LOINC2HPO: Improving Translational Informatics by Standardizing EHR Phenotypic Data Using the Human Phenotype Ontology

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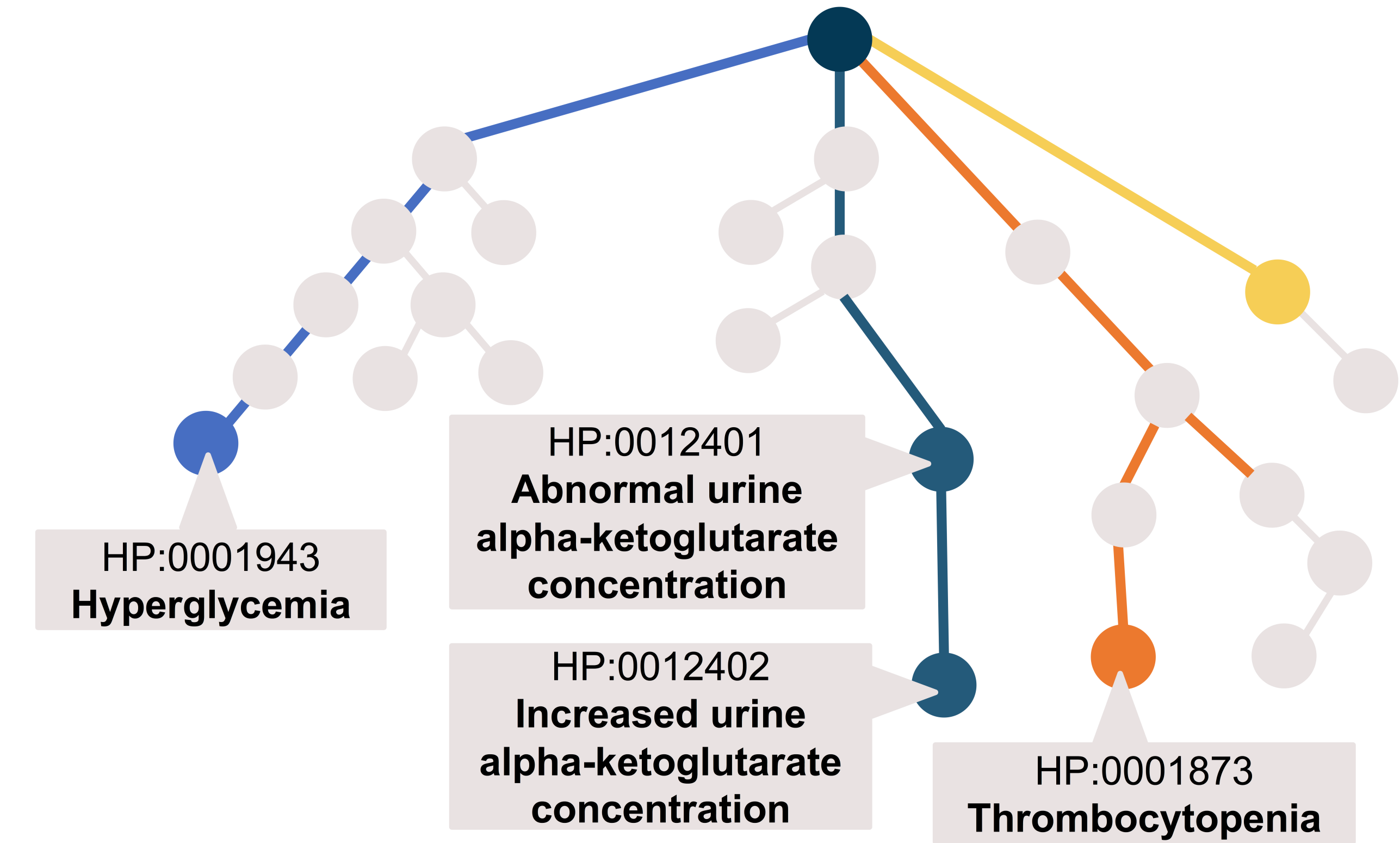
ABSTRACT

Clinical laboratory tests are identified with Logical Observation Identifier Names and Codes (LOINC) codes but many similar tests are represented with different LOINC codes.

