Case report: Reducing the diagnostic odyssey in Behcet's disease using a digital health approach into primary care UK

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Introduction

- Behcet's disease is a rare, multisystemic disease that is poorly understood[1].

- Under-diagnosis is common due to the complexity of the disease and low physician awareness[2].

- We show that a digital health approach may lead to earlier diagnosis by scanning Electronic Health Records (EHR), at scale, to identify patients with rare undiagnosed diseases.
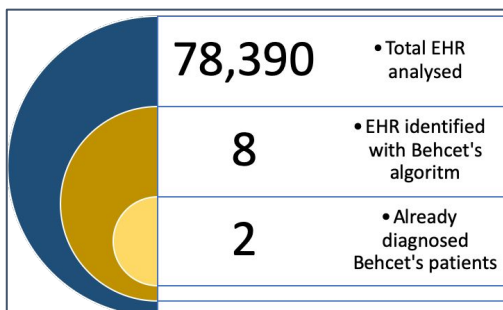
Methods

- This digital health approach used Clinical Terms Version 3 (CTV3) codes to build algorithms based on published rare disease diagnostic or case finding criteria.

- We used these algorithms to search primary care EHR and highlight patients who breached the encoded criteria.

- The algorithm for Behcet's disease was derived from the International Criteria for Behcet's Disease (ICBD)[3], a criteria in routine clinical use with a score of four-points or more indicating a diagnosis with a sensitivity of 94.8% (95% CI: 93.4-95.9%), and a of specificity (90.5%, 95% CI: 87.9-92.8%)[3].

| International Criteria for Behcet's Disease (ICBD) criteria ||
Sign / symptom	Points
Ocular lesions	2
Genital aphthosis	2
Oral aphthosis	2
Skin lesions	1
Neurological manifestations	1
Vascular manifestations	1
Positive pathergy test	1

- We applied the algorithm to a primary care EHR dataset of n=78,390. We then performed an analysis of patients identified by the algorithm which had previously received a diagnosis of Behcet's.


78,390	• Total EHR analysed
8	• EHR identified with Behcet's algoritm
2	• Already diagnosed Behcet's patients

- Previously diagnosed patients were reviewed to see if our approach would have identified these patients earlier.

Results

- Eight patients breached the clinical criteria, of which two patients had already been diagnosed with Behcet's.

- One patient breached the criteria with five points at 32 years old, four years earlier than the first recorded diagnostic code for Behcet's disease in their EHR at 36 years old. This patient's findings were:

Sign / symptom	EHR	Patient points
Ocular lesions	"Acute conjunctivitis" (XE16X)	2
Genital aphthosis		
Oral aphthosis	"Oral aphthae or mouth ulcer" (J082.)	2
Skin lesions	"Acne vulgaris" (XE1BO)	1
Neurological manifestations		
Vascular manifestations		
Positive pathergy test		
Total		*5 points*

- "Acne vulgaris" (XE1BO) and Acute conjunctivitis (XE16X) were encoded at the age of 31, "Oral aphthae or mouth ulcer" (J082.) at the age of 32.
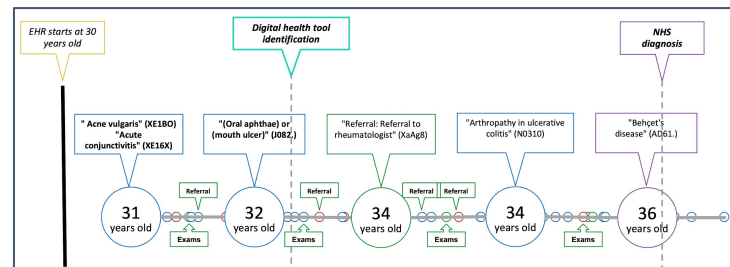
Discussion

- This approach has the potential to identify patients with Behcet's disease earlier than current clinical practice. Further analysis in larger datasets will enable an assessment of the sensitivity and specificity of this approach, and its clinical utility as an adjunctive tool in routine clinical practice.

References

- [1] Sakane T, Takeno M, Suzuki N, Inaba G. Behçet's disease. *New England Journal of Medicine* 1999; 341(17): 1284–1291. Doi: 10.1056/ NEJM199910213411707
- [2] Kiss E, Dohán J, Németh J, Poór G. Egy ritkán felismert ritka betegség: a Behcet-kór [Behcet's disease: a rarely recognized orphan disorder]. *Orv Hetil.* 2013; 154(3):93–101. Doi: 10.1556/ OH.2013.29528
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Case report: cost-benefit of earlier diagnosis in a patient with Behcet's disease using CTV3 codes at primary care UK

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Introduction

- It is challenging to build robust economic models for rare diseases since few epidemiologic and economic studies exist[1].
- We sought to establish a cost-benefit model for earlier diagnosis using real-world data collected from primary care Electronic Health Records (EHR) using Clinical Terms Version 3 (CTV3) codes[2].
- Our aim was to determine the healthcare utilisation and costs saved by making an earlier diagnosis of a patient with Behcet's disease.

Methods

- We analysed the Electronic Health Record (EHR) of a patient with Behcet's disease, who could have been diagnosed four years earlier by using digital encoding of an established clinical guideline[3].
- We extracted the Clinical Terms Version 3 (CTV3) codes from their primary care EHR. We then calculated the time to diagnosis. This was defined as the first time a coded entry of a clinical feature associated with Behcet's disease was encoded until the time that a diagnosis was recorded.

