








Utility of optical coherence tomography in basal cell naevus syndrome: A case report

Genevieve Ho MD, MMed^{1,2,3}  | Rodrigo J. Schwartz MD^{1,4}  | Bruna Melhoranse-Gouveia MD, MPhil^{1,2}  | Pascale Guitera MD, PhD, FACD^{1,2,5}  | Niamh A. O'Sullivan MBBCh, PhD, MRCS (Ed), FACD¹  | Veronica K. Y. Cheung MChD⁶ | Sydney Ch'ng MBBS, PhD, FRACS^{1,2,7}  | Linda K. Martin BSc(Med), MBBS(Hons), MMed, FACD^{1,2,3} 

¹Melanoma Institute Australia, Sydney, New South Wales, Australia

²Faculty of Medicine and Health, University of Sydney, Sydney, New South Wales, Australia

³Faculty of Medicine and Health, University of New South Wales, Sydney, New South Wales, Australia

⁴Department of Dermatology, Faculty of Medicine, University of Chile, Santiago, Chile

⁵Sydney Melanoma Diagnostic Centre, Royal Prince Alfred Hospital, Sydney, New South Wales, Australia

⁶Department of Tissue Pathology and Diagnostic Oncology, Royal Prince Alfred Hospital, Camperdown, New South Wales, Australia

⁷Royal Prince Alfred Hospital, Sydney, New South Wales, Australia

Correspondence

Genevieve Ho, Melanoma Institute Australia, 40 Rocklands Road, North Sydney NSW 2065, Australia.
Email: genevieve.ho@melanoma.org.au

Abstract

Optical Coherence Tomography (OCT) is a useful non-invasive diagnostic tool for diagnosing and monitoring treatment of basal cell carcinomas. We describe the use of OCT in a patient with Basal Cell Naevus Syndrome. Through measuring tumour depth on OCT, management of individual tumours was triaged accordingly using 0.4 mm tumour depth as a cut-off for surgical and non-surgical management. OCT has potential to reduce unnecessary excisions and associated morbidity in this population of patients.

KEYWORDS

basal cell naevus syndrome, case report, Gorlin syndrome, optical coherence tomography, skin cancer

INTRODUCTION

Basal cell naevus syndrome (BCNS, OMIM #109400), also known as Gorlin syndrome or Gorlin-Goltz syndrome, is a genodermatosis caused by mutations affecting PTCH1, PTCH2 or SUFU of the sonic hedgehog signalling pathway. It is characterized by numerous early onset basal cell

carcinomas (BCCs) and other characteristic cutaneous and skeletal features including odontogenic keratocysts and palmoplantar pits. These younger patients pose a management challenge in balancing adequate treatment of BCCs and treatment-related morbidity from a cosmetic and psychological standpoint. Even the simplest histopathological assessment of BCCs with shave biopsy may

This is an open access article under the terms of the [Creative Commons Attribution-NonCommercial-NoDerivs](https://creativecommons.org/licenses/by-nc-nd/4.0/) License, which permits use and distribution in any medium, provided the original work is properly cited, the use is non-commercial and no modifications or adaptations are made.

© 2022 The Authors. *Australasian Journal of Dermatology* published by John Wiley & Sons Australia, Ltd on behalf of Australasian College of Dermatologists.

eventually feel too invasive considering the number of lesions in these patients. As such, effective and non-invasive diagnostic techniques are welcome.

Optical coherence tomography (OCT) utilizes low-coherence infrared light and interferometry. As different structures of the skin have varying optical properties, their varying refractions are captured and combined to produce an image *in vivo*. It can penetrate skin to depths of up to 2.5 mm depending on the machine and systems used, and can visualize a 6 × 6 mm field. The software allows the user to measure the thickness of structures within the image. In diagnosing BCC, OCT has a sensitivity and specificity ranging from 79%–94% and 85%–96% respectively.^{1,2} We report a case of use of OCT for non-invasive diagnosis and management planning in a patient with BCNS.

