Case Report

A case of Sandaj Sakati in a 6-year-old Child with Severe Resistant Pulmonary Infections

Mahmoud Ahmed Hassanin[*] [1], Omnia Adel [2], Abdelrahman Sallam [1], Omnia Magdy [4]

Intern, Alexandria University [1], Pediatrics resident, Alexandria University[2], Professor of pediatrics, Alexandria University [4]

Background

Sanjad Sakati was first identified in 1991 in Saudi Arabia and occurs almost exclusively in people of Arabian descent. Our case highlights the challenges in the treatment of the sanjad-sakati case from Egypt and what was done to achieve normal levels of calcium. Although it is proven that the syndrome is caused by a mutation in TBCE gene, little is known about the pathophysiology of the disease. Moreover, the effect of this mutation seems to have multiple effects, such as short stature, GH deficiency, hypo-cortisolemia [2], together with typical manifestations of micrognathia, prominent forehead, broad nasal bridge, and microcephaly [1]. The main aim of this research is to highlight the association between Sanjad Sakati syndrome and severe pulmonary infections as a serious cause of morbidity and mortality in these patients.

Case presentation

The case is a 6-year-old boy from Egypt who was born through normal vaginal delivery for consanguineous marriage. He has 4 sisters and 2 brothers who died from severe infections, who also experienced hypocalcemia and convulsions. Peculiarly, all his 4 sisters are in perfect health.

The mother had a normal prenatal course without any complications and had regular follow-ups. After birth, the parents didn't notice any concerns; however, at 40 days of age, he had generalized tonic-clonic seizures and the measured serum calcium was low, as well as a high phosphate level, suggesting hypoparathyroidism. After that, measured serum PTH was also found to be low, indicating the cause of hypocalcemia. He received calcium supplementation, and seizures were aborted.

Management and outcomes

He was discharged since then on regular calcium supplementation as well as active form of vitamin D but he had recurrent infections and fever requiring admission to the hospital for instance at 1.5 years of age he presented with fever, bronchopneumonia and hypocalcemia (5.5 mg/dl total calcium) despite adherence to calcium supplementation (calcium carbonate 1gm/8h) that required giving Ceftriaxone and IV calcium. Phosphorus level (6.9U/ml) was in the higher normal range. Calcium/ creatinine ratio was elevated (0.68 mg ca /mg cr). Immunophenotyping and flow cytometry reveal that all B cells, T cells and NK all lie within normal range, except for a mildly low CD4/CD8 ratio, which doesn't explain why the child has

recurrent severe infections. The thyroid profile was also normal.

At 2.5 years of age, he had cardiac arrest due to hypocalcemia, but he was successfully resuscitated.

The child has characteristic dysmorphic features: deep-set eyes, beaked nose, micrognathia, and frontal and occipital bossing.

He also has severe growth retardation and macrocephaly as well as global developmental delay, especially affecting speech and language development (saying only one-syllable words with vocabulary limited to just 2 words), although his speech understanding is preserved and follows different simple commands, and he feeds himself. He doesn't have any cardiac abnormalities and the main finding at the current presentation was fever and bronchopneumonia, indicated by reduced air entry on both sides of the chest, tachypnea and crackles supported by X-ray. His complete blood count also reveals microcytic hypochromic anemia (9.6 g/dl) as well as an elevated white blood cell count (17.9 thousand per mm3) with absolute neutrophilia and relative lymphopenia. His pneumonia was so severe at the current presentation that he required sustained oxygen supply through face mask due to decreased SO2 (80 %) in room air.

Table (1): Serum calcium, phosphorous and ALP at 4.5 years of age with adherence to treatment

Test	Level	Normal range
Total calcium	7.3 mg/dl	8.1 -10.4 mg/dl
phosphorus	9.3 mg/dl	2.5-5.0 mg/dl
ALP	186 U/L	40-129 U/L

Figure 1

Figure 2

Figures (1 &2): The child is 6 years old, presenting with deep-set eyes, frontal bossing, and micrognathia as indicated in the photos.

Figure (3): Oxygen supplementation through a face mask to reach normal oxygen saturation.

Discussions

This case shows two interesting key points, as he is the third case in the family after the passing of his two brothers due to the same condition. However, his 4 sisters are healthy, which seems to be a rare incident. The second is his prolonged and protracted hospital admission due to severe persistent pneumonia requiring multiple antibiotics. This association has not been explained before, but an assumption may be made that, despite a normal white blood cell count the genetic mutation affecting tubulin protein affects microtubule functions, which may impair neutrophil migration and phagocytosis of microorganisms. Pulmonary infections remain one of the major causes of morbidity and mortality for this group. [3]

Conclusion

Sanjad Sakati syndrome is a genetic syndrome that not only affects intellectual abilities or growth but also appears to have multiple effects and patients are prone to severe pulmonary infections that necessitate close follow-up and further research.

References

1. Sanjad SA, Sakati N, Abu-Osba YK, Kaddoura R, Milner R. A new syndrome of congenital hypoparathyroidism, severe growth failure, and dysmorphic features. 1991 Feb 1;66(2):193–6.
2. Padidela R, Kelberman D, Press M, Al-Khawari M, Hindmarsh PC, Dattani MT. Mutation in theTBCEGene Is Associated with Hypoparathyroidism-Retardation-Dysmorphism Syndrome Featuring Pituitary Hormone Deficiencies and Hypo-plasia of the Anterior Pituitary and the Corpus Callosum. The Journal of Clinical Endocrinology & Metabolism. 2009 Aug 1;94(8):2686–91.
3. Aminzadeh M, Galehdari H, Shariati G, Malekpour N, Ghandil P. Clinical features and tubulin folding cofactor E gene analysis in Iranian patients with Sanjad–Sakati syndrome. Jornal de Pediatria. 2020 Jan;96(1):60–5.