Late presentation was a marker that patients either did not have cellulitis or also had lymphoedema or eczema as a comorbidity to LLC. Patients with LLC may be categorized into two broad clinical groups: firstly, patients with straightforward LLC that responds to antibiotics, and secondly, a complex group of patients with comorbid lymphoedema and eczema in whom the diagnosis and treatment of LLC are more challenging. This latter group may benefit from early specialist input. It has been shown that a shared-care approach between primary and secondary care improved early and accurate diagnosis and the avoidance of prolonged antibiotics.[7]

The participants represented a population from a specialist service who were likely to have more complex disease. Therefore the results may have reflected the referral pattern to secondary care. The proportion of all patients in primary care being treated for cellulitis and eventually found not to have cellulitis remains unclear, so these findings are applicable to secondary care. It would be useful to validate this study in a prospective primary care cohort, where patients may present more acutely. However, this analysis is clinically useful because this subset presents a common diagnostic and management challenge. This analysis assumed a correct diagnosis from the service. Experienced dermatologists may have used the duration of symptoms to help reach a diagnosis of LLC or other diagnoses. If validated diagnostic criteria are developed for LLC these could be utilized in future studies.

In conclusion, patients with a shorter history of symptoms were more likely to have LLC rather than other diagnoses. The duration of symptoms may help predict a diagnosis of LLC. Patients with complex LLC with comorbid lymphoedema or eczema could be referred to specialist services earlier to reduce prolonged courses of antibiotics with no clinical improvement.

K. Mistry and N.J. Levell

Department of Dermatology, Norfolk and Norwich University Hospital, Norwich, UK

Email: Khaylen.Mistry@nnuh.nhs.uk

References

1 Brindle R, Williams M, Barton E *et al.* Assessment of antiobitic treatment of cellulitis and erysipelas – a systematic review and meta-analysis. *JAMA Dermatol* 2019; **155**:1033–40.

2 National Institute for Health and Care Excellence. Cellulitis – acute. Available at: https://cks.nice.org.uk/cellulitis-acute (last accessed 20 May 2020).

3 Thomas KS, Brindle R, Chalmers JR *et al.* Identifying priority areas for research into the diagnosis, treatment and prevention of cellulitis (erysipelas): results of a James Lind Alliance Priority Setting Partnership. *Br J Dermatol* 2017; **177**:541–3.

4 Patel M, Lee SI, Akyea RK *et al.* A systematic review showing the lack of diagnostic criteria and tools developed for lower-limb cellulitis. *Br J Dermatol* 2019; **181**:1156–65.

5 Mistry K, Sutherland M, Levell NJ. Lower limb cellulitis: low diagnostic accuracy and underdiagnosis of risk factors. *Clin Exp Dermatol* 2019; **44**:e193–5.

6 Weng QY, Raff AB, Cohen JM *et al.* Costs and consequences associated with misdiagnosed lower extremity cellulitis. *JAMA Dermatol* 2017; **153**:141–6.

7 Levell NJ, Wingfield CJ, Garioch JJ. Severe lower limb cellulitis is best diagnosed by dermatologists and managed with shared care between primary and secondary care. *Br J Dermatol* 2011; **164**:1326–8.

Funding sources: This research received no specific grant from any funding agency in the public, commercial or not-for-profit sectors.

Conflicts of interest: The authors declare they have no conflicts of interest.

Detection of genetic tumour predisposition syndromes using electronic health records

DOI: 10.1111/bjd.19215

Dear Editor, The restricted tumour repertoire of certain genetic skin tumour syndromes, such as CYLD cutaneous syndrome (CCS), is well recognized.[1] Patients with CCS develop multiple benign cylindromas and spiradenomas, a presentation of skin appendageal tumours recognized to be exclusively associated with loss of *CYLD* function.[2] Consequently, pathology coding records, which capture tumour-level data, can be interrogated to detect patients who may have undiagnosed CCS based on the presentation of multiple CCS-specific tumours over time. We explored the feasibility of this approach in a tertiary hospital serving a population of 3 million people, where a national genetic testing centre for CCS was also based.

We interrogated the hospital pathology coding system, using SNOMED codes and free-text searches to identify patients with cutaneous cylindromas and spiradenomas presenting over a 3-year period during which *CYLD* gene testing data was available (2012–2015). Trichoepithelioma, seen in some patients with CCS, was excluded, as the presentation of multiple trichoepitheliomas may be associated with other tumour syndromes such as naevoid basal cell carcinoma syndrome. During this period, 62 803 skin cases were identified, of which 951 were appendageal tumours. Ninety-three cylindromas, spiradenomas or tumours with histological features of both were detected from 52 patients. We excluded patients who presented with one tumour only (44 tumour specimens) as these were more likely to be sporadic cases. We included data for patients who had more than one tumour, or a combination of any of these lesions. This identified 14 patients with 49 tumour specimens. Electronic case note review was then performed to determine whether these patients had undergone *CYLD* testing according to the UK Gene Testing Network criteria,[3] or had been diagnosed on clinical grounds. The majority (12 of 14 patients) had undergone dermatogenetic review and gene testing and were confirmed to have CCS; however, two did not appear to have a clinical diagnosis of CCS.

