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Case Report
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The Case Number 104 of Sanjad Sakati Richardson Kirk Syndrome

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Abstract

The syndrome of severe growth retardation, mental retardation, and chronic hypocalcemia caused by hypoparathyroidism was first reported by Sanjad, Sakati, and Abu-Osba in 1988.

Sanjad, Sakati, and Abu-Osba presented a part of a full description of the syndrome in five infants at the 58th Annual Meeting of the Society for Pediatric Research, Washington DC, May 1988. Later, they published a more completed description of the syndrome in 1991. The paper of 1991 which was authored by more authors and included twelve patients.

However, the syndrome was first fully described in 1990 by Ricky J Richardson from the Sick Children Hospital of Great Ormond Street in London, and Jeremy MW Kirk from St Bartholomew's Hospital in London.

Richardson and Kirk emphasized that this association of a previously un-described congenital anomalies represented a new syndrome that was observed in eight children of Middle Eastern origin. They thought that early recognition of this rare disorder may decrease the associated morbidity and mortality.

The total number of the reported patients with Sanjad-Sakati-Richardson-Kirk syndrome is 103.

Sanjad-Sakati-Richardson-Kirk syndrome has not been reported before in Iraq. The main aim of paper is to describe the first case of this syndrome in Iraq which is the case number 104 in the world.

Keywords: Sanjad-Sakati-Richardson-Kirk syndrome; Hypoparathyroidism; Chronic hypocalcemia

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Introduction

The syndrome of severe growth retardation, mental retardation, and chronic hypocalcemia caused by hypoparathyroidism was first reported by Sanjad, Sakati, and Abu-Osba in 1988 [1].

However, the syndrome was first fully described in 1990 by Ricky J Richardson from the Sick Children Hospital of Great Ormond Street in London, and Jeremy MW Kirk from St Bartholomew's Hospital in London. Richardson and Kirk emphasized that this association of a previously un-described congenital anomalies represented a new syndrome that was observed in eight children of Middle Eastern origin. They thought that early recognition of this rare disorder may decrease the associated morbidity and mortality [2].

Sanjad, Sakati, and Abu-Osba presented a part of a full description of the syndrome in five infants at the 58th Annual Meeting of the Society for Pediatric Research, Washington DC, May 1988. Later, they published a more completed description of the syndrome in 1991. The paper of 1991 which was authored by more authors, and included twelve patients [3].

The total number of the reported patients with Sanjad-Sakati-Richardson-Kirk syndrome is 103 [4,5].

Thirty eight patients with Sanjad-Sakati-Richardson-Kirk syndrome were reported during the 1990's including eight patients reported by Richardson and Kirk, 1990; twelve patients reported by Sanjad et al. [3], one patient reported by Kalam and Hafeez in 1992, six patients reported by Hershkovitz, 1995. Two patients reported by Parvari, 1998; One patients reported by al-Gazali and Dawodu, 1997; and eight patients reported by Diaz, 1999 [4].

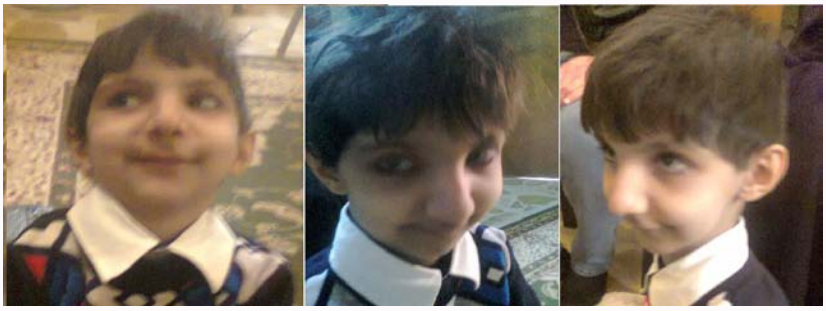

Figure 1: The boy had deep set eyes, thin lips, long philtrum, and beaked nose.

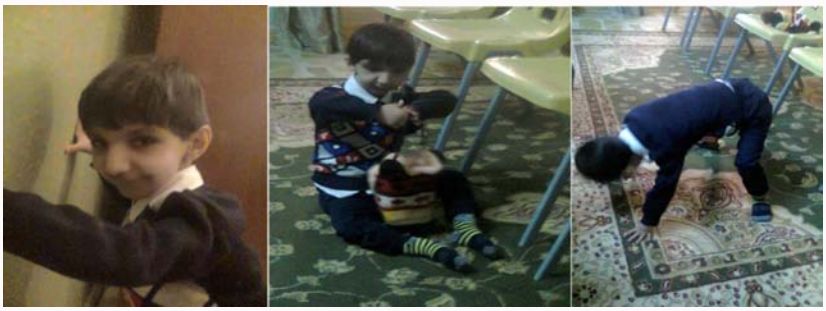

Figure 2: During December, 2018m the boy could stand with some assistance holding the wall. However, the boy was unable to stand from sitting alone without assistance or holding something.

