A digital health approach: Reducing the diagnostic odyssey of Hereditary Hemorrhagic Telangiectasia using UK Primary Care Electronic Health Records

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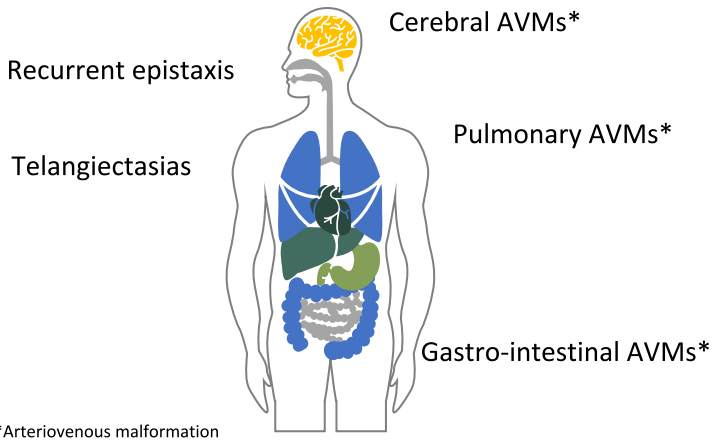
Objective

- **To demonstrate how a digital health tool that scans EHRs may lead to an earlier diagnosis of Hereditary Hemorrhagic Telangiectasia (HHT)**

Introduction

- HHT is a rare multisystemic disease that is poorly understood.[1] **Under-diagnosis is common** due to the disease's complexity and low physician awareness.[2]

Figure 1. Signs & Symptoms of HHT



Methods

- A **digital health tool**, takes published disease criteria and maps these to the appropriate Snomed CT code to create a digital criteria algorithm.
- The algorithm for HHT was derived from the Curaçao diagnostic criteria.[3]

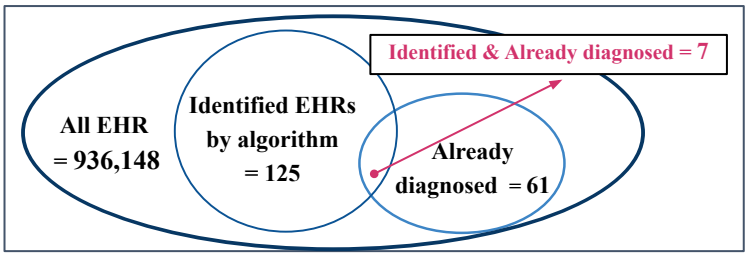
Curaçao diagnostic criteria for HHT

Sign / symptom	Points	Classification
Recurrent epistaxis	1	2 points = Possible diagnosis 3 points = Diagnosis
Telangiectasias	1	
Family history of HHT	1	
AVM malformations	1	

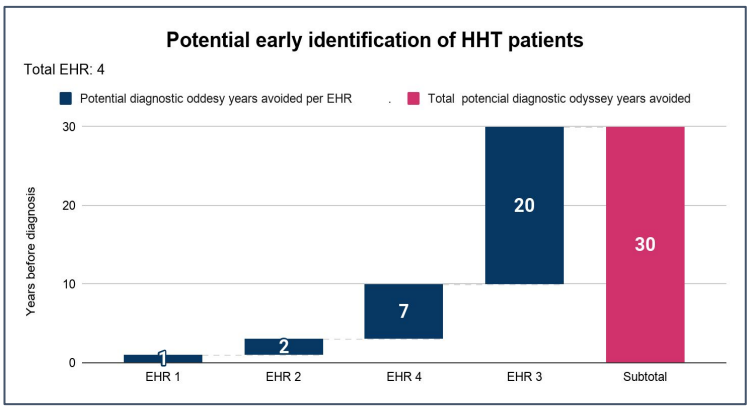
- This digitised criteria algorithm was applied to the primary care **EHRs of 936,148 patients** (434,960 Biobank and 501,188 from a single primary care practice federation) highlighting those that match the algorithm.
- We identified the patients' EHRs that both met the HHT algorithm criteria and had an existing diagnostic code for HHT (21877004). We then analysed these EHRs to explore their pre-diagnostic primary care record.