We created a library that mapped laboratory test results, such as low, normal or high outcomes to medical implications in the Human Phenotype Ontology (HPO). Using this mapping, we can infer patients' phenotypic abnormalities using HPO terms, thus allowing semantic integration of laboratory tests and extracting patient phenotypes in large scale.

LOINC

- A universal standard for identifying medical laboratory observations in Electronic Health Records (EHRs)
- Lab tests, encoded as observations in Fast Healthcare Interoperability Resource (FHIR), are uniquely identified with LOINC codes
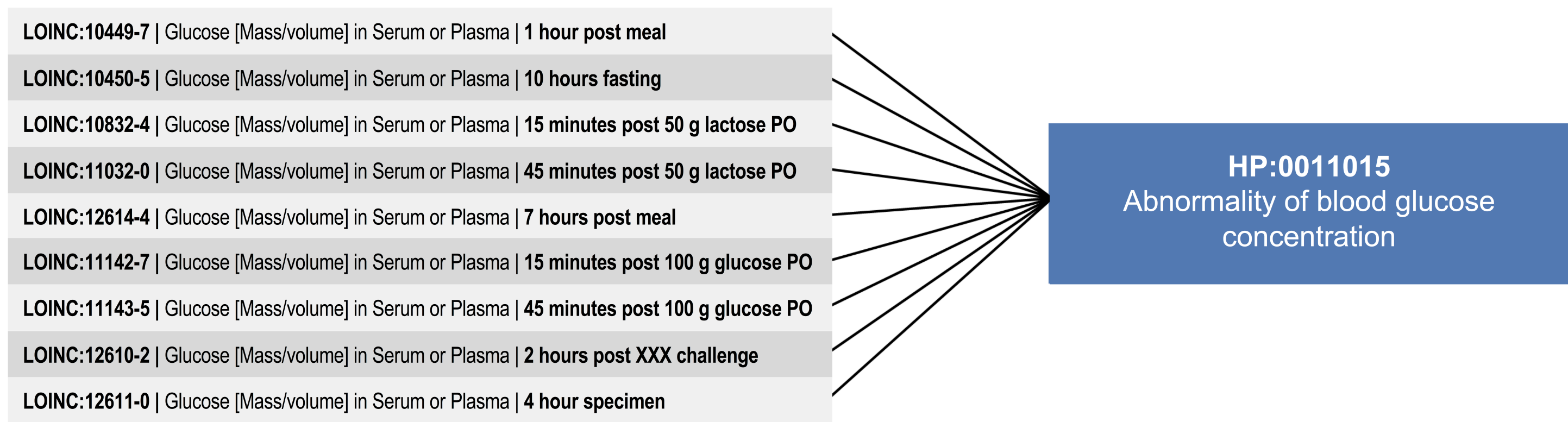
LOINC	Name	Component	Property	Time	Aspect	System	Scale	Method
10450-5	Glucose [Mass/volume] in Serum or Plasma --10 hours fasting	Glucose*post 10H CFst	MCnc	Pt		Ser/Plas	Qn	
777-3	Platelets [#/volume] in Blood by Automated count	Platelets	NCnc	Pt		Bld		Automated count
5769-5	Bacteria [#/area] in Urine sediment by Microscopy high power field	Bacteria	Naric	Pt		Urine sed	Qn	Microscopy.light.HPF

HUMAN PHENOTYPE ONTOLOGY

HP:0001943 Hyperglycemia
HP:0012401 Abnormal urine alpha-ketoglutarate concentration
HP:0012402 Increased urine alpha-ketoglutarate concentration
HP:0001873 Thrombocytopenia

The HPO contains over 13,000 classes that represent phenotypic abnormalities encountered in human diseases.

