

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

Orlando Buendia<sup>1</sup>, Rudy Benfredj<sup>1</sup>, Timothy Halford<sup>1</sup>, Isabella Rustiglioni<sup>1</sup>, Peter Fish<sup>1</sup>, Will Evans<sup>1</sup>, Renate Apse<sup>1</sup>, Nicolas Aussel<sup>1</sup>, Connor Toal<sup>1</sup>, Valeria Garzon<sup>1</sup>.  
<sup>1</sup>Mendelian Ltd, London, United Kingdom

## Introduction

- Behcet's disease is a rare, multisystemic disease that is poorly understood<sup>1</sup>.
- Under-diagnosis is common due to the complexity of the disease and low physician awareness<sup>2</sup>.
- 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.

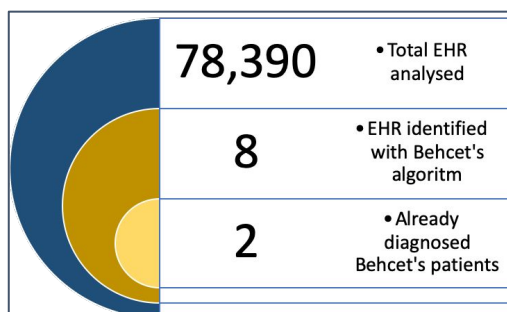
## Methods

- 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)<sup>3</sup>, 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%)<sup>3</sup>.

### 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.



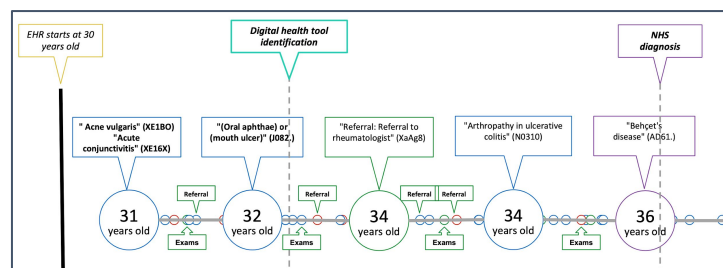
- Previously diagnosed patients were reviewed to see if our approach would have identified these patients earlier.

## 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		
<b>Total</b>		<b>5 points</b>

- "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

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