# Capturing Well-structured Patient Data in 5 minutes (not including questions)

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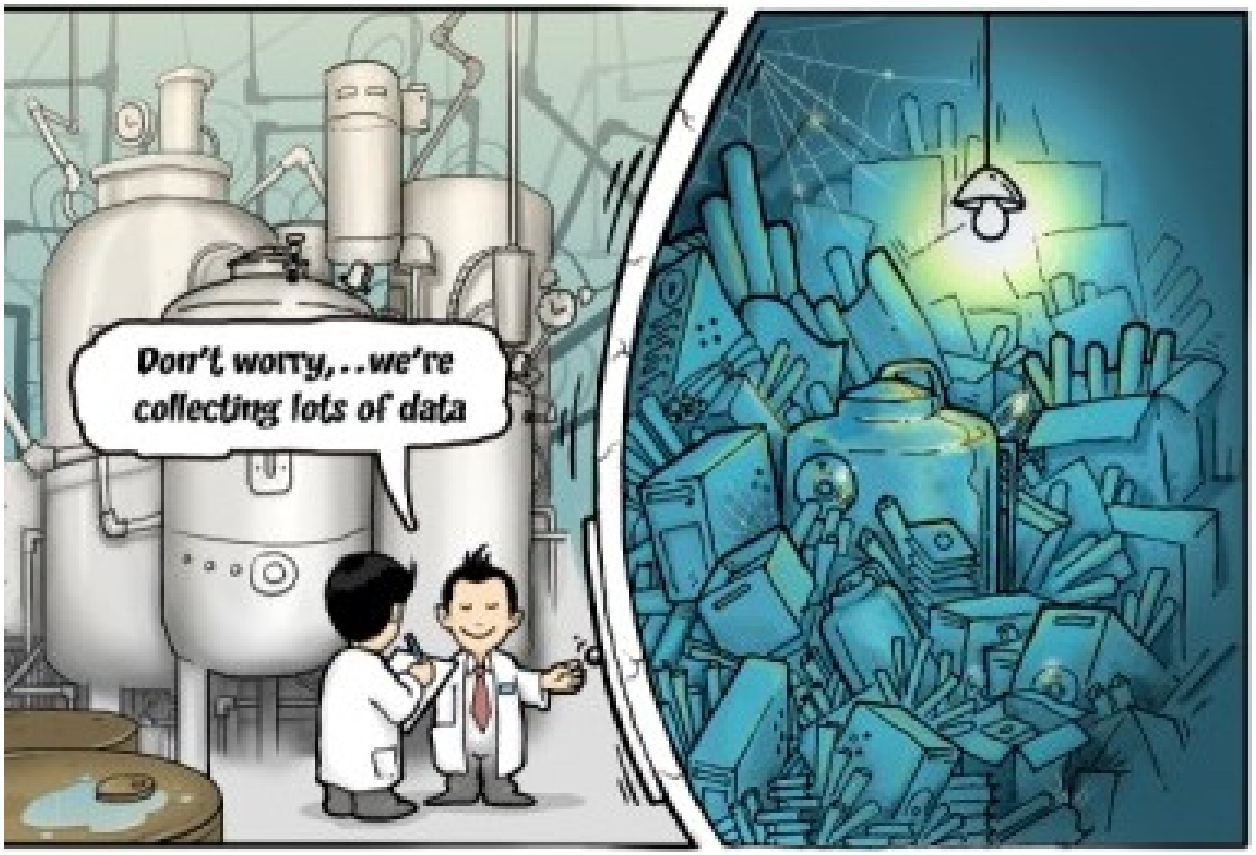
# Patient Data

- Has to come from somewhere



# Previous State of Clinical Phenotyping

- Important to collect phenotype data to cross-reference against cases observed across sites
- Currently Two Alternatives: free text or checkboxes

Dysmorphic features
- df
- dysmorphic
- dysmorphic faces
- dysmorphic features

Congenital malformation/anomaly:
- congenital anomaly
- congenital malformation
- congenital anamoly
- congenital anomly
- congential anomaly
- congentital anomaly
- cong. m.
- cong. Mal
- cong. malfor
- congenital malform
- congenital m.
- multiple congenital anomalies
- multiple congenital abormalities
- multiple congenital abnormalities

