PerturbDB: A Database of Huntington's Disease Perturbation Studies
https://www.hdinnhd.org/

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Abstract
PerturbDB is a database of Huntington's disease (HD) perturbation studies incorporating methods, outcomes, and experimental details from >10,000 experiments curated from >1,200 publications and CHDI study reports. In addition to maintaining provenance, several classes of experimental metadata have been curated, collectively describing the nature of the: a) HD model; b) perturbation; and c) readout/results. Wherever possible, metadata are standardized according to existing ontologies and custom vocabularies. HD models are characterized as either in vitro, in vivo, ex vivo, or in silico and organisms in scope range across those described in the literature. Details about the model, such as cell line, animal strain, or cell type, are collected alongside additional experimental metadata, such as number of Q repeats in mHtt, and type of mHtt used (e.g., full length or exon 1 fragment).

Perturbation Experiment
A perturbation is defined broadly as any type of intervention applied to an HD model. In PerturbDB, the most common perturbation category is Knockdown via Gene Delivery followed by Small Molecule treatments.

Sources of Perturbation Experiments

<table>
<thead>
<tr>
<th>Source Category</th>
<th>Number of Experiments</th>
</tr>
</thead>
<tbody>
<tr>
<td>Publications (PubMed)</td>
<td>1,205</td>
</tr>
<tr>
<td>CHDI Reports</td>
<td>68</td>
</tr>
<tr>
<td>Preprints (bioRxiv)</td>
<td>4</td>
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</tbody>
</table>

Models
HD models are characterized as either in vitro, in vivo, ex vivo, or in silico and organisms in scope range across those described in the literature. Details about the model, such as cell line, animal strain, or cell type, are collected alongside additional experimental metadata, such as number of Q repeats in mHtt, and type of mHtt used (e.g., full length or exon 1 fragment).

Results are described relative to the phenotype of the HD model without perturbation.

Conclusions and Future Directions
PerturbDB enables rapid data mining of thousands of HD perturbation experiments to understand how perturbations of a single gene across a spectrum of interventional paradigms impact the HD phenotype. The integration of PerturbDB within HD Explorer places these perturbations results within a greater HD experimental context, further facilitating interpretation and hypothesis generation.

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References