PheNominal: An EHR-Integrated Application for Annotation of Human Phenotype Ontology Terms at the Point of Care

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INTRODUCTION

Diagnosis of genetic disorders requires correlation between phenotypic and genotypic data. Human Phenotype Ontology (HPO) is a widely used standard for annotating phenotypic data. However, *post hoc* approaches to extract HPO terms from clinical notes using NLP have insufficient recall and precision for diagnostic interpretation, while point-of-care approaches are time-consuming in a busy clinical setting.

We sought to develop a tool that could be used at the point of care and that facilitated capture of discrete HPO terms for secondary use. The tool needed to serve the needs of many groups: clinical users, the genetic diagnostic laboratory, and researchers who study the genetic causes of disease.

METHODS

We developed a web application that runs within the EHR during a clinical encounter. The app communicates bidirectionally in real-time with the EHR using vendor-specific webservices.

Users can review, browse, add, and delete HPO terms, traverse parent/child terms as needed, and flag terms for association with the patient's medical record using a "shopping cart" metaphor.

Once saved, the terms are linked to the patient's record and formatted for one-click import into a clinic note as well as downstream reuse for diagnostic laboratories and researchers.

RESULTS

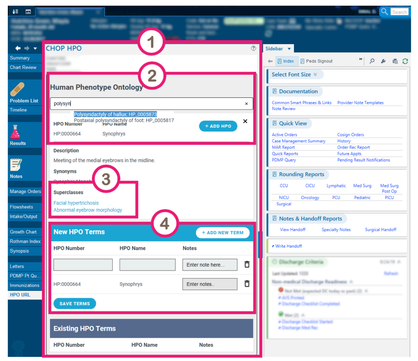
PheNominal has been used to capture discrete HPO terms for over 300 patients in both the RIMGC and Neurology clinics. On average, time to annotate HPO terms was reduced from ~15 minutes per patient via manual entry to ~5 minutes per patient, with fewer typographic errors and an average of 8 terms per patient.

PheNominal is a web-based tool that allows users to quickly tag a patient's record with **Human Phenotype Ontology terms** during a clinical encounter.

Terms are fetched and stored in the **EHR** using **web services** and can easily be pulled into **clinical** and **diagnostic** workflows, including **variant filtering pipelines**.

These high-fidelity discrete phenotypes can help to **accelerate research** for human genetic disorders.

Take a picture to learn more about the Roberts Individualized Medical Genetics Center at CHOP



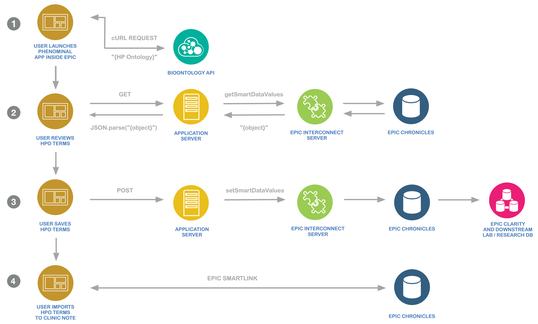
1. PheNominal runs within the EHR, alongside other clinical documentation tools.
2. Using a "shopping cart" metaphor, users can easily browse the entire HPO database, including 13K terms and 150K annotations. The search bar has an adaptive auto-complete feature as users type. Once the correct term is identified, users select "+ ADD HPO"
3. HPO is a true ontology, with parent and child terms. Users can easily traverse the ontology to find the best / most-specific term.
4. All terms are visible for final review and annotation before they are saved to the EHR. The list of terms are saved twice: once as an RTF string to permit inclusion into the note, and once as a JSON string to permit secondary reuse outside of the EHR.

Icon by dDara from the Noun Project

INTENDED USE

Genetic counselors and other expert users can use PheNominal to quickly annotate a patient's chart with HPO terms.

SYSTEM ARCHITECTURE



LIMITATIONS

- Character limit on SmartData Elements in Clarity
- Dependency on vendor-specific web services
- Dependency on vendor-specific mechanism to import terms into a clinical note (e.g. Epic "SmartLink")
- Intended for staff with deep domain expertise
- No widely-adopted standard for representation of phenotype data in HL7 FHIR

INTEROPERABILITY & FUTURE WORK

Future releases should explore the SMART on FHIR framework, which would require a number of changes:

1) Standard representation of HPO terms in the PhenoPackets format, an emerging standard
2) Storage/Retrieval of data as a FHIR "Observation" resource with associated EHR flowsheet values
3) Demonstration of integration in both Epic and non-Epic EHRs via the SMART platform
4) Identify equivalent function of a "SmartLink" to import terms into a clinical note
5) Permit domain-specific ontologies other than HPO

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