CASE HISTORY

Patient X is a female in her 30s who first developed BCCs at age 25 and who already had 15 BCCs curetted within the preceding 5-year period. She is of Fitzpatrick phototype II, has no history of excessive sun exposure and a family history of a first degree relative with multiple BCCs at a young age. At her first appointment with us, she had

24 lesions suspicious for BCC identified on dermoscopy (Figure 1a). She was noted to have bilateral syndactyly of the second and third toes of each foot. She had a background of papulonodular acne. No other specific features of BCNS were noted. She was confirmed to have a pathogenic variant in the PTCH1 gene (c.3307-1G>A), thus meeting major criteria for BCNS diagnosis.³

OCT (VivoSight Dx, Michelson Diagnostics Ltd.) was performed on all lesions to guide further management. Diagnostic features of BCC^{4,5} were identified in 19/24 lesions, with the remaining five papules presumed to be inflammatory acneiform lesions. The total time taken to scan all 24 lesions was under 30 min. Depth on OCT of BCCs was recorded, and along with anatomical location was used to guide treatment. Tumour depth was measured from the top of the epidermis to the lower margin of the deepest tumour nest. Of the 19 lesions with diagnostic BCC features on OCT, 5/19 lesions were <0.4 mm in depth and were treated as superficial BCCs with topical imiquimod (5 days/week for 6 weeks), excision was performed for 4/19 BCCs >0.4 mm in depth in high risk or cosmetically sensitive areas by a plastic surgeon, and curettage and cautery (C&C) was used to address the remaining lesions due to patient preference (Figure 1b). OCT was used at each subsequent visit to

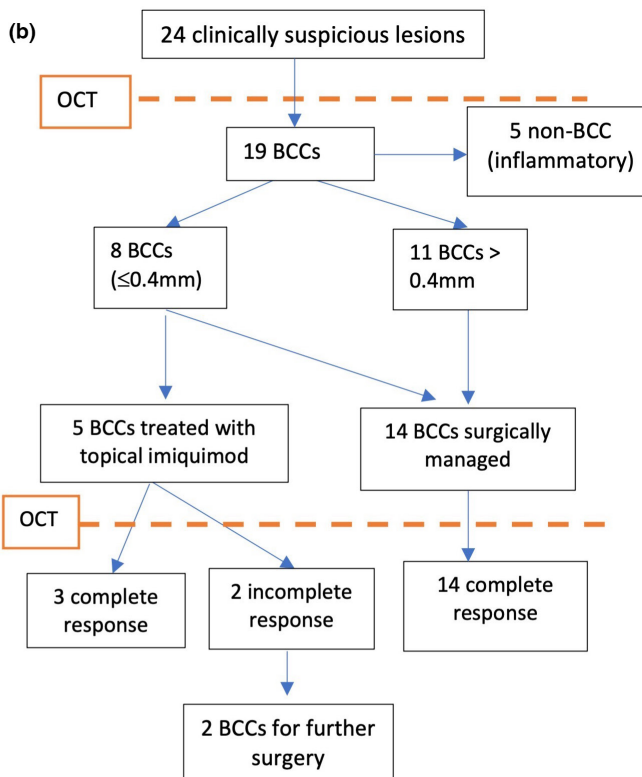


FIGURE 1 (a) Photograph of the patient's back at her initial visit showing the location of 12 lesions dermoscopically suspicious for basal cell carcinoma (BCC) identified for OCT assessment. (b) Subsequent management and outcomes for initially identified lesions suspicious for BCC

assess response to treatment, and 2/5 BCC's initially treated with imiquimod had incomplete response and were subsequently treated with C&C. OCT was helpful in distinguishing between keloidal scarring at BCC excision sites from recurrent BCC (Figures 2 and 3). At subsequent reviews over the following 9 months, five new BCC's were identified, and again triaged for excisional surgery, topical imiquimod or C&C according to depth of invasion and anatomical location.

DISCUSSION

OCT was a useful tool in triaging and guiding management in this case of BCNS, particularly for dermoscopically equivocal or difficult lesions. When added to dermoscopy, it may increase sensitivity from 90.6% to 95.7%, and specificity from 54.3% to 75.3% ($p < 0.001$).⁴ In our case, OCT was also useful in distinguishing BCC from acneiform lesions, and recurrence from scar.

Histopathological correlation with OCT findings has been previously described.^{4,5} The main OCT diagnostic

features seen in this case were epidermal protrusions into the upper dermis with a dark rim, dark oval structures at varying depths, some with peripheral rimming and disruption of the epidermis.