LOINC2HPO MAPPING STRATEGY

LOINC:10449-7 | Glucose [Mass/volume] in Serum or Plasma | 1 hour post meal
LOINC:10450-5 | Glucose [Mass/volume] in Serum or Plasma | 10 hours fasting
LOINC:10832-4 | Glucose [Mass/volume] in Serum or Plasma | 15 minutes post 50 g lactose PO
LOINC:11032-0 | Glucose [Mass/volume] in Serum or Plasma | 45 minutes post 50 g lactose PO
LOINC:12614-4 | Glucose [Mass/volume] in Serum or Plasma | 7 hours post meal
LOINC:11142-7 | Glucose [Mass/volume] in Serum or Plasma | 15 minutes post 100 g glucose PO
LOINC:11143-5 | Glucose [Mass/volume] in Serum or Plasma | 45 minutes post 100 g glucose PO
LOINC:12610-2 | Glucose [Mass/volume] in Serum or Plasma | 2 hours post XXX challenge
LOINC:12611-0 | Glucose [Mass/volume] in Serum or Plasma | 4 hour specimen

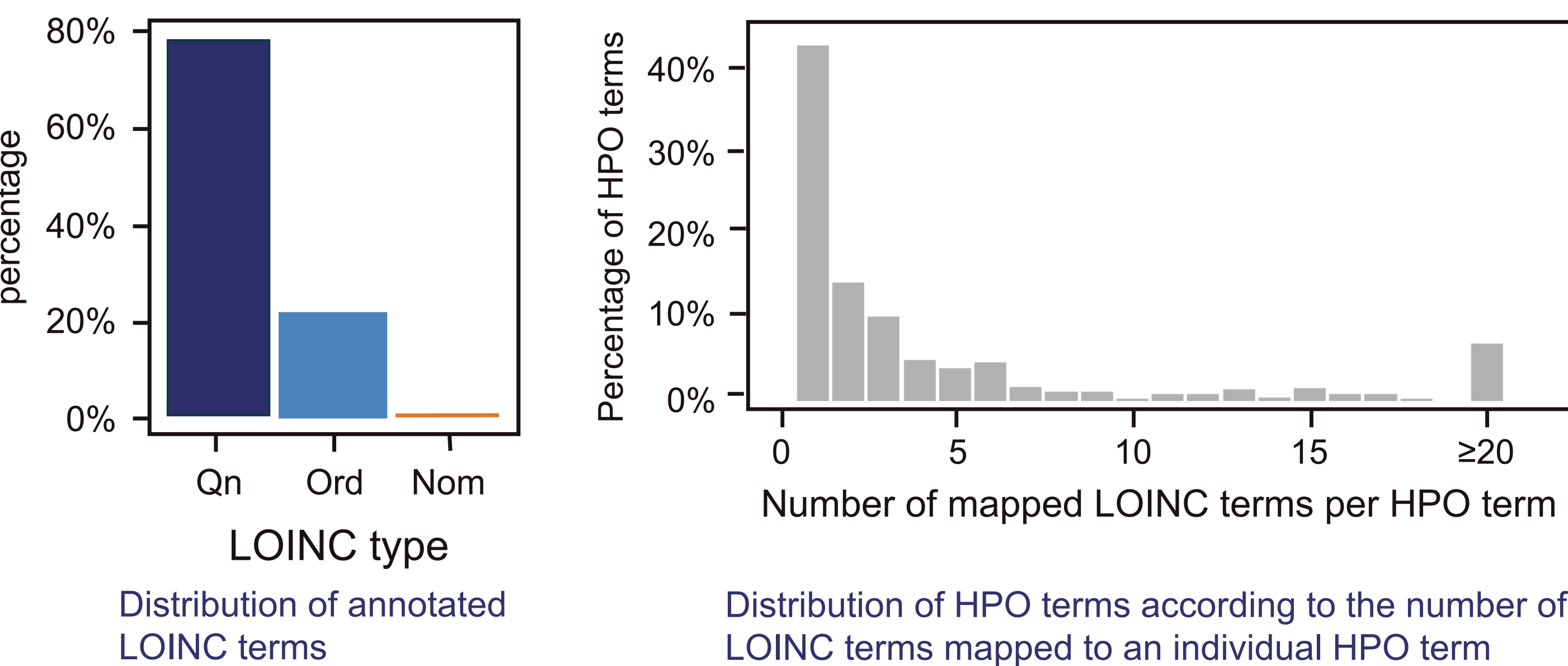HP:0011015 Abnormality of blood glucose concentration

