Kobe J. Med. Sci., Vol. 56, No. 6, pp. E239-E241, 2010

Brachydactylia Associated with Mitochondrial Disorder in an Octogenarian

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Received 30 November 2010/ Accepted 26 December 2010

Key Words: respiratory chain, metabolic disease, oxidative phosphorylation, skeletal deformity, dysmorphism

ABSTRACT

In a 80yo female with acute pancreatitis, myopathy, polyneuropathy, short stature, diabetes mellitus, hypothyroidism, hypoacusis, atrial fibrillation, hepatopathy, and renal cysts, mitochondrial disease was diagnosed. The family history for the disease was negative. Interestingly, the patient additionally presented with brachydactylia, which was also found in her son and father and has not been reported in association with mitochondrial disease before. Whether the relation between brachydactylia and mitochondrial disease was causal or coincidental remains speculative.

INTRODUCTION

Mitochondrial disorders (MIDs) have been reported in association with skeletal abnormalities such as facial dysmorphism [1]. The association of a MID with brachydactylia has not been described.

CLINICAL CASE

The patient is a 80yo Caucasian, HIV-negative female, height 160 cm, weight 82 kg with a history of diabetes mellitus type 2, polyneuropathy, restless leg syndrome, bilateral hypoacusis, thyroidectomy for struma nodosa with consecutive hypothyroidism, hepatopathy in the absence of alcoholism or hepatitis, arterial hypertension, atrial fibrillation, implantation of a pacemaker for bradycardia, resection of a basalioma on the nose, and multiple left-sided renal cysts. She was regularly taking anti-hypertensive drugs and laevothyroxin.

At age 80y she had been admitted for acute swelling of the right parotid gland. Diagnostic work-up revealed leucocytosis, anaemia, thrombocytopenia, hypokaliaemia, hypocalcaemia, hypomagnesaemia, markedly elevated alpha-amylase (992 U/l, n: 28-100 U/l), elevated transaminases, elevated gamma-glutamyl transpeptidase, hyperuricaemia, and markedly elevated C-reactive protein. A CT-scan of the face revealed an enlarged parotid gland on the right side with indications for inflammation. Acute parotitis was diagnosed and an antibiotic therapy with amoxicillin plus clavulanic acid was initiated. Despite this therapy a phlegmona developed requiring incision and drainage of the parotid gland with success. Several cultures from the material grew staphylococcus aureus in concentrations of maximally 10^3.

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Clinical neurologic investigation revealed bilateral hypoacusis, bilateral ptosis, weak head anteflexion, generally absent deep tendon reflexes, and surprisingly brachydactylia of all fingers and toes (figure 1). She reported brachydactylia also in her son and her father in the absence of any MID features in her relatives. For the restless leg syndrome L-DOPA was prescribed with success.

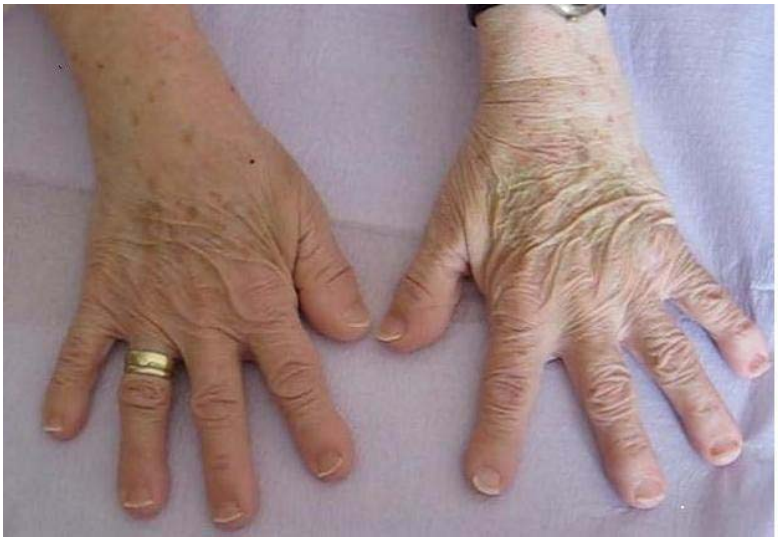

Figure 1. Hands of the described patient. View of the dorsum mani showing brachydactylia

DISCUSSION

Based upon the clinical presentation including myopathy with ptosis, polyneuropathy, short stature, diabetes mellitus, hypothyroidism, hypoacusis, atrial fibrillation, hepatopathy, and renal cysts, a MID was suspected. Whether anaemia, thrombocytopenia, hypokalaemia, hypocalcaemia, and hypomagnesaemia were also manifestations of the MID or infect-associated, remains speculative. However, such abnormalities have been previously reported in association with MIDs [2]. Also speculative remains if the parotitis was a manifestation of the MID or a coincidental finding. A causal relation cannot be excluded since at least pancreatitis has been repeatedly described as a phenotypic feature of a MID [3].

The surprising occurrence of brachydactylia together with MID in the presented patient may be a coincidental finding or causally related. Either the patient had a genetic disorder other than MID, of which brachydactylia is a key clinical feature (Table I), or brachydactylia was a manifestation of the MID. In the presented patient there was no indication for a disorder previously described in association with brachydactylia. Arguments for a manifestation of a MID are the multiorgan nature of the MID in the presented patient, suggesting brachydactylia as one of the phenotypic manifestations and that MIDs may occasionally go along with dysmorphic features and skeletal abnormalities [4]. Arguments against this assumption are that such an association has not been described previously and that her father and brother presented with brachydactylia in the absence of MID features.

Table I. Disorders associated with brachydactylia

Poland syndrome
Smith Magenis syndrome
Pseudohypoparathyroidism (Albright hereditary osteodystrophy)
Drinkwater's type brachydactylia
Alagille syndrome
Cranio-facio-mandibular dysostosis
Brachydactylia with synphalangia
Hereditary type E brachydactylia
Congenital dyslocation of head of radius with brachydactylia
Tetrameric poly-syndactylia combined with brachydactylia
Dominant hereditary brachydactylia with synostosis of the joints
Marble bone disease
Congenital brachydactylia
Familial brachydactylia

This case suggests that brachydactylia may be a rare phenotypic feature of a MID, although the causal nature of such a relation needs to be elucidated.

REFERENCES

1. **Berio, A., and Piazzi, A.** 2007. Facial anomalies in a patient with cytochrome-oxidase deficiency and subsequent Kearns-Sayre syndrome with growth hormone deficiency. Minerva Med **98:** 81-85.
2. **Lee, Y.S., Yap, H.K., Barshop, B.A., Lee, Y.S., Rajalingam, S., Loke, K.Y.** 2001. Mitochondrial tubulopathy: the many faces of mitochondrial disorders. Pediatr Nephrol **16:** 710-712.
3. **Debray, F.G., Drouin, E., Herzog, D., Lortie, A., Lambert, M., Garel, L., Mitchell, G.A., Michaud, J.L.** 2006. Recurrent pancreatitis in mitochondrial cytopathy. Am J Med Genet A **140:** 2330-2335.
4. **Yüksel, A., Seven, M., Cetincelik, U., Yeşil, G., Köksal, V.** 2006. Facial dysmorphism in Leigh syndrome with SURF-1 mutation and COX deficiency. Pediatr Neurol **34:** 486-489.