Relevant Behcet's CTV3 codes identified
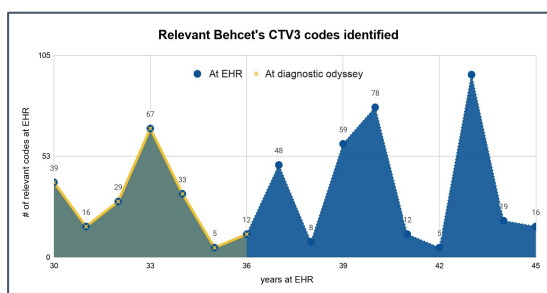
- We identified all the codes at the diagnostic odyssey that could be related to Behcet's disease (GP visits, signs & symptoms, tests, referrals, interventions and misdiagnosis.


Diagnostic odyssey CTV3 codes categorization
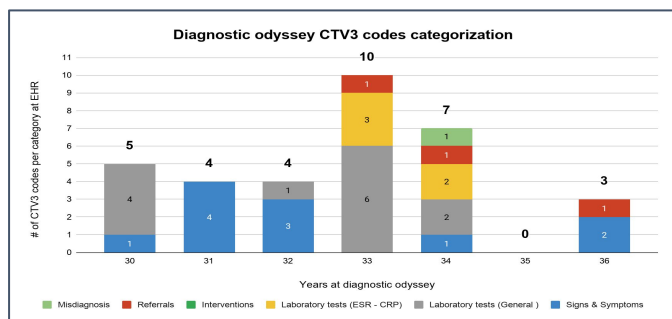
- We assigned for each identified code a cost attributed to the National Health Service (NHS)[4]. Then we calculated the potential total health service utilisation and the possible cost of the diagnostic odyssey.
- We compared costs incurred during the patient's real diagnostic odyssey (six years), to the shorter journey to diagnosis (two years) had the digitally encoded diagnostic criteria been used.

Results

- Total healthcare costs over the 6 year period were £1038. Of which £812 was incurred as costs during the 4 years after the earlier potential diagnostic date had the digital diagnostic criteria been used, a 78% reduction.

Diagnostic odyssey costs before and after digital health identification
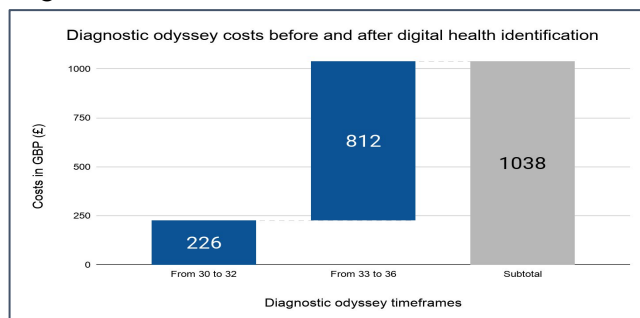
- Annual healthcare costs were highest in the 4 years prior to diagnosis.

Diagnostic odyssey costs per year in GPB (£)
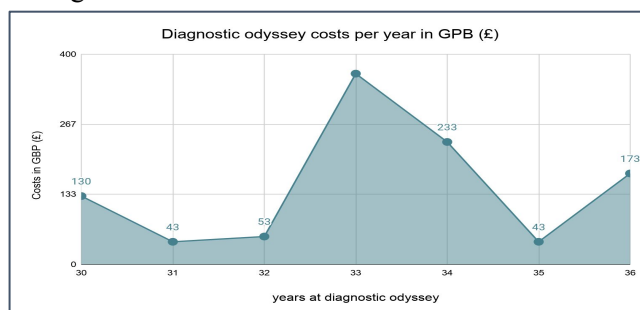
- This saving in health care utilization included the potential avoidance of 3 unnecessary referrals, investigations and 1 misdiagnosis.

Health utilisation at diagnostic odyssey
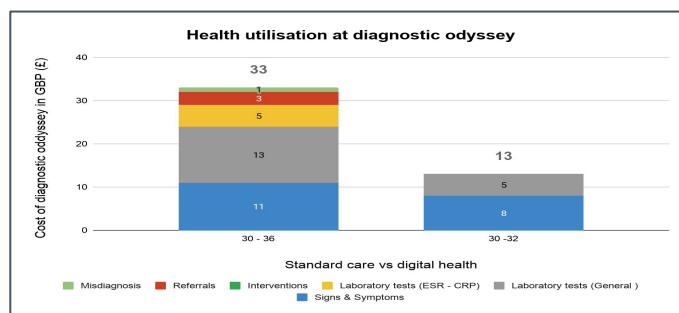
Discussion

- Data from primary care EHR allows the stratification of healthcare resources and calculation of costs.
- This study demonstrates that developing economic models at an individual patient level can help quantify the cost savings of earlier diagnosis in Rare Diseases.
- Further work is needed to replicate these findings in other cases and diseases.

References

- [1] Sakane T, Takeno M, Suzuki N, Inaba G. Behçet's disease. *New England Journal of Medicine* 1999; 341(17): 1284–1291. Doi: 10.1056/NEJM199910213411707
- [2] NHS Digital. 2020. *Read Codes - NHS Digital*. [online] Available at: <https://digital.nhs.uk/services/terminology-and-classifications/read-code

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Funded by Innovate UK