We relied on tumour depth to guide management. The use of ≤ 0.4 mm as a cut-off for non-surgical management is based on previous studies where tumour thickness > 0.4 mm was predictive of topical imiquimod treatment failure⁶ and because OCT/histopathological correlation is best when tumour depth is ≤ 0.4 mm.⁷ Depth correlation of OCT becomes less accurate with deeper tumours.^{1,7} Morphological features on OCT have recently been proposed to better discriminate superficial from non-superficial BCC subtypes, as is done histopathologically.⁸ Ideally, all BCCs > 0.4 mm should be excised for depth correlation and to confirm subtype and margins. In vivo reflectance confocal microscopy may also assist in confirming diagnosis of BCCs with cellular resolution of tumour morphology, although has a depth limitation of 0.2 mm and is less helpful in guiding treatment option.

In the setting of innumerable BCCs, use of OCT to triage and select tumours for non-surgical management

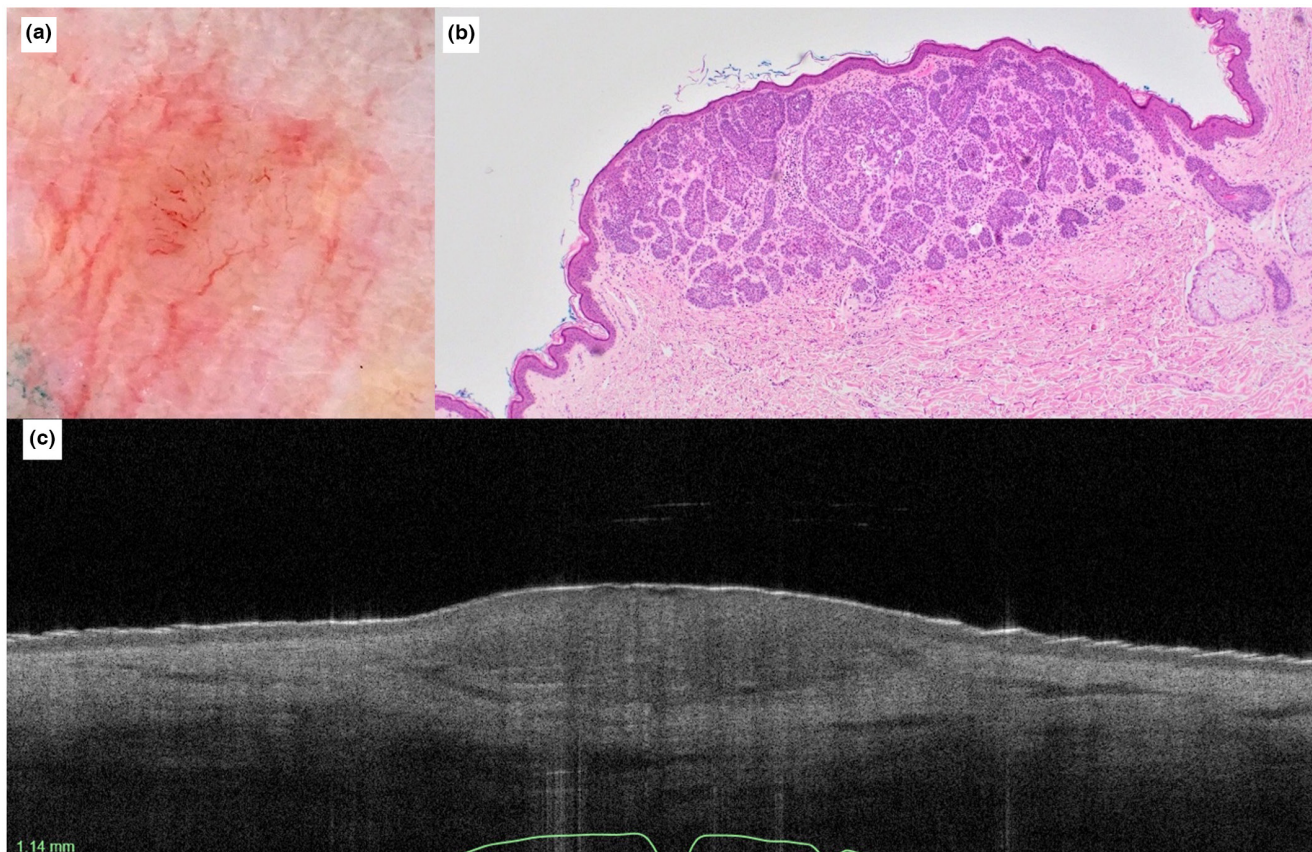


FIGURE 2 (a) Dermoscopy (taken with DermLite DL 4). (b) Histopathology and (c) OCT of BCC on the left neck. Dermoscopy shows an amelanotic lesion of 2 mm diameter with linear vessels (b) histopathology shows multiple tumour islands of nodular BCC to depth of 0.7 mm (haematoxylin and eosin stain; X10 magnification). (c) OCT shows hypoechoic (dark grey) ovoid structure, protruding into the upper dermis with clefting corresponding to tumour islands

