# Phenotype Curation in PomBase

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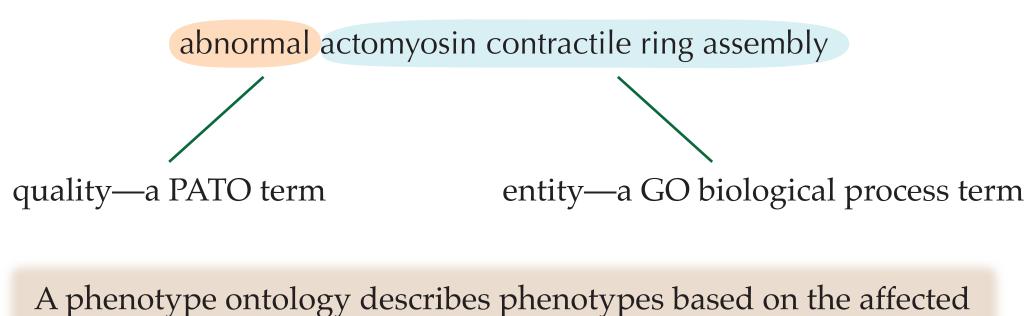
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# Introduction

PomBase, the recently established online fission yeast resource, has made the comprehensive and detailed representation of phenotypes a high priority. To support highly specific phenotype annotation, we are actively developing the FissionYeast Phenotype Ontology (FYPO), a modular ontology that uses terms from several OBO Foundry ontologies in logical phenotype definitions. Phenotype curation is featured in Canto, the PomBase community curation tool. Phenotype annotations are displayed on PomBase gene pages, and can be queried in the PomBase advanced search.

# FYPO: Fission Yeast Phenotype Ontology

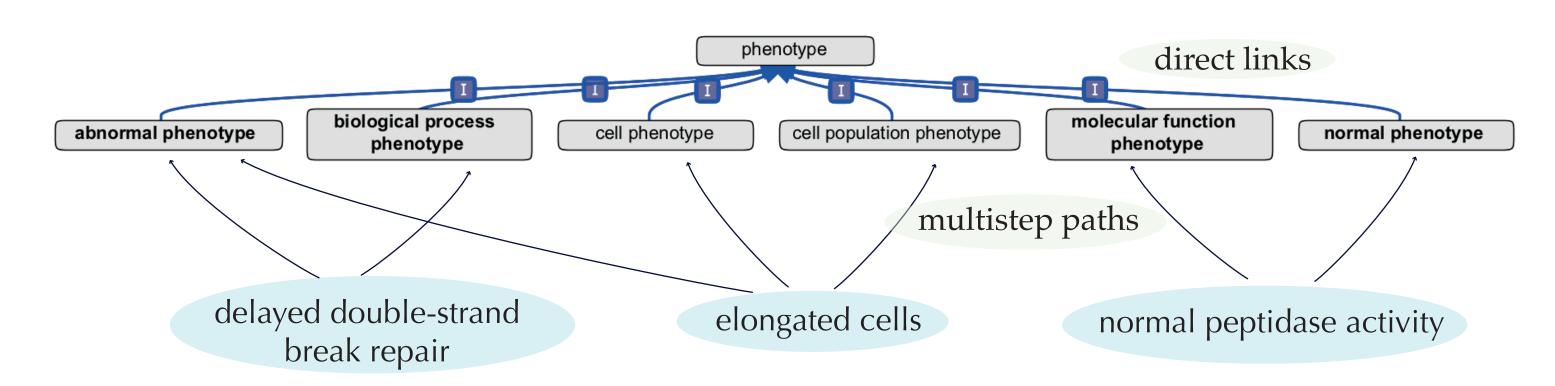
The basics: Entity–quality (EQ) definitions



A phenotype ontology describes phenotypes based on the affected entity—a structure or process—and the quality that specifies how the entity is affected. FYPO uses terms from several ontologies, including ChEBI and all three branches of GO, and the Phenotypic Quality Ontology (PATO) for qualities.

