

Brachyonychia in a patient with Bardet-Biedl syndrome: Case report and review of this rare syndrome



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Key words: Bardet-Biedl syndrome; brachydactyly; brachyonychia.

INTRODUCTION

Bardet-Biedl syndrome (BBS) is a variably expressive autosomal recessive multisystem disorder caused by more than 20 distinct genes involved in cilia functioning, with a prevalence of approximately 1:100,000 in North America and Europe.¹ Primary features include polydactyly (with supernumerary digits), genital abnormalities, rod-cone dystrophy resulting in retinitis pigmentosa and blindness, obesity, renal defects, and learning difficulties. Secondary features include brachydactyly (shortening of the digits) or syndactyly (fusion of 2 or more digits), dental defects, ataxia or poor coordination, developmental delay, speech deficit, olfactory deficit, diabetes mellitus, and congenital heart disease.¹ A clinical diagnosis of BBS can be assigned when a patient has either 4 primary features or 3 primary and 2 secondary features.¹ Features of BBS progressively emerge during or after the first decade of life, and multigene sequencing can help confirm the diagnosis.

Manifestations of BBS involving the skin and nails are rarely reported in the literature. We present a novel case of brachyonychia in a patient with BBS. Brachyonychia (or racquet nails) is a clinical term defining nail units in which the width of the nail plates and nail beds is greater than the length.

CASE REPORT

A 44-year-old woman with a history of BBS complicated by postaxial polydactyly of the right foot, obesity, chronic kidney disease, hypertension, and blindness caused by retinitis pigmentosa presented to the dermatology clinic for keratosis pilaris

Abbreviation used:

BBS: Bardet-Biedl syndrome

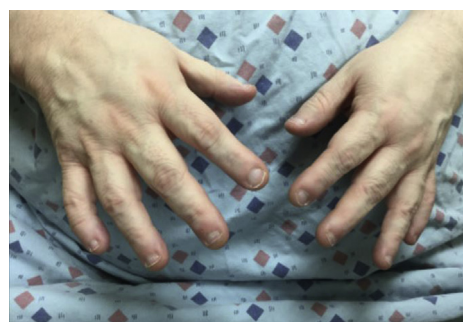


Fig 1. Clinical brachyonychia of the fingernails.

and retention hyperkeratosis. She had no history of psoriasis or eczema. On examination, clinical brachyonychia of the fingernails was noted (Fig 1).

Radiographs of the hands confirmed brachyphalangy (shortening of the phalanges) of the distal phalanges, involving the right second digit and the bilateral third and fourth digits (Figs 2 and 3). The degree of distal brachyphalangy for digits 2 through 5 was determined by calculating the relative ratios of the length of the distal phalanges to the length of the thumb distal phalanges, given the thumb has the longest distal phalanx.² The expected distal phalanx relative length was compared with the calculated relative length of our patient's distal phalanges, using a standard deviation of 1 mm (Table 1).² There was

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Fig 2. Radiograph of right hand shows brachyphalangy of the second, third, and fourth distal phalanges and the fifth middle phalanx, with clinodactyly of the fifth phalanx.



Fig 3. Radiograph of left hand shows brachyphalangy of the third and fourth distal phalanges and the fifth middle phalanx, with clinodactyly of the fifth phalanx.

also bilateral clinodactyly (abnormal curvature of the digit in the medial-lateral plane) and brachyphalangy of the fifth middle phalanges (Figs 2 and 3).

The patient had no history of nail biting, and no other nail abnormalities were present. Parathyroid hormone, phosphate, and calcium levels, which

Table I. The expected distal phalanx relative length was compared with the calculated relative length of our patient's distal phalanges

	Expected ratio of the distal phalanx length relative to the length of the distal thumb phalanges (%)	Ratio of the distal phalanx length relative to the length of the distal thumb phalanges in our patient, with a standard deviation of 1 mm (%)
Right 2nd phalanx	82	72 (66-77)
Left 2nd phalanx	82	77 (72-83)
Right 3rd phalanx	86	73 (67-78)
Left 3rd phalanx	86	71 (65-77)
Right 4th phalanx	87	79 (73-85)
Left 4th phalanx	87	78 (74-86)
Right 5th phalanx	78	75 (69-80)
Left 5th phalanx	78	76 (71-82)

Expected ratios of the distal phalanges relative to the thumb distal phalanges² were higher than the ratios calculated in our patient for the right second digit and the bilateral third and fourth digits. A standard deviation of 1 mm for distal phalanx length was used, as proposed by Aydinlioglu et al.²

were checked routinely because of her underlying renal disease, were within normal limits.

DISCUSSION

Involvement of the hair, skin, and mucous membranes is rarely reported with BBS and may be underrecognized. It is important for dermatologists to be aware of several dermatoses associated with BBS. Haws et al³ recently reported a series of 31 patients with BBS in which the 2 most common cutaneous findings were keratosis pilaris associated with xerosis (80.6%) and seborrheic dermatitis (19.3%). Less common findings included hidradenitis suppurativa, psoriasis, eczema, ichthyosis vulgaris, and confluent and reticulated papillomatosis.³ Skin findings associated with obesity including acanthosis nigricans, striae, acrochordons, and intertrigo were also noted in these patients.³ Gingival overgrowth⁴ and linear porokeratosis⁵ have been described in BBS as well.

BBS is a ciliopathy that is associated with polydactyly, syndactyly, and brachydactyly. Cilia are important in embryonic and postnatal bone development, and they serve as chemosensors and mechanosensors for bone formation and maintenance.⁶ Brachyonychia is caused by underlying distal brachyphalangy that can be seen on a radiograph,⁷ as demonstrated in our patient's radiographs.

Brachyonychia can be inherited in an autosomal dominant fashion and is typically limited to the thumb nail but may involve multiple nails.

Acquired racquet nails have been associated with hyperparathyroidism with underlying resorption of the distal terminal phalanges⁸ and psoriatic arthropathy and nail biting.⁹ Acquired generalized racquet nails with resorption of the terminal phalanx has been reported in Erasmus syndrome, a syndrome characterized by the co-occurrence of systemic sclerosis with silicosis.¹⁰

We contribute to the literature a novel case of brachyonychia in BBS. Given that brachyonychia is the clinical presentation of a shortened distal phalanx and that BBS is characterized by brachydactyly, we believe that brachyonychia is likely an under-recognized clinical feature in BBS. The addition of this case to the literature expands the clinical description of features present in BBS and may facilitate diagnosis of this syndrome characterized by serious systemic associations.

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