**Phenotypic description (Clinical symptoms)**

**Behavior, Cognition and Development**
- □ Global development delay
- □ Fine motor delay □ Gross motor delay
- □ Language delay
- □ Learning disability
- □ Mental retardation
  - □ Mild
  - □ Moderate
  - □ Severe
- □ Attention deficit hyperactivity disorder
- □ Autism
- □ Pervasive developmental delay
- □ Psychiatric disorders (Specify below)
- □ Other: _____

**Neurological**
- □ Hypotonia
- □ Seizures
- □ Ataxia
- □ Dystonia
- □ Chorea

**Cardiac**
- □ ASD
- □ VSD
- □ AV canal defect
- □ Coarctation of aorta
- □ Tetralogy of fallot
- □ Other: _____

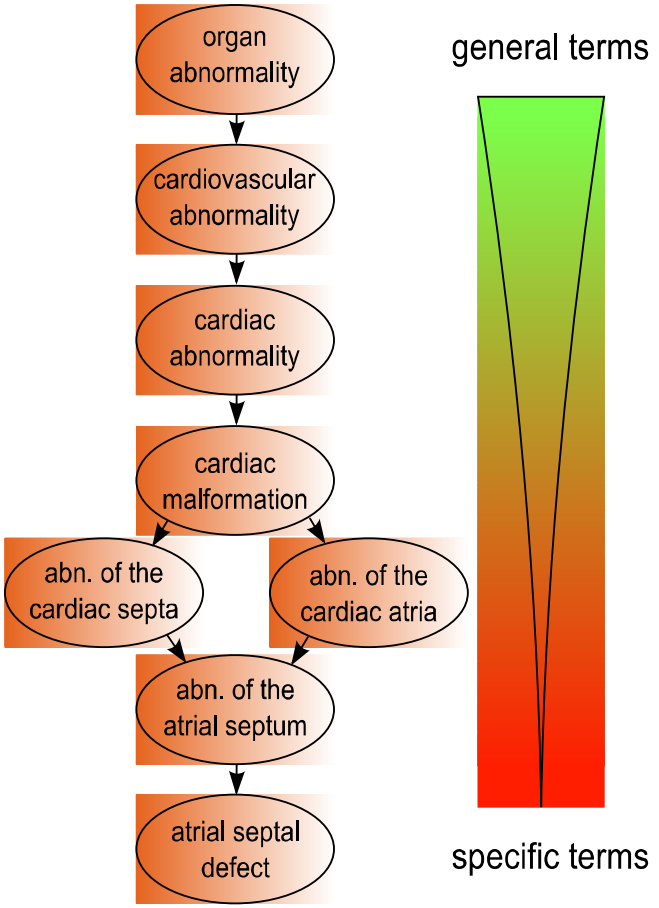
**Craniofacial**
- □ Craniosynostosis
- □ Cleft lip □ Cleft palate
- □ Microretrognathia □ Retrognathia
- □ Facial dysmorphism (Specify below)
- □ Other: _____

**Eye Defects**
- □ Blindness
- □ Coloboma
- □ Epicanthus
- □ Eyelid abnormality (Specify bellow)
- □ Other: _____

Examples of lists:
* dd. cong. malfor. behav. pro.
* dd. mental retardation
* df< delayed puberty
* dr&it
* dd df mr
* ~~mental retar.short stature~~

# Human Phenotype Ontology

- 10,200+ terms
- 57,000+ links to 5,000+ OMIM Disorders




Peter Robinson et al.

# PhenoTips: Key Features

- Ontologies are large (HPO has > 10,000 terms) and difficult to use
- Predictive Search (with synonyms): allowing rapid phenotype entry with HPO terms
- Extensive measurements (e.g. height, weight, eye distance, arm length...), with charting and automated selection of relevant phenotypic terms
- Built-in Diagnosis Assistance: suggest a list of differentials to consider
- Built-in Exam Assistance: suggest related but unselected phenotypes, so none are missed

# What PhenoTips Enables

- Fast search of previously seen patients to identify cohorts for retroactive studies
- High-throughput identification of genetic causes of disorders via mining of HPO/nosology connections
- Effective sharing of patient phenotype data between many databases (Global Alliance/IRDIRC Matchmaker project)
- Identification of second families with undiagnosed genetic disorders based on phenotype matching

Acknowledgements

**PhenomeCentral/PhenoTips Team**

Marta Girdea, Orion Buske, Sergiu Dumitriu, Andriy Misyura, Michael Kozakov

**NIH-UDP**

Neal Boerkoel, David Adams, Camilo Toro, Cyndi Tifft, et al.

# HPO & Monarch

Peter Robinson, Melissa Haendel, Damian Smedley