Article published online: 2021-08-09

CASE REPORT

Kenny-Caffey syndrome type 1

Tony El Jabbour, Tarek Abboursheid[1], Mohammad Baraa Keifo[1], Ismael Maksoud[2], Diana Alasmar[3]

Department of Anatomic Pathology, Faculty of Medicine, Lebanese University, Lebanon, [1]Department of Pediatrics, Faculty of Medicine, [2]Pediatrics University Hospital, [3]Pediatric Endocrinology and Metabolic Diseases Unit, University Children Hospital, Faculty of Medicine, Damascus University, Damascus, Syria

Access this article online

Website: www.avicennajmed.com

DOI: 10.4103/2231-0770.133340

Quick Response Code:

ABSTRACT

Kenny-Caffey syndrome type 1 is a rare hereditary skeletal disorder. We present here a documented case of a 7-month-old girl with the characteristic symptoms of growth retardation, dysmorphic features, and hypoparathyroidism.

Key words: Hypoparathyroidism, Kenny-Caffey, Sanjad-Sakati

INTRODUCTION

Kenny-Caffey syndrome type 1 (KCS 1) is a rare autosomal recessive skeletal disorder. It's manifested by growth retardation, thickening of the long bones, medullary stenosis (thin marrow cavities) and dysmorphic features, in addition to recurrent episodes of hypocalcemia caused by hypoparathyroidism which occur in early life.[1]

This disease has been observed almost exclusively in the Middle East. The only data available about its prevalence is in Saudi Arabia and is estimated between 1:40,000 and 1:100,000 live births.[2] Although in Syria, there is no conclusive data. This article presents a case of a 7-month-old Syrian girl who showed all of the characteristics of KCS 1.

CASE REPORT

A 7-month-old girl was admitted to the children hospital in Damascus for the first time complaining of failure to thrive. She was the sixth-born child to consanguineous parents who had their first girl child died 24 days after birth following a story of jaundice, whereas the other siblings were normal. The baby was born after a full-term pregnancy with no considerable maternal health issues. Her weight was 1500 g afterbirth (below 1% percentile following the WHO child growth standards), 1700 g at the end of the second month and 2950 g on admission and remained below 1% percentile throughout. The parents mentioned the occurrence of a convulsion episode 10 days prior to the admission that lasted for 20 min.

Examination revealed noticeable dysmorphic features including beaked nose tip, thin lips, micrognathia, and prominent forehead. She also exhibited a right cornea cloudiness and alternative strabismus in both eyes. The head circumference was 36.5 cm (below 2% percentile, following the WHO child growth standards). Developmentally, she had started to recognize her mother since 2 months and lately, she's become partially able to roll over from pronation to supination.

Laboratories revealed normal blood count, hemoglobin 10.9 g/dl, and mean corpuscular volume 87 fl. All plasma electrolytes were normal except for: Calcium 6.6 g/dl, phosphorus 11.1 g/dl and magnesium 1.8 g/dl (normal values of magnesium being between 1.9 and 2.5). Serum parathyroid hormone (PTH) was extremely low 7.9 pg/ml and 25-hydroxy vitamin D was 121 ng/ml. Liver functions test and urinalysis was normal.

Plain X-ray images showed absence of diploic space in the skull bones [Figure 1] and cortical thickening and medullary stenosis of the long bones [Figure 2]. Abdominal ultrasonography and echocardiogram were normal.

Address for correspondence: Dr. S Tony El Jabbour, Saint Elie Street, Joseph Attieh's Building, 4th Floor, Dekweneh, Lebanon.
E-mail: tonijab@hotmail.com

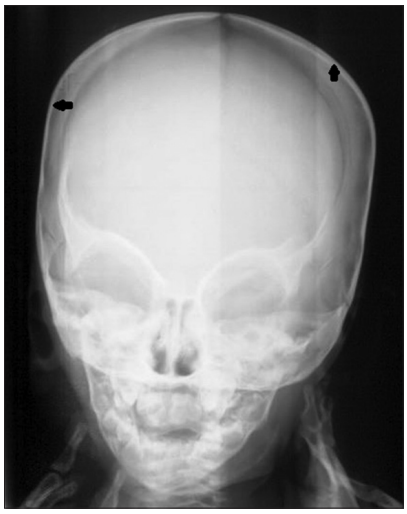

Figure 1: Skull X-ray showing absent diploic space in the skull bones

Ophthalmologic consult reported no evidence for cysteine crystals and the cloudiness of the right cornea can be attributed to a previous inflammatory event.

In respect of the patient's family history, her cousin had been admitted to the same hospital due to a growth failure with low plasma PTH level and somewhat similar findings. Genetic consult suggested a possible hereditary disorder with autosomal recessive type of inheritance.

The patient was readmitted to the hospital at the age of 16 months for reevaluation and to adjust the doses of the medications. No improvement in her growth was remarked with 4 kg of weight and a mental age didn't exceed 7 months. The investigations results were similar to those obtained previously. Hormonal tests and celiac disease screening were performed to interpret poor growth and they were both negative.

