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Intent vs. impact: Calling time on outdated phenotypic labels in dermatology

Active social justice movements are exposing the structural disempowerment of minority groups throughout Western societies, including in healthcare. In the realm of rare genetic skin diseases, the term ‘turban tumour’ persists on PUBMED searches as recently as 2021 and is a pejorative and culturally insensitive description. Whilst likely to have been coined with benign intent, the impact of this term to the estimated 0.4% of the global population who are Sikh for example, is not the same. ‘Turban tumour’ is a term used to describe the presentation of confluent scalp tumours seen in *CYLD* cutaneous syndrome (CCS)¹, and has been used widely since at least 1933². CCS, or the eponymous Brooke-Spiegler Syndrome, is a rare genodermatosis predisposing to predominantly benign skin tumours without a recognised ethnic preponderance, caused by pathogenic variants in the *CYLD* gene³. We felt it would be bizarre to offer this term as a diagnosis to an 85-year-old Australian woman from Melbourne who recently presented with confluent cylindromas over her scalp, face and trunk (**Figure 1**), leading us to consider the barriers to change that exist and steps that could be taken to improve the lexicon.

In the pre-genetic era, Stillians in 1954² suggested cylindroma as a preferred term to describe these confluent tumours of the scalp in accordance with their histological architecture. Yet the term ‘turban tumour’ remains in use, for reasons that may include its infrequent clinical requirement, its alliterative nature, and its presence in key genetic references such as the Online Mendelian Inheritance in Man catalogue (at the time of writing). We would advocate for the term ‘turban tumour’ to be expunged from the medical literature, given its implicit cultural bias as well as its potential to be offensive to members of the Sikh community, as well as people of other faiths who use similar head coverings. Pejorative language impacts negatively on access to health for minority groups, who may be less likely to engage with health services as a result⁴. Despite this, we recognise change may be slow. Culturally offensive and inaccurate colloquialisms in dermatology, such as the term “Mongolian blue spot”⁵ used to describe congenital dermal melanocytosis, continue in everyday use despite being raised as problematic⁵ almost 30 years ago. It is encouraging to

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see that in the UK, the Royal College of Paediatrics and Child Health is using Twitter in 2021 to shift away from “Mongolian blue spot”.

We propose that in the context of genetic disease, the revision of online resources is key. We have now appropriately updated the National Centre for Biotechnology Information resource GeneReviews, the National Institute of Health Genetic and Rare Disease Information Centre, the US National Library of Medicine Medline Plus, the British Association of Dermatologists patient information leaflets and Orphanet with the inclusive term *CYLD* cutaneous syndrome. This is in an effort to move towards a label that is based on the genetics that drive the diverse phenotypes seen in this condition, where there is no relationship to *CYLD* genotype. Intrinsically, the use of a gene-based diagnosis focuses the consultation on genetics and will enhance genomic literacy. We suggest that the use of outdated phenotypic labels is considered by editors and reviewers of peer reviewed medical literature, as they are also well placed to end the perpetuation of such terminology. The impact of our lexicon has to be considered in a global context, and this shift enhances cultural safety and the provision of equity of access to health care services.

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Figure Legend

Fig.1 CYLD cutaneous syndrome presenting with confluent scalp tumours, a favoured site for this hair follicle tumour syndrome.



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