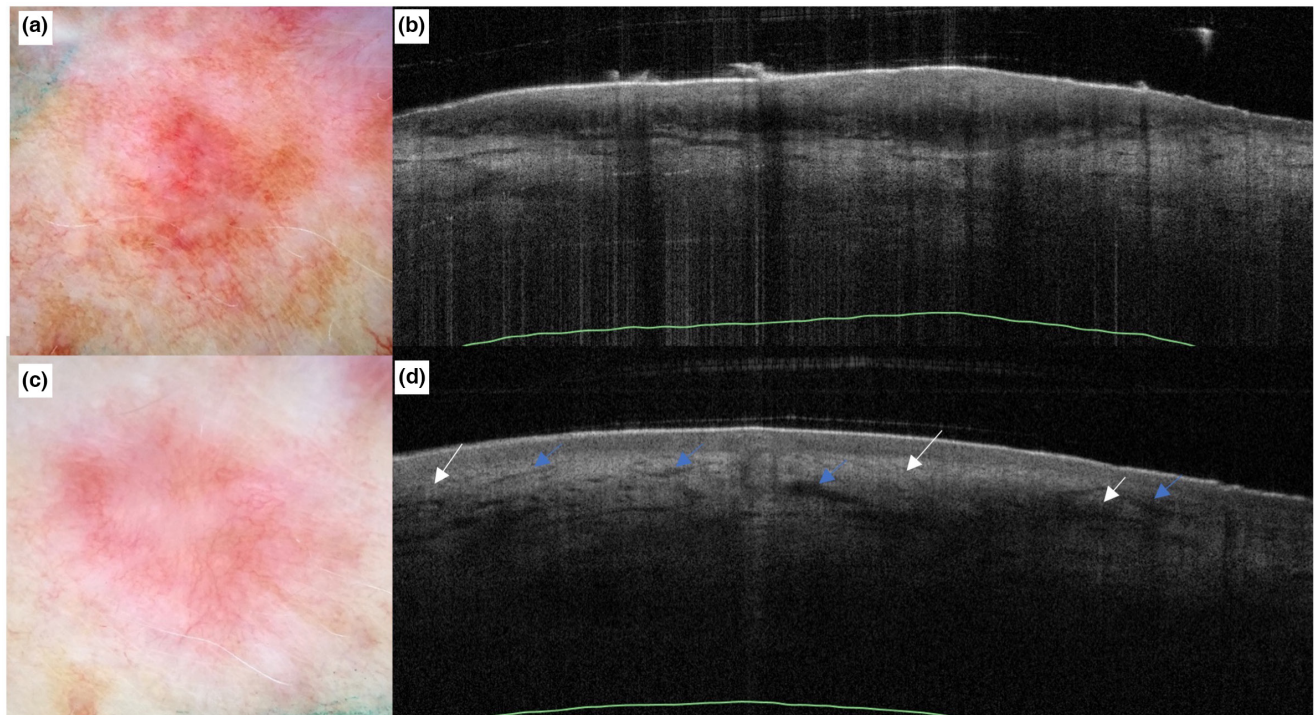


FIGURE 3 (a) Dermoscopy image taken at baseline of a lesion on the patient's right upper back. (b) OCT image of the BCC at baseline, showing hypoechoic tumour and clefting, consistent with a superficial BCC. (c) 9 months post treatment of a superficial basal cell carcinoma with atypical vessels and a keloid scar at previously curretted site on the patient's right upper back. (d) OCT image: There are no tumour islands visible; instead, there are hyper-reflective collagen bundles (white arrows) and numerous vessels (blue arrows) consistent with scar

may reduce the cumulative surgical burden in BCNS patients and optimize effective use of non-invasive therapies. OCT is helpful for monitoring treatment response and detecting recurrence, in a time-saving manner (15 s per structural scan). Given that 85% of BCNS patients require some form of annual surgical excision, reported to be the most disliked treatment modality as well as one associated with the highest psychosocial burden,⁹ non-invasive technologies have an important role in this patient cohort. Larger studies of standardized methodology would be helpful to further define the use of OCT in this population.