Twenty five patients with Sanjad-Sakati-Richardson-Kirk syndrome were reported during the 2000s including one Saudi girl reported by Al-Malik, 2004; two Saudi siblings reported by Hellani, 2004; one girl reported by Courtens, 2006; and twenty one patients reported by Naguib, 2009 [4].

Forty patients with Sanjad-Sakati-Richardson-Kirk syndrome were reported after 2010 including eight Jordanian patients reported by Albaramki, 2012; One Tunisian patient reported by Kerkeni, 2015; One Jordanian patients reported by Ajarmeh and Al Tamimi, 2018; One patient reported by Ryabets-Lienhard, 2018; and twenty nine Arab patients from Iran reported by Aminzadeh, 2018 [4].

Sanjad-Sakati-Richardson-Kirk syndrome has not been reported before in Iraq. The main aim of this chapter is to describe the first case of this syndrome in Iraq which is the case number 104 in the world.

Case Presentation

A. Cesar was born during the year 2010 and was seen early during the year 2018 because of resistant hypocalcemia and seizures. The parents were relatives and healthy and had another four children, three were normal and doing well at school and one child died during early infancy after developing hypocalcemia and seizures, and septicemia was considered as the cause of death.

Before referral, the hypocalcemia was treated with low dose vitamin D followed with low dose of one alpha hydroxycholecalciferol (2 drops daily) and the boy continued to have seizures.

Parathyroid hormone level was within normal (23 pg/ml; Normal: 10 to 65 pg/ml) even when serum calcium was below 6.5 mg/dL (Normal: 9 to 11 mg/dL).

The calcium level remained below 7 mg/dL, but it was ignored by the treating physicians, and electroencephalography was performed, and showed frequent focal epileptic discharges more prominent in central leads and mild slowing of cerebral activity.

The child was treated with anticonvulsant medications mostly sodium valproate which reduced the seizures but didn't stop.

When the child was first seen, he had severe growth retardation (Body weight: 8 Kilograms), and marked developmental delay.

He was not able to stand and was crawling with difficulty. He was not saying any words and not responding to simple commands.

The child was treated with one alpha hydroxycholecalciferol 1 μ twice daily and increased to 4 μ daily over three months. Calcium was maintained above 8 mg/dL. Seizures stopped and sodium valproate was stopped. Intensive nutritional support was also provided.

After controlling the hypocalcemic seizures and improving the child general condition and nutritional status, the disorder of the child was studied more carefully during December, 2018.

The child had dysmorphic features including (Figure 1):

- Deep set eyes
- Beaked nose
- Thin lips
- Long philtrum
- Large floppy ears
- Micrognathia

Despite the improvement in growth with his weigh became 10 Kilograms, and some improvement in development, the boy was considered to have mental retardation. His speech was initiated, and he was saying many words. He was crawling much easier.

He was climbing the chair to stand holding the chair and could

stand with some assistance holding the wall. However, he was unable to stand from sitting alone without assistance or holding something (Figure 2).

Discussion

There is often limited professional knowledge, experience and awareness of the manifestations of very rare genetic disorders, and their most appropriate management, because of the small number of patients having each one of them. It is generally difficult to diagnose a rare disease or disorder because it is impracticable for doctors to be familiar with thousands of rare conditions. However, early diagnosis of rare genetic disorders plays a vital role in preventing the disorder through appropriate genetic counseling. Sometimes early recognition of a rare disorder can help in reducing the associated morbidity and mortality [4]. In this case, the parents was not provided with appropriate genetic counseling because the early consulted physicians couldn't make the correct diagnosis or them nor made the appropriate referral. Late referral also delayed the provision of more appropriate treatment.

Syndromes in medicine are often named after the physician or group of physicians that discovered them or initially provided the full clinical picture or the best description of the syndrome. However, many of the rare syndromes have been described by physicians in many areas of the world before the era of the internet which has been associated with easy access to clinical reports throughout the world. Unfortunately, sometimes, when naming a syndrome, the enormous role of the physicians who provided the first full published description was unfairly ignored [6]. In this paper, an attempt was made to give the appropriate credit for the authors who provided the first full description of the syndrome.

Acknowledgement

This paper was presented at the Royal College of Paediatrics and Child Health 9th Europaediatrics Congress (13 to 15 June Dublin Ireland, 2019) and the abstract was published in [Archives of Disease in Childhood 2019; 104(Suppl 3):A355].

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