- The entire LOINC table has ~85,000 entries
- The top 2,000 LOINC represent about 98% of all tests in real world applications
- Multiple LOINC codes map to single HPO terms

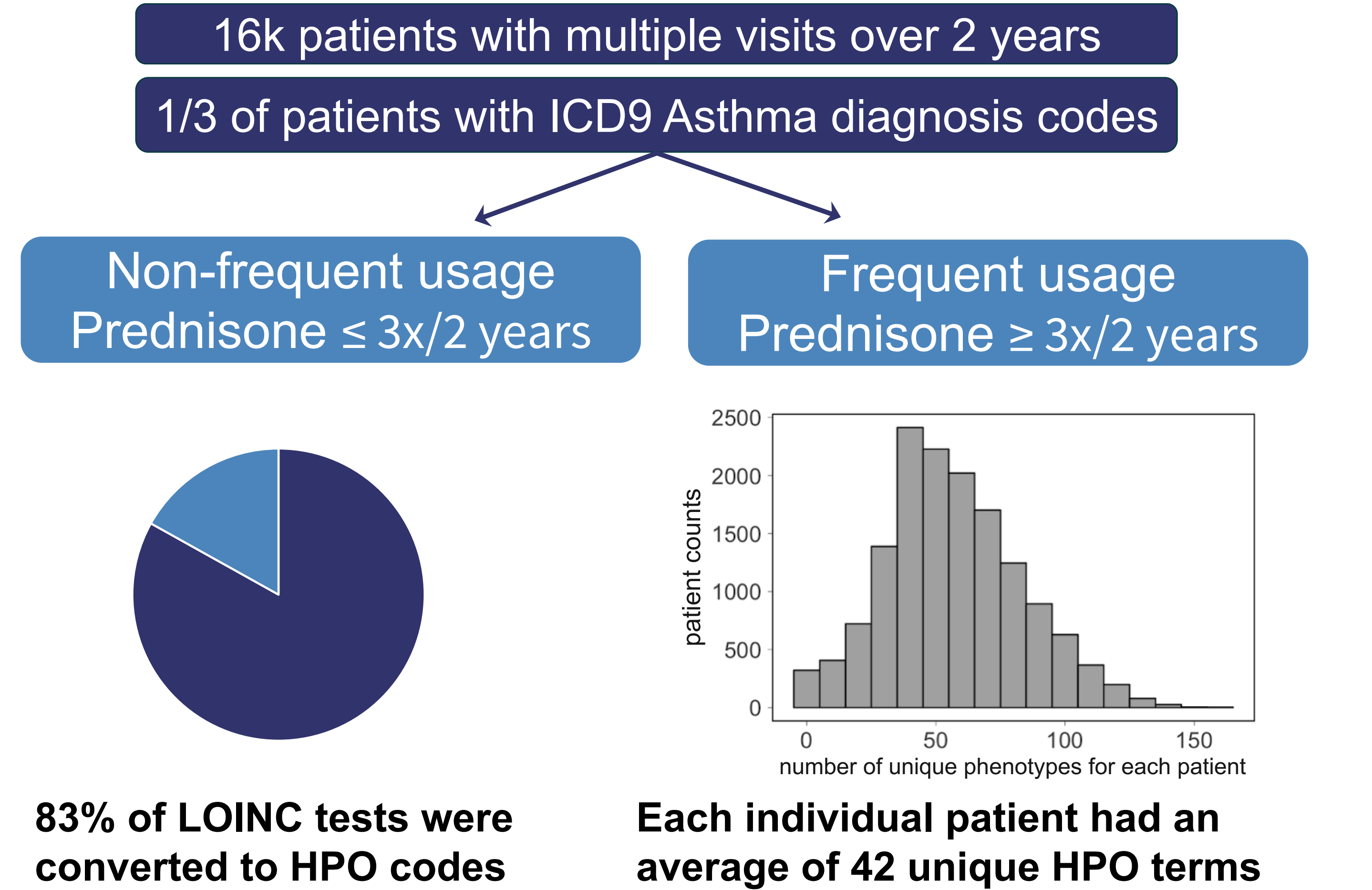*Our library transforms LOINC codes plus FHIR encoded results into HPO terms*