AUTHOR CONTRIBUTIONS

GH, RS, BMG and LM contributed to conception and design; GH, RS, PG, NAS, VC, SC and LM contributed to acquisition and interpretation of data; all authors were involved in drafting and revising the manuscript. All authors have given final approval of the version to be published and agree to be accountable for all aspects of the work.

FUNDING INFORMATION

GH is supported by NHMRC CRE funding (#1135285). RJS is supported by the Goodridge Foundation MIA Dermatology Fellowship. LKM is supported by the

Warwick L Morison Professorship of Dermatology, UNSW. Sunpharma donated the VivoSightDx device used in this report as a third party sponsored investigator-initiated research grant.

ACKNOWLEDGEMENT

Open access publishing facilitated by The University of Sydney, as part of the Wiley - The University of Sydney agreement via the Council of Australian University Librarians.

CONFLICT OF INTEREST

The authors have no conflicts of interest to declare.



DATA AVAILABILITY STATEMENT

Data sharing is not applicable to this article as no new data were created or analysed in this study.

INFORMED CONSENT


The patient in this manuscript has given written informed consent to the publication of their case details.

ORCID

Genevieve Ho  <https://orcid.org/0000-0003-3769-8951>
Rodrigo J. Schwartz  <https://orcid.org/0000-0003-1609-143X>

Bruna Melhoranse-Gouveia  <https://orcid.org/0000-0002-6322-5675>

Pascale Guitera  <https://orcid.org/0000-0001-9519-110X>

Niamh A. O'Sullivan  <https://orcid.org/0000-0002-1882-8303>

Sydney Ch'ng  <https://orcid.org/0000-0002-4011-1253>

Linda K. Martin  <https://orcid.org/0000-0001-9519-7077>

REFERENCES

1. Cheng HM, Guitera P. Systematic review of optical coherence tomography usage in the diagnosis and management of basal cell carcinoma. *Br J Dermatol*. 2015;173(6):1371–80.
2. Ulrich M, von Braunmuehl T, Kurzen H, Dirschka T, Kellner C, Sattler E, et al. The sensitivity and specificity of optical coherence tomography for the assisted diagnosis of nonpigmented basal cell carcinoma: an observational study. *Br J Dermatol*. 2015;173(2):428–35.
3. Bree AF, Shah MR. Consensus statement from the first international colloquium on basal cell nevus syndrome (BCNS). *Am J Med Genet A*. 2011;155A(9):2091–7.
4. Hussain AA, Themstrup L, Jemec GBE. Optical coherence tomography in the diagnosis of basal cell carcinoma. *Arch Dermatol Res*. 2015;307(1):1–10.
5. Gambichler T, Plura I, Kampilafkos P, Valavanis K, Sand M, Bechara FG, et al. Histopathological correlates of basal cell carcinoma in the slice and en face imaging modes of high-definition optical coherence tomography. *Br J Dermatol*. 2014;170(6):1358–61.
6. McKay KM, Sambrano BL, Fox PS, Bassett RL, Chon S, Prieto VG. Thickness of superficial basal cell carcinoma (sBCC) predicts imiquimod efficacy: a proposal for a thickness-based definition of sBCC. *Br J Dermatol*. 2013;169(3):549–54. <https://doi.org/10.1111/bjd.12402>
7. Cheng HM, Lo S, Scolyer R, Meekings A, Carlos G, Guitera P. Accuracy of optical coherence tomography for the diagnosis of superficial basal cell carcinoma: a prospective, consecutive, cohort study of 168 cases. *Br J Dermatol [Internet]*. 2016;175(6):1290–300. <https://doi.org/10.1111/bjd.14714>
8. Adan F, Mosterd K, NWJ K-S, Nelemans PJ. Diagnostic value of optical coherence tomography image features for diagnosis of basal cell carcinoma. *Acta Derm Venereol*. 2021;101(11):adv00607. <https://doi.org/10.2340/actadv.v101.421>
9. Ali FR, Collier NJ, Evans DG, Costello M, Webster S, Lear JT. National survey of patients with Gorlin syndrome highlights poor awareness, multiple treatments and profound psychosocial impact of disease. *J Eur Acad Dermatol Venereol*. 2016;30(2):371–3.

How to cite this article: Ho G, Schwartz RJ, Melhoranse-Gouveia B, Guitera P, O'Sullivan NA, Cheung VKY, et al. Utility of optical coherence tomography in basal cell naevus syndrome: A case report. *Australas J Dermatol*. 2022;63:e331–e335. <https://doi.org/10.1111/ajd.13921>