly associated congenital cardiac defects, a variety of cardiac defects may occur in this syndrome. Atrial septal defect, and patent ductus arteriosus are other well-recognized cardiac defects of this syndrome. The diagnosis of Noonan syndrome is entirely clinical as there is no specific diagnostic available

Materials and methods: A ten month old boy who was referred to the pediatric neuropsychiatric clinic of the Children Teaching Hospital of Baghdad Medical City because of developmental delay associated with multiple congenital abnormalities was studied.

Results: The boy had growth and developmental retardation, low set ears, hypertelorism, and smooth philtrum, undescended testes. Echocardiography showed interatrial septum, small atrial septal defect and closing patent ductus arteriosus.

Conclusion: Noonan syndrome was previously reported only in one girl from Iraq. The first Iraqi boy with Noonan syndrome is reported in association with unique cardiac defects. The previously reported case and the case in this report demonstrates the variability of the phenotype of this syndrome.

Keywords: noonan syndrome; cardiac defects; interatrial septum; iraq.

Introduction

Noonan syndrome is a heterogeneous congenital disorder that can occur sporadically or inherited as an autosomal dominant disorder. It is characterized by a wide spectrum of phenotypic abnormalities that vary greatly in range and severity, and two patients with Noonan syndrome may have two different characteristic features. In many patients the syndrome characterized by craniofacial abnormalities including low set ears, hypertelorism, congenital heart defect, short stature, and undescended testes. Although pulmonary stenosis is the commonly associated congenital cardiac defects, a variety of cardiac defects may occur in this syndrome. Atrial septal defect, and patent ductus arteriosus are other well-recognized cardiac defects of this syndrome. The diagnosis of Noonan syndrome is entirely clinical as there is no specific diagnostic available [1-5].

Materials and methods

A ten month old boy who was referred to the pediatric neuropsychiatric clinic of the Children Teaching Hospital of Baghdad Medical City because of developmental delay associated with multiple congenital abnormalities was studied.

Results

The boy was experiencing growth and developmental delay and was unable to sit. He had facial dysmorphic features consisting of low set ears, hypertelorism, and smooth philtrum (Figure-1). Examination of the heart suggested the presence of congenital heart disease and his testes were not palpable in the scrotum. Echocardiography showed interatrial septum, small atrial septal defect and closing patent ductus arteriosus. Abdominal

ultrasound showed ectopic intra-abdominal testes located at the inlet of the inguinal, and both testes measures 10 mm in diameter (Figure-2). Chromosomal analysis showed normal female karyotype.

Parents were consanguineous and family history was negative for similar condition.



Figure-1: The infant had facial dysmorphic features consisting of low set ears, hypertelorism, and smooth philtrum



Figure-2: Abdominal ultrasound showed ectopic intra-abdominal testes located at the inlet of the inguinal, and both testes measures 10 mm in diameter

Discussion

During the 1940s, 1950s, and early 1960s Noonan syndrome was considered to be Turner syndrome occurring in males [6-11]. However, at the meeting of the Mid-Western Society for Pediatric Research in 1962, Jacqueline A. Noonan (Figure-3) presented nine patients who had growth retardation, hypertelorism syndrome of valvular pulmonary stenosis and other congenital abnormalities. In 1968, Jacqueline A. Noonan published reporting 19 patients including males and females [1, 2]. In 1971, Hellebusch called the condition Noonan syndrome [12]. Noonan syndrome was previously reported only in one girl from

Iraq. She had also a sporadic form of Noonan with predominant cerebral manifestations and without congenital heart defect [5]. The previously reported case and the case in this report demonstrates the variability of the phenotype of this syndrome.

Conclusion

The first Iraqi boy with Noonan syndrome is reported in association with unique cardiac defects.



Figure-3: Jacqueline Noonan, a pediatric cardiologist at the University of Iowa

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- 2- The author has the copyright of the sketch in figure-3.

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