Our data reveal several interesting observations. Firstly, skin tumours that are strongly associated with specific gene disorders can be used to detect respective patients with genetic conditions from electronic health records. A related approach has been described, where diagnostic coding data are refined using multiple electronic records such as prescription data to improve identification of patients with diabetes.[4,5] Patients with CCS in this series who had not been clinically diagnosed stand to benefit from genetic testing, counselling and skin surveillance. Secondly, the presence of associated tumours in CCS such as salivary gland tumours[6] and malignant transformation[2] can be monitored.

The generalizability of this approach to other genetic disorders beyond CCS, where cancer and skin appendageal tumours may occur in combination, is important. Muir–Torre syndrome, where sebaceous carcinoma and bowel cancer develop, Birt–Hogg–Dubé syndrome, where trichodiscomas and renal cell carcinoma present, and Reed syndrome, where leiomyomas and renal cell carcinoma present, are pertinent examples. In these conditions, screening for malignancy could be initiated in relevant individuals, and in the case of Muir–Torre syndrome chemoprevention with aspirin can reduce cancer risk.[7]

Recently in the UK, national registration has been initiated for skin cancer.[8] As we transition to a digital health service, it is conceivable that all skin pathology cases will be searchable, including skin appendageal tumours. We propose that the application of filtering algorithms could be applied to increasingly comprehensive electronic health records, allowing patients with an underlying genetic condition to be detected. Limitations of this proof-of-principle study are that some patients with CCS will have only one biopsy at the time of study, and typographical errors in free-text coding can result in omission of patients. Our method found that most patients with potential CCS identified from electronic pathology records during this interval had a confirmed diagnosis of CCS. Future studies, as genetic testing for CCS becomes established as a standard of care, should also investigate the number of patients with a confirmed diagnosis of CCS who are not identified using our method in the electronic pathology records. Nonetheless, these data are relevant, at a time when artificial-intelligence-based analysis of large datasets such as national cancer registries is underway.

The ability to detect any potential genetic disease by proxy methods raises issues regarding consent, similarly to those raised in the context of conventional genetic testing. Public perception of genetic testing is changing, and patient group engagement will be important in guiding how such algorithms may be acted upon when they detect a patient with a genetic predisposition. For example, it may involve notification only when a helpful intervention is possible. It would be beneficial to conduct qualitative interviews in individuals with conditions such as CCS to capture their perceptions of the risks and benefits of being diagnosed in this manner. Patients have the right not to know about a genetic diagnosis, yet some may consider it a failing of a digital healthcare system if data point to a genetic diagnosis where screening or chemoprevention is possible, and yet no action is taken. We highlight that electronic health records are a rich resource, and continue to offer new facets that can impact on patient care.

S. Cook,[1] V. Wilson,[2] J. Ness,[1] J. Burn,[3] A. Husain[1] and N. Rajan[3,4]

[1]*Department of Pathology and* [4]*Department of Dermatology, Royal Victoria Infirmary, Newcastle upon Tyne, UK;* [2]*Molecular Diagnostics, Northern Genetics Service, Newcastle upon Tyne, UK and* [3]*Translational and Clinical Research Institute, Newcastle University, Newcastle upon Tyne, UK*

Correspondence: Neil Rajan.

Email: neil.rajan@newcastle.ac.uk

References

1 Rajan N, Ashworth A. Inherited cylindromas: lessons from a rare tumour. *Lancet Oncol* 2015; **16**:e460–9.
2 Davies HR, Hodgson K, Schwalbe E *et al.* Epigenetic modifiers DNMT3A and BCOR are recurrently mutated in CYLD cutaneous syndrome. *Nat Commun* 2019; **10**:4717.
3 Dubois A, Wilson V, Bourn D, Rajan N. *CYLD* genetic testing for Brooke–Spiegler syndrome, familial cylindromatosis and multiple familial trichoepitheliomas. *PLOS Curr* 2015; **7**:eogt.45c4e63d-d43d62e12228cc5264d6a0db.
4 Makam AN, Nguyen OK, Moore B *et al.* Identifying patients with diabetes and the earliest date of diagnosis in real time: an electronic health record case-finding algorithm. *BMC Med Inform Decis Mak* 2013; **13**:81.
5 Wilke RA, Berg RL, Peissig P *et al.* Use of an electronic medical record for the identification of research subjects with diabetes mellitus. *Clin Med Res* 2007; **5**:1–7.
6 Jungehulsing M, Wagner M, Damm M. Turban tumour with involvement of the parotid gland. *J Laryngol Otol* 1999; **113**:779–83.
7 Burn J, Gerdes AM, Macrae F *et al.* Long-term effect of aspirin on cancer risk in carriers of hereditary colorectal cancer: an analysis from the CAPP2 randomised controlled trial. *Lancet* 2011; **378**:2081–7.
8 Venables ZC, Nijsten T, Wong KF *et al.* Epidemiology of basal and cutaneous squamous cell carcinoma in the U.K. 2013–15: a cohort study. *Br J Dermatol* 2019; **181**:474–82.

Funding sources: the work of N.R. was supported by a Wellcome Trust-funded Intermediate Clinical Fellowship WT097163MA. The research of N.R. is also supported by the Newcastle NIHR Biomedical Research Centre.

Conflicts of interest: The authors declare they have no conflicts of interest.

The prevalence of hidradenitis suppurativa is shown by the Secure Anonymised Information Linkage (SAIL) Databank to be one per cent of the population of Wales

DOI: 10.1111/bjd.19210

Dear Editor, Hidradenitis suppurativa (HS) is a chronic inflammatory skin disease with multiple inflammatory skin lesions