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Enucleation of Cylindromas in Brooke-Spiegler Syndrome: A Novel Surgical Technique

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Brooke Spiegler Syndrome (BSS), is a rare, autosomal dominantly inherited genetic condition characterised by the development of cylindromas, trichoepitheliomas and spiradenomas. Multiple cylindromas in BSS cause discomfort, functional impairment and significant cosmetic disfigurement. Patient management requires a considered approach, with appreciation for available treatment options and patient preference. We describe enucleation of multiple cylindromas in patients with BSS as a procedure that reduces tumour burden, spares scalp skin, and hair and provides excellent cosmesis. Enucleation allows for rapid healing and has a high patient acceptability. Our BSS patients have preferred this approach to alternative reconstructive techniques including electrosurgical resurfacing, excision with second intention healing, local flap repair or full thickness skin grafting.

Germline mutations in the tumour suppressor gene *CYLD* are thought to be central to the development of cylindromas, as well as other cutaneous adnexal tumours, namely spiradenomas and trichoepitheliomas.¹ Cylindromas are non-encapsulated adnexal tumours with a low malignant potential that usually involve the head and neck.² They usually arise in the second decade as erythematous, firm and sometimes painful nodular tumours that increase in size and number with age. This may result in considerable, confluent areas of tumour involvement that can pose a significant surgical challenge when extensive excision is balanced against conservation of normal tissue and function. Pain, cosmetic disfigurement, and functional impairment (for example if tumours encroach upon the auditory canal or arise on the eyelids) are common reasons for presentation. Whilst the molecular dissection of *CYLD* as a negative regulator of oncogenic cell signalling pathways such as NFκB and TRK may lead to effective medical therapy in the future³, the onus for treatment currently lies with surgery to provide control of tumour burden.

Although perhaps not an intuitive method of removing non-encapsulated dermal tumours such as cylindromas, we report success using surgical enucleation; a minimally invasive technique which preserves hair follicles and surrounding scalp by not excising the overlying

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skin. Individualisation of treatment is important, taking into account patient expectation and previous experience as well as tumour extent and location. Enucleation under local anaesthesia is not appropriate for treatment of the entire scalp, but may be used for the treatment of several tumours per session.

The area(s) to be treated should be appropriately delineated and then infiltrated with local anaesthetic. Respecting lines of skin tension, a superficial linear incision is made over the involved skin in order to reveal the tumour. Although cylindromas are non-encapsulated, it is possible, through blunt dissection and lateral fingertip pressure, to shell out the tumour bulk without compromising the overlying epidermis. Remnants can then be carefully dissected out and the defect closed with simple sutures, whilst excess skin caused by tumour stretch, may need to be excised in order to produce a flat scar. It is important to note that we explain to our patients that enucleation is essentially a debulking technique, not a curative procedure; hence there is a likelihood of tumour recurrence over the treated areas, as well as the development of new tumours.

Various other surgical modalities have been described in the literature⁴. Options include conventional excision, curettage, laser (CO₂ and Neodymium YAG), electrosurgery, Mohs micrographic surgery and, for extensive scalp involvement, complete scalp excision with split skin grafting – an outcome described in up to 1 in 4 *CYLD* mutation carriers.⁵

Our patients who have had their tumours enucleated have found this type of conservative surgery preferable, reporting an excellent cosmetic outcome. We highlight this surgical pearl as a novel method to manage tumour burden in appropriately selected cases.

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Figure 1. Cylindromas on the left temple.

(1a) Pre-operative. (1b) A linear incision is made that clearly reveals the tumour bulk. (1c) Fingertip pressure is applied to express the tumour. (1d) Post-operative. The wound edges are approximated with nylon sutures.