Results

- **61 EHRs** had a SNOMED CT code for HHT (21877004).



- **125 EHRs** matched the digitised criteria, of which **7** patients' had a previous diagnosis of HHT.
- Of these **7** already diagnosed patients, **4 would have matched the digitised criteria in advance** of the HHT diagnostic code appearing in their EHR.
- 3 EHRs matched the digitised criteria with 2 points (possible diagnosis) **1, 2 and 20 years before** the diagnostic code for HHT appeared on their record.
- 1 EHR matched the digitised criteria with 3 points (definite diagnosis) **7 years before** the diagnostic code for HHT appeared on their record.



Conclusions

with HHT **earlier than** current **clinical practice** with important implications for clinical management.

- Further **prospective studies** are planned to evaluate the sensitivity and specificity of this digital approach and its implementation as an **adjunctive tool** in routine clinical practice.

References

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- [2] Donaldson JW, McKeever TM. The UK prevalence of hereditary haemorrhagic telangiectasia and its association with sex, socioeconomic status and region of residence: a population-based study. Thorax. 2014 Feb;69(2):161-7. doi: 10.1136/thoraxjnl-2013-203720. Epub 2013 Nov 4. PMID: 24188926.
- [3] Shovlin CL, Guttmacher AE. Diagnostic criteria for hereditary hemorrhagic telangiectasia (Rendu-Osler-Weber syndrome). Am J Med Genet. 2000 Mar 6;91(1):66-7. doi: 10.1002/(sici)1096.

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Identifying potential cases of Juvenile Polyposis - Hereditary Haemorrhagic Telangiectasia syndrome using Primary Care Electronic Health Records in the UK

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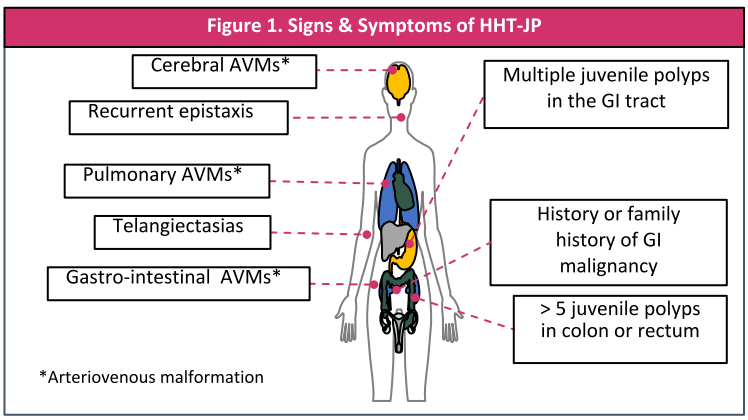
Objective

Identify patients who may have Juvenile Polyposis syndrome - Hereditary Haemorrhagic Telangiectasia (JP-HHT) from a cohort of HHT patients by using a digital health tool.

Introduction

- Juvenile Polyposis Syndrome (JPS) is an autosomal dominant condition characterised by >5 gastrointestinal (GI) hamartomatous polyps and a predisposition to GI cancer. Hereditary Haemorrhagic Telangiectasia (HHT) is an autosomal dominant vascular dysplasia affecting multiple organs.[1]
- A rare **combined syndrome, JP–HHT**, caused by a mutation in the **SMAD4 gene**, has recently been recognised with patients having features of both individual diseases.[1]
- JP-HHT has been previously reported in **22%** of patients with HHT.[2]