LOINC Type	Example code	Test Outcome	HPO term
Quantitative (Qn) Result can be low, high or normal	LOINC:6298-4 **Potassium in blood**	FHIR L	Hypokalemia
		FHIR N	NOT Abnormality of potassium homeostasis
		FHIR H	Hyperkalemia
Ordinal (Ord) Result can be absent (normal) or present (abnormal)	LOINC:5802-4 **Nitrite in Urine by test strip**	FHIR NEG	NOT Nitrituria
		FHIR POS	Nitrituria
Nominal (Nom) Result can one of a series of categories, each of which is abnormal in its own way	LOINC:5778-6 **Color of urine**	SNOMED CT 44911100 0124104	Blue colored urine
		SNOMED CT 44910100 0124102	Brown colored urine
		SNOMED CT 44909100 0124108	Green colored urine
		SNOMED CT 44908100 0124105	Orange colored urine

QUANTIFICATION OF MAPPING

Distribution of annotated LOINC terms

Distribution of HPO terms according to the number of LOINC terms mapped to an individual HPO term

PILOT STUDY: ASTHMA EHR DATA

Analysis of patient EHR data with asthma or an asthma-like condition from University of North Carolina

16k patients with multiple visits over 2 years

1/3 of patients with ICD9 Asthma diagnosis codes

Non-frequent usage Prednisone ≤ 3x/2 years

Frequent usage Prednisone ≥ 3x/2 years

83% of LOINC tests were converted to HPO codes

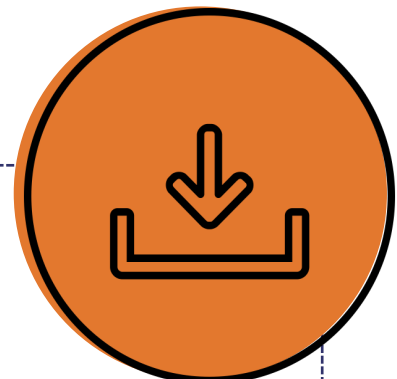**Each individual patient had an average of 42 unique HPO terms**

Example HPO terms that were significantly overrepresented in the frequent usage group

- Increased hematocrit
- Decreased mean corpuscular hemoglobin concentration
- Decreased lipoprotein lipase activity
- Hypercholesterolemia
- Reduced blood urea nitrogen
- Hyposthenuria

CONCLUSIONS AND NEXT STEPS

- LOINC2HPO can be used:
  - To identify rare disease patients and target diagnostics
  - To support phenotype-driven diagnostics
  - For discovery and mechanistic research

HUMAN PHENOTYPE ONTOLOGY
https://hpo.jax.org/app/
ANNOTATION FILE:
https://w3id.org/loinc2hpo/annotations