PhenoTips & PhenomeCentral: Deep Phenotyping and Data Sharing for Rare Disorders

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Despite the availability of cheap genome sequencing and continuing progress in identifying novel disease genes, a significant number of patients seen at a genetics clinic remain without a diagnosis. To help enable the accurate diagnosis of these patients, we have developed PhenoTips software for standardized patient phenotype recording and PhenomeCentral, a privacy-aware rare disease data sharing portal. PhenoTips is open source software for collecting and analyzing phenotypic information. PhenoTips closely mirrors clinician workflows, and supports recording of measurements, drawing of pedigrees and selection of phenotypes standardized using the Human Phenotype Ontology. Once recorded, data can be securely shared using PhenomeCentral: a portal that allows clinicians to find second families and closely related solved disorders while respecting patient privacy.