Figure 1. Signs & Symptoms of HHT-JP

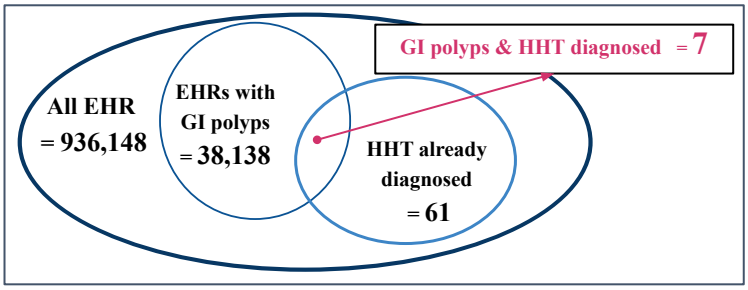


Methods

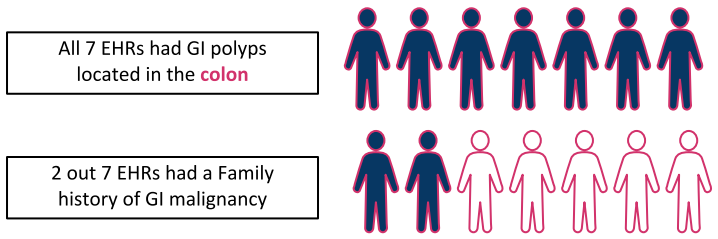
- A formal diagnostic criteria for JP–HHT has **not yet been established.**[2] We created a digital health tool which allowed us to highlight patients with features suggestive of both conditions for further evaluation.
- We analysed **936,148 pseudo-anonymised electronic health records** (EHRs) (434,960 Biobank and 501,188 from a single primary care Clinical Commissioning Group) to select those with the **diagnostic code for HHT** (SNOMED CT code: 21877004). These EHRs were then analysed to identify patients who also carried diagnostic SNOMED CT codes indicating **gastrointestinal polyposis**.
- Identified patients who met **both conditions** were analysed for metrics related to both conditions: age of polyp finding, location of polyp, GI malignancy, anaemia, AVM malformation, age of HHT diagnosis, telangiectasia, epistaxis, digital clubbing, family history of HHT and history of genetic referral.

Results

- **61** EHR were identified with a diagnosis of HHT by the presence of the associated SNOMED CT code, **7** of these EHRs also had a code indicating GI polyposis.

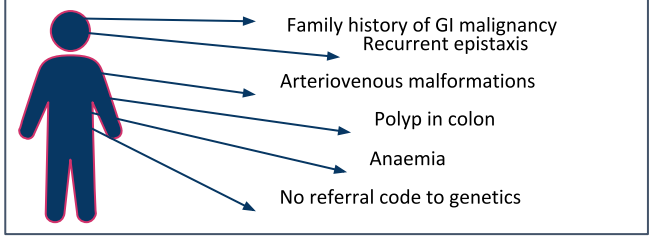


- All identified EHRs had GI polyps located in the **colon** along with a diagnosis of HHT. The age of diagnosis of GI polyp in this cohort ranged from **46 to 80 years (average age 59 years).**
- 3 out of 7 EHRs had a **genetic referral** indicating that **4 out of 7 have not** yet received input from clinical genetics.



- 1 out of 7 EHRs had AVM, consisted with previous JP–HHT reports exhibiting a high percentage of AVMs (87%).[1]

1 out of 7 EHRs had multiple HHT/JP overlapping findings



Discussion

- Identifying the subset of HHT patients who potentially have combined JP-HHT is important as **clinical management is different** for each aspect of the disease and may involve clinicians from multiple specialities.
- The use of **digital health tools to identify** patients with phenotypic features of JP-HHT could **assist clinicians** in earlier diagnosis of this **very** rare condition, facilitating improved clinical management and care.

References

[1] Overlapping spectra of SMAD4 mutations in juvenile polyposis (JP) and JP-HHT syndrome. *American journal of medical genetics. Part A*, *152A*(2), 333–339..
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