A Digital Health Approach: Early Identification Of Undiagnosed Rare Disease Patients At Scale

ASHG VIRTUAL MEETING 2020 OCTOBER 27-30

Connor Toal[1], Renate Apse[1], Orlando Buendia[1], Peter Fish[1], Will Evans[1]

[1]Mendelian Ltd, London, United Kingdom

Introduction

- Rare disease patients frequently experience a significant delay in diagnosis, on average 7.6 years in the US[1].
- This is due to their multifaceted complexity and low levels of awareness amongst clinicians leading to misdiagnoses, unnecessary investigations, referrals and treatments[1].
- This study shows that a novel digital approach, scanning Electronic Health Records (EHRs) at scale, may lead to earlier diagnosis.

Methods

- Mendelian has developed a series of algorithms to assist in identifying patients with rare diseases from their EHRs.
- Published clinical criteria for a range of rare diseases were digitised using SNOMED CT codes to create this algorithm.
- This algorithm was deployed across a population of 501,188 patients' NHS (UK) primary care EHRs. .
- Chronic Progressive External Ophthalmoplegia (CPEO)[2] and Behçet's[3] disease are used as examples for this poster.
- A number of patients who had previously breached criteria were identified, some of which had already been diagnosed with that rare disease as per their EHR.
- We reviewed the EHRs of theses previously diagnosed patients to assess if our algorithm could have identified them earlier, and if so, what effect this may have had on the diagnostic odyssey.

Results

- Across the two diseases, 237 patients had breached the criteria of which 9 had already been diagnosed (Figure 1.)

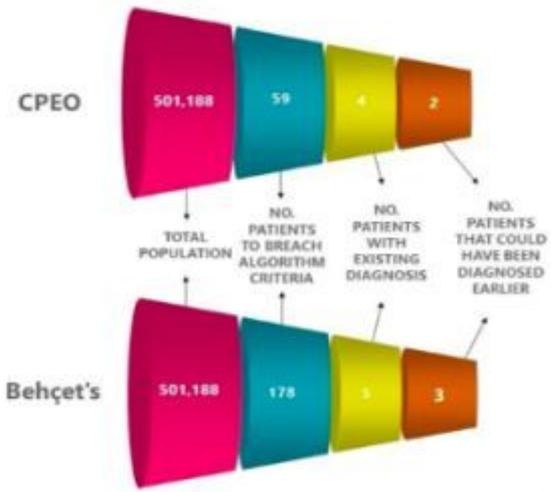

Figure 1 *Mendelscan identification*

- 5 of the diagnosed patients could have been identified earlier with our approach (Table 1.)
- An example diagnostic odyssey for a Behçet's patient is illustrated below, with details altered to protect patient privacy (Figure 2.)

Patient	Diagnosed on EHR (Years)	Algorithm breached (Years)	Difference (Years)
CPEO #1	68	60	**8**
CPEO #2	74	67	**7**
Behçet's #1	36	32	**4**
Behçet's #2	32	30	**2**
Behçet's #3	47	38	**9**

Table 1 *Years saved for 5 identified patients*

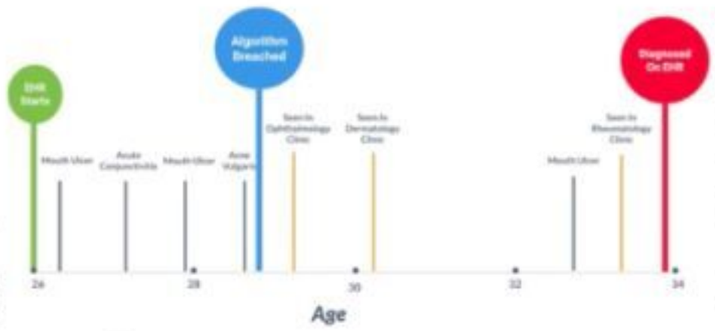

Figure 2 *Example diagnostic odyssey*

Conclusions

- This research demonstrates the potential of using digital analysis of EHRs to diagnose patients with rare diseases earlier.
- Patients who breach the algorithm criteria are first reviewed internally alongside disease experts, with suitable cases highlighted for further investigations and referrals.
- Further prospective studies in an expanded panel of rare diseases are ongoing to assess the clinical utility and economic impact of this digital approach.

References:

1) Vandeborne, L., van Overbeeke, E., Dooms, M. et al. Information needs of physicians regarding the diagnosis of rare diseases: a questionnaire-based study in Belgium. Orphanet J Rare Dis 14, 99 (2019). https://doi.org/10.1186/s13023-019-1075-8

2) POLG-Related DisordersCohen BH, Chinnery PF, Copeland WC. POLG-Related Disorders. 2010 Mar 16 [Updated 2018 Mar 1].

3) International Team for the Revision of the International Criteria for Behçet's Disease (ITR-ICBD). The International Criteria for Behçet's Disease (ICBD): a collaborative study of 27 countries on the sensitivity and specificity of the new criteria. J Eur Acad Dermatol Venereol. 2014;28(3):338–347. doi:10.1111/jdv.12107

info@mendelian.co

Funded by Innovate UK

ASHG American Society of Human Genetics