



Figure S1. Compilation of Personalized Gene Panel using HPO.

The cartoon panels demonstrate the procedure to generate personalized gene panel (PGP) from patient's phenotypic manifestation using HPO in Phenoxome. The left panel is a representation of the initial state of a sub-network of HPO containing relevant phenotypes. Limb hypertonia and Central Hypotonia are the reported phenotypes in the patient example, which are the primary phenotypes and are highlighted in blue. The right panel reflects the end state of the sub-network where pertinent phenotypes are retrieved and personalized gene panel generated. The phenotypes in orange are children nodes of the primary phenotypes while the phenotypes in green are the immediate parents of the primary phenotypes. Genes associated with any of the primary or secondary phenotypes are compiled in PGP, such as GPHN (Spasticity and Abnormality of Central Motor Function) and SPATA5 (Limb Hypertonia).