

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



Shannon Q. Detty, MD,<sup>a</sup> Molly A. Hinshaw, MD,<sup>a</sup> Michael J. Tuite, MD,<sup>b</sup> and Alexander D. Means, MD<sup>a</sup>  
Madison, Wisconsin

**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.<sup>1</sup> 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.<sup>1</sup> A clinical diagnosis of BBS can be assigned when a patient has either 4 primary features or 3 primary and 2 secondary features.<sup>1</sup> Features of BBS progressively emerge during or after the first decade of life, and multigene sequencing can help confirm the diagnosis.

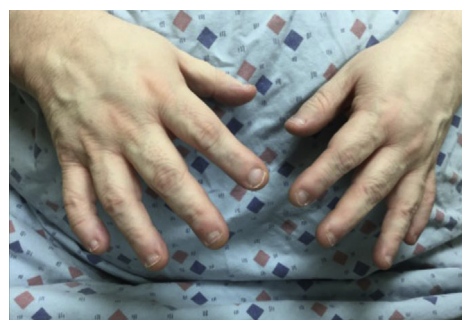
Manifestations of BBS involving the skin and nails are rarely reported in the literature. We present a novel case of brachyonychia in a patient with BBS. Brachyonychia (or racquet nails) is a clinical term defining nail units in which the width of the nail plates and nail beds is greater than the length.

## 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.<sup>2</sup> 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).<sup>2</sup> There was

From the Departments of Dermatology<sup>a</sup> and Radiology,<sup>b</sup> University of Wisconsin-Madison.

Funding sources: None.

Conflicts of interest: None disclosed.

Correspondence to: Shannon Q. Detty, MD, University of Wisconsin-Madison, Department of Dermatology, 1 S. Park Street, 7<sup>th</sup> Floor, Madison, WI 53715. E-mail: [sdetty@uwhealth.org](mailto:sdetty@uwhealth.org).

JAAD Case Reports 2019;5:672-4.  
2352-5126

Published by Elsevier on behalf of the American Academy of Dermatology, Inc. This is an open access article under the CC BY-NC-ND license (<http://creativecommons.org/licenses/by-nc-nd/4.0/>).

<https://doi.org/10.1016/j.jidcr.2019.05.015>



**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 phalanges<sup>2</sup> 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.<sup>2</sup>

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 al<sup>3</sup> 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.<sup>3</sup> Skin findings associated with obesity including acanthosis nigricans, striae, acrochordons, and intertrigo were also noted in these patients.<sup>3</sup> Gingival overgrowth<sup>4</sup> and linear porokeratosis<sup>5</sup> 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.<sup>6</sup> Brachyonychia is caused by underlying distal brachyphalangy that can be seen on a radiograph,<sup>7</sup> 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 phalanges<sup>8</sup> and psoriatic arthropathy and nail biting.<sup>9</sup> 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.<sup>10</sup>

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.

#### REFERENCES

1. Suspitsin EN, Imyanitov EN. Bardet-Biedl syndrome. *Mol Syndromol*. 2016;7(2):62-71.
2. Aydinlioglu A, Akpınar F, Tosun N. Mathematical relations between the lengths of the metacarpal bones and phalanges: surgical significance. *Tohoku J Exp Med*. 1998; 185(3):209-216.
3. Haws RM, McIntee TJ, Green CB. Cutaneous findings in Bardet-Biedl syndrome. *Int J Dermatol*. 2019. <https://doi.org/10.1111/jid.14412> [Epub ahead of print].
4. Drugowick RM, Da RósGonçalves L, Barrôso AS, et al. Treatment of gingival overgrowth in a child with Bardet-Biedl syndrome. *J Periodontol*. 2007;78:1159-1163.
5. Tappel AC, Tiwari N, Zlotoff B. Linear porokeratosis associated with Bardet-Biedl syndrome: a case report. *Pediatr Dermatol*. 2019. <https://doi.org/10.1111/pde.13776> [Epub ahead of print].
6. Yuan X, Yang S. Cilia/IFT protein and motor-related bone diseases and mouse models. *Front Biosci (Landmark Ed)*. 2015; 20:515-555.
7. Tosti A, Piraccini BM. Nail disorders. In: Bologna JL, Schaffer JV, Cerroni L, eds. *Dermatology*. New York, NY: Elsevier; 2018: 1203-1219.
8. Baran R, Turkmani MG, Mubki T. Acquired racquet nails: a useful sign of hyperparathyroidism. *J Eur Acad Dermatol Venereol*. 2014;28(2):257-259.
9. Tosti A, Baran R, Dawber RPR, et al. Nail configuration abnormalities. In: Baran R, Dawber RPR, Haneke E, et al., eds. *A Text Atlas of Nail Disorders*, 3rd ed. London: Martin Dunitz, Taylor & Francis Group plc; 2003:10-57.
10. Vetrichevvel TP, Renita L, Shobana S, Anandan S. Acquired racquet nails in Erasmus syndrome. *Int J Dermatol*. 2010;49(8): 932-933.