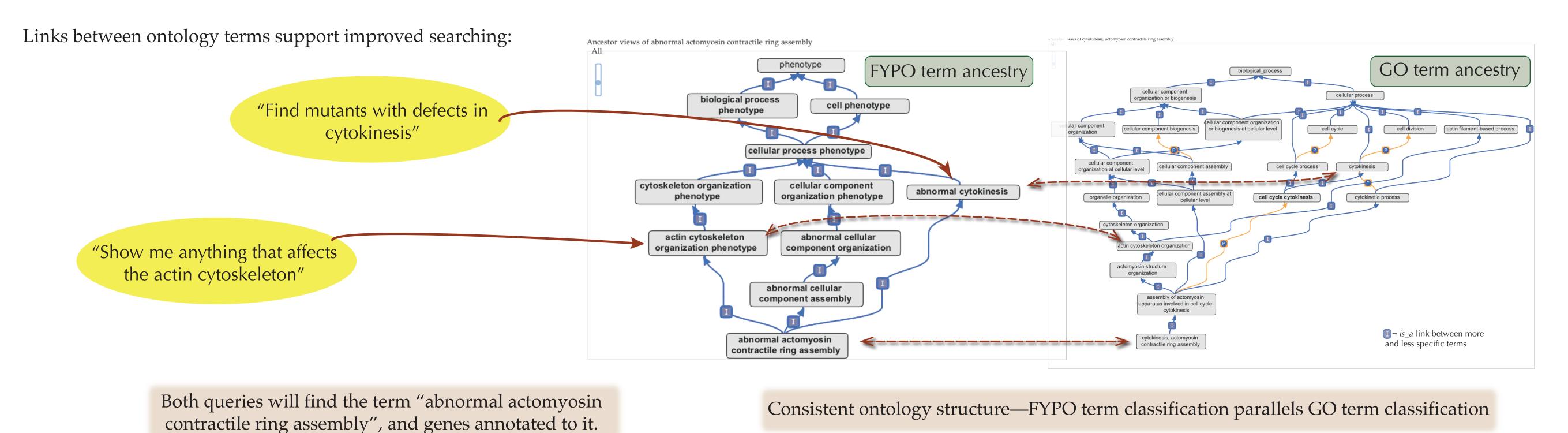
# High-level organization

- FYPO organizes terms along three axes:
  - ♦ normal vs. abnormal
    ♦ cell vs. cell population ("level")
  - ♦ biological process or molecular function (or cellular component)



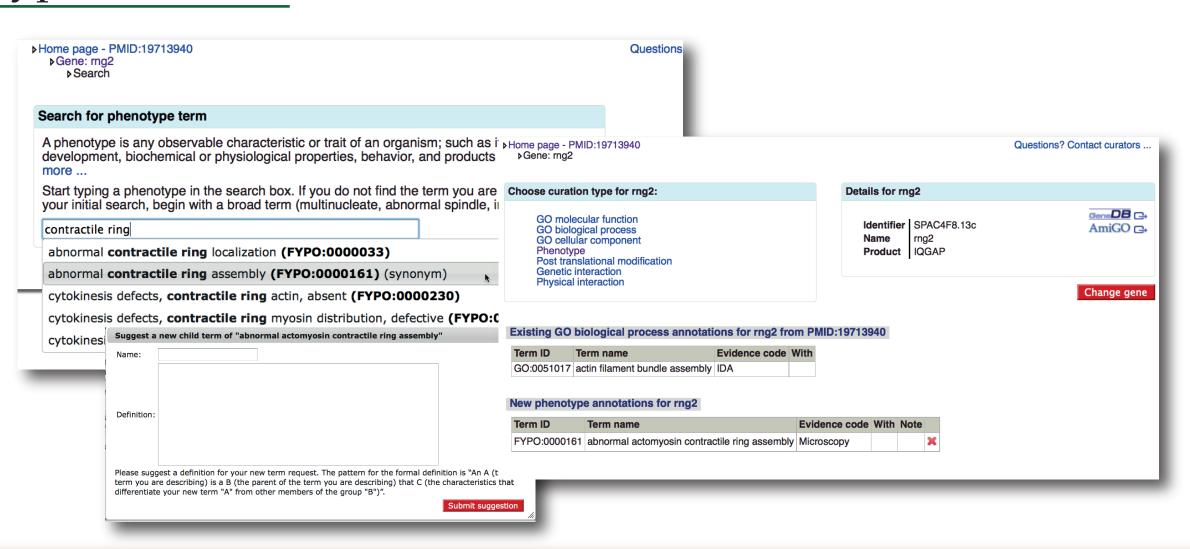
#### Using FYPO's logical structure

EQ definitions and links to other ontologies confer several benefits. Reasoning software uses EQ definitions to help maintain internal consistency, and ensure that FYPO's structure is consistent with GO, PATO and ChEBI



