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A Case Study in Using ZFA and PATO for Describing Histological Phenotypes in the Larval Zebrafish

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When used in conjunction with an appropriate ontology of quality-bearing entity terms, such as the Zebrafish Anatomy and Development (ZFA) Ontology [1], the Phenotype and Trait Ontology (PATO) [2] permits the description of phenotypic qualities by means of a bipartite "entity-quality" (EQ) data structure. Potentially, the use of these ontologies can mitigate ambiguity in phenotypic interpretation that might otherwise occur using free-text, plain language descriptions. For example, the phrase "fin edges look ratty" ascribed to the stereomicroscopic characterization of the zebrafish insertional mutant *parvaa* is intended to describe defects in the epithelial tissue of the fin, but this might not be apparent to someone unfamiliar with such colloquialisms. A possible coercion of this phrase to the ZFA-PATO format could be expressed as "surface structure: abnormal," as it is currently described in the Zebrafish Information Network (ZFIN) database [3]. However, some precision is lost in the conversion – the researcher does not immediately know *which* surface structure is abnormal, nor does he or she know *how* abnormal it is.

Here, we will present our findings from a case study in which we have explored the use of ZFA and PATO for the description of histological phenotypes in larval zebrafish. Like the *parvaa* example, we have found that ZFA-PATO is imprecise with respect to tissue and cell type affected. Many zebrafish mutants can only be distinguished at microscopic levels of histological detail that current ZFA-PATO term combinations cannot fully describe. Based on our experience, we advocate the adoption of more highly specific terms into ZFA and PATO, perhaps based on terms "borrowed" from other model organism ontologies that currently accommodate the desired level of precision. We also propose that ontology terms (or at least their definitions) reflect the assay and resolution used (e.g., "dissecting microscope") so that researchers can easily recognize the limits of current knowledge in phenotype databases such as ZFIN. In addition, we demonstrate that even further precision (such as for describing the *extent* of abnormality) can be achieved by employing the under-utilized *modifier* term in the PATO data structure, which can allow for quantitative measurements as well as semi-quantitative "grades" not unlike those used by pathologists in characterizing disease progression. Enabling such precision gains may be the only way to describe phenotypes at the level of "granularity" needed for the forthcoming Zebrafish Phenome Project [4].

References

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