SunKrist Publishing
Accelerate Scientific Knowledge in Open Access

SunKrist Clinical and Medical Case Reports Journal

Case Presentation *Volume: 1, Issue: 1* *Scientific Knowledge*

Coffin Siris Syndrome with Significant Autistic Features

Al-Mosawi AJ[1,2*]

[1]Department of Pediatrics and Pediatric Psychiatry, Children Teaching Hospital of Baghdad Medical City, Iraq

[2]Head, Iraq Headquarter of Copernicus Scientists International Panel, Iraq

1. Abstract

Coffin Siris Syndrome is a rare genetic dysmorphic disorder that was first described by Coffin and Siris in 1970. It is associated with mental retardation and distinctive coarse facial features including wide nose, wide mouth with thick everted upper and lower lips, thick eyebrows and lashes, hirsutism/ hypertrichosis. Sparse scalp hair particularly in the temporal areas.

Coffin Siris syndrome has been reported only once in Iraq. The first Iraq patient with Coffin Siris syndrome (Al Mosawi AJ, 2006) was also the first patient in the Arab. The occurrence of significant autistic features in form of pervasive developmental disorder or autism has been very infrequently reported in association with Coffin Siris syndrome. This aim to report which is most probably the seventh case of Coffin Siris Syndrome associated with significant autistic features. The case is the second case of this syndrome in Iraq which is also the case number 170 in the world.

2. Introduction

Coffin Siris Syndrome is a rare genetic dysmorphic disorder that was first described by Coffin and Siris in 1970. It is associated with mental retardation. Levy and Baraitser thought that there had been thirty-one reported cases by 1991.In addition to mental retardation, patients had [1-4]: 1-Distinctive coarse facial features including: A-Wide nose. B-Wide mouth with thick everted upper and lower lip. C-Thick eyebrows and lashes. 2-Hirsutism/ hypertrichosis. 3-Sparse scalp hair particularly in the temporal areas 4-Feeding difficulties, slow growth, hypotonia.

The facial features typically coarsen with time and the characteristic facies may not be evident or obvious early during life. Patients having short fifth digits with hypoplastic or absent nails were considered to have the classical syndrome. However, fifth finger nail/distal phalanx hypoplasia or aplasia was not considered mandatory to make the diagnosis [1-4]. The occurrence of significant autistic features in form of pervasive developmental disorder or autism has been very infrequently reported in association with Coffin Siris syndrome [4-6]. This aim to report which is most probably the seventh case of Coffin Siris Syndrome associated with significant autistic features.

A total of 169 cases have been reported in the literature including the eight patients reported by Schrier et al. (2012) [4,7]. The case is the second case of this syndrome in Iraq case number 170 reported in the literature.

3. Case Presentation

Z.K was first seen at the age of twelve years during

*Corresponding author: Al-Mosawi AJ, Department of Pediatrics and Pediatric Psychiatry, Children Teaching Hospital of Baghdad Medical City, Iraq, E-mail: almosawiAJ@yahoo.com

Received Date: August 19, 2019; **Accepted Date**: August 26, 2019; **Published Date**: August 29, 2019

November, 2018 because of very poor feeding, severe malnutrition, difficulty in walking and markedly diminished responsiveness to the external environment. She had evidence of marked wasting in the lower limbs and chest.

The girl had severe form of mental retardation with global delay in motor development and didn't develop speech despite normal hearing and also didn't develop the normal adaptive behaviors of controlling bowels and self-care.

Early during childhood she was rather active, but during the previous months she experienced reduction in her activity. Her parents were normal and had three other normal children.

The girl also had significant autistic features including not responding to her name and didn't show any eye contact.

The girl had feeding difficulties, slow growth and hypotonia in addition to several physical abnormalities

Figure 1: The girl had distinctive facial features including thick eyebrows, depressed and wide nasal bridge, large mouth with thick everted upper and lower lips, hypertrichosis and sparse scalp hair particularly in the temporal regions.

including dysmorphic facial features. She has: Thick eyebrows, Depressed and wide nasal bridge, Low set ears, Large mouth with thick everted upper and lower lips, relatively long fingers and rather spindle in shape, Hypertrichosis, Sparse scalp hair particularly in the temporal regions, Generalized Hirsutism and facial Hirsutism with a thin moustache.

Abdominal ultrasound performed during this year showed normal size kidneys.

4. Discussion

Al Mosawi AJ (2006) reported the first Iraqi patient with Coffin Siris syndrome when he was head of the department of Pediatrics at Al Kadhymiyia University Hospital and the case was presented at several conferences. The patient was first case of the syndrome in the Arab [2].

The occurrence of significant autistic features in form of pervasive developmental disorder or autism has been very infrequently reported in association with Coffin Siris syndrome [4-6].

Hersh and colleagues reported the occurrence of childhood autism in a girl with Coffin Siris Syndrome. Swillen et al. (1995) [6] from the University of Leuven in Belgium described twelve children and adolescents with Coffin-Siris syndrome (Nine girls and three boys). The patients' age was between 2.5 to19 years.

Three patients had mild mental retardation and nine patients had moderate mental retardation.

Speech onset was severely retarded with little interest in language. In the older group (seven patients aged 7 to 19 years), language comprehension was appropriate to the mental level.

Aggressive disturbed behavior was observed particularly in the youngest children, whereas mixed disturbed behavior was observed in the oldest patients. Five patients developed symptoms of pervasive developmental disorder and in two of them the symptoms were in the pathological range. Obsessive interests, strong dependence on patterns and rituals and unusual fears were characteristic behavioral problems when patients got older.

The patient described in this paper most probably represents the seventh case of Coffin Siris Syndrome associated with significant autistic features.

References

1. Coffin GS, Siris S. Mental retardation with absent fifth fingernail and terminal phalanx. Am J Dis Child. 1970; 119: 433-439.

2. P Levy, M Baraitser. Coffin-Siris syndrome. J Med Genet. 1991; 28: 338-341.

3. Al Mosawi AJ. Genetic drift. Letter from Baghdad: Coffin-Siris syndrome in a girl with absent kidney. Am J Med Genet A. 2006; 140: 1789-1790.

4. Al-Mosawi AJ. Coffin Siris syndrome. 1st ed., Saarbrücken; LAP Lambert Academic Publishing: 2018.

5. Hersh JH, Bloom AS, Weisskopf B. Childhood Autism in a female with Coffin Siris Syndrome. J Dev Behav Pediatr 1982; 3: 249-252.

6. Swillen A, Glorieux N, Peeters M, Fryns JP. The Coffin-Siris syndrome: data on mental development, language, behavior and social skills in 12 children. Clin Genet. 1995; 48: 177-182.

7. Schrier SA, Bodurtha JN, Burton B, Chudley AE, Chiong MA, D'avanzo MG, et al. The Coffin-Siris syndrome: a proposed diagnostic approach and assessment of 15 overlapping cases. Am J Med Genet A. 2012; 158: 1865-1876.

Citation: Al-Mosawi AJ. Coffin Siris Syndrome with Significant Autistic Features. SunKrist Clin Med Case Rep J. 2019; 1: 1002.