# PomBase Phenotype Annotation

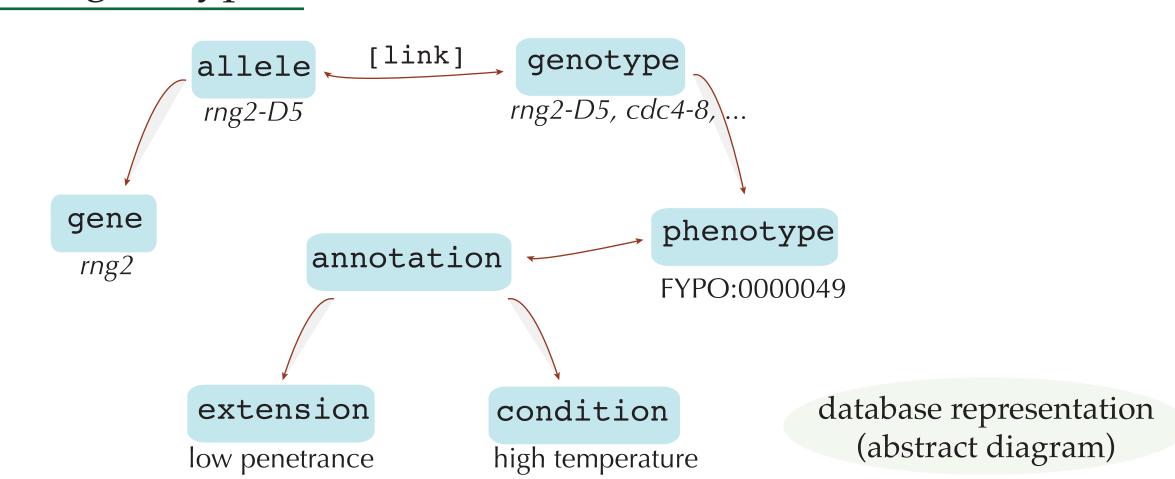
# Phenotypes in Canto



Phenotype annotation is featured in the Canto annotation tool, allowing researchers to annotate specific phenotypes using FYPO terms, based on publications. The system allows users to curate new alleles and recommend new phenotype terms for addition to FYPO.

#### **PomBase** PomBase gene page SPBC106.10 git6, sam1, sam5, sam6, sam7, to Phenotype annotation details are shown on PomBase gene pages. Each FYPO ID links to a page with term details and a list of annotated genes. FYPO:0000544 protein modification Other direct assay sam5 (G441E) (2011)amino aci abolished assayed\_using pka1 pka1delta assayed\_using pka1 Other direct assay (deletion) assayed\_using pka1 Other direct assay Gupta DR et al. (2011) sam7 (G441R) amino acid mutation Gupta DR et al. (2011) wild\_type Overexpression standard glucose FYPO:0000553 increased Microscopy not recorded pseudohyphal growth (wild type) minimal medium during cellular response FYPO:0000584 T356D (T356D) amino\_acid\_mutation standard glucose rich Gupta DR et al. (2011) Cell growth assay decreased sporulation medium

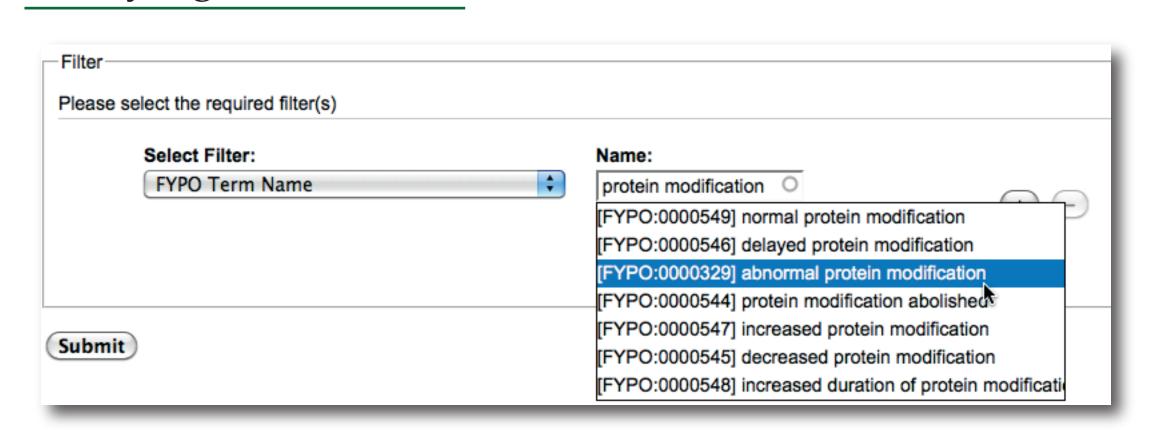
# Alleles & genotypes



Canto and the Chado database underlying PomBase support associating phenotypes with genotypes, to capture

- different phenotypes for different alleles of a single gene
- double, triple, etc. mutant phenotypes
- annotation extensions such as penetrance or expressivity

# Querying in PomBase



FYPO terms can be queried by name or ID in the PomBase advanced search. A list of annotated genes is returned, and can be used in further queries. Allele querying will be supported in the future.

# Acknowledgements