DISCUSSION

Kenney-Caffey is a rare syndrome, reported firstly by Kenny and Linarelli in 1966;[3] Caffey described the radiological characteristics in 1976;[4] Lee detailed the facial features in 1983.[5]

We are presenting the case of 7-month-old girl with refractory failure to thrive hypoparathyrodism, dysmorphic facies, characteristic bone changes and mild mental retardation. Consanguinity and a cousin with similar findings were found in the family. Laboratory studies showed hypocalcemia, hyperphosphatemia, and mild hypomagnesemia. All those findings are in the perspective of KCS 1 (OMIM 244460).[1]

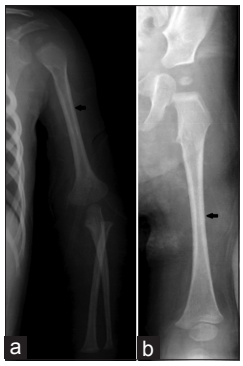

Figure 2: Plain X-ray images showing cortical thickening and medullary stenosis of the long bones: (a) Humerus (b) Femur

The differential diagnosis of hypoparathyrodism in this age includes: KCS 2, Sanjad-Sakati syndrome, Di-George syndrome and transient hypoparathyrodism of diabetic mother.

Kenny-Caffey syndrome type 2 (OMIM 127000), is an autosomal dominant disorder, sharing some of those clinical and radiological finding. However, it is distinguished by corneal and retinal calcifications, congenital cataracts, normal mentality, deficient T-cell immunity, and transient hypoparathyrodism.[6] Although corneal clouding confused initially our case, ophthalmology consult ruled out KCS 2.

Another differential diagnosis is Sanjad-Sakati syndrome (OMIM 241410) an autosomal recessive disorder characterized by hypoparathyrodism, retardation and dysmorphism.[6-8] It is thought that subclavian steal syndrome (SSS) and KCS 1 are due to a mutation in the tubulin specific chaperone E gene[9] suggesting that both are allelic disorders if not the same. The typical findings of cortical thickening and medullary stenosis were not described in SSS,[8] making the diagnosis of KCS 1 more probable.

Di-George syndrome may present with hypoparathyrodism, growth retardation and abnormal facial features. Chest radiograph showing normal thymic shadow and normal echocardiogram and ultrasonogram abdomen distinguished our case from Di-Georges syndrome.

Hypoparathyrodism can present in the offspring of a diabetic mother, but it is mild and transient. Osteopetrosis and pycnodysostosis are characterized by increased bone thickness and density, but they lack hypoparathyrodism.

The diagnosis of KCS 1 was then established depending on the previous data given that no other possible diagnosis

matched the findings, but the confirmation with a specific genetic test was not done.

ACKNOWLEDGMENT

The authors would like to thank Al Hamid Naji MD, Diana Alasmar MD.

REFERENCES

1. Naguib KK, Gouda SA, Elshafey A, Mohammed F, Bastaki L, Azab AS, *et al*. Sanjad-Sakati syndrome/Kenny-Caffey syndrome type 1: A study of 21 cases in Kuwait. East Mediterr Health J 2009;15:345-52.
2. Franceschini P, Testa A, Bogetti G, Girardo E, Guala A, Lopez-Bell G, *et al*. Kenny-Caffey syndrome in two sibs born to consanguineous parents: Evidence for an autosomal recessive variant. Am J Med Genet 1992;42:112-6.
3. Kenny FM, Linarelli L. Dwarfism and cortical thickening of tubular bones. Transient hypocalcemia in a mother and son. Am J Dis Child 1966;111:201-7.
4. Caffey J. Congenital stenosis of medullary spaces in tubular bones and calvaria in two proportionate dwarfs - Mother and son; coupled with transitory hypocalcemic tetany. Am J Roentgenol Radium Ther Nucl Med 1967;100:1-11.
5. Lee WK, Vargas A, Barnes J, Root AW. The Kenny-Caffey syndrome: Growth retardation and hypocalcemia in a young boy. Am J Med Genet 1983;14:773-82.
6. Sanjad SA, Sakati NA, Abu-Osba YK, Kaddoura R, Milner RD. A new syndrome of congenital hypoparathyroidism, severe growth failure, and dysmorphic features. Arch Dis Child 1991;66:193-6.
7. Sanjad S, Sakati N, Abu-Osba Y. Congenital hypoparathyroidism with dysmorphic features: A new syndrome. Pediatr Res 1988;23:71A.
8. Parvari R, Hershkovitz E, Grossman N, Gorodischer R, Loeys B, Zecic A, *et al*. Mutation of TBCE causes hypoparathyroidism-retardation-dysmorphism and autosomal recessive Kenny-Caffey syndrome. Nat Genet 2002;32:448-52.
9. Kelly TE, Blanton S, Saif R, Sanjad SA, Sakati NA. Confirmation of the assignment of the Sanjad-Sakati (congenital hypoparathyroidism) syndrome (OMIM 241410) locus to chromosome lq42-43. J Med Genet 2000;37:63-4.

Cite this article as: El Jabbour T, Aboursheid T, Keifo MB, Maksoud I, Alasmar D. Kenny-Caffey syndrome type 1. Avicenna J Med 2014;4:74-6.

Source of Support: Nil, **Conflict of Interest:** None declared.