

Supp Table 1 Top phenotype-gene pairs according to the hypergeometric index, based on the Orphanet dataset using the protein-target based methodology. HPO: Human Phenotype Ontology, Hyl: hypergeometric index.

HPO	HPO name	Gene Entrez	Gene Symbol	Hyl
HP:0001056	Milia	1294	<i>COL7A1</i>	16.10
HP:0001030	Fragile skin	1294	<i>COL7A1</i>	13.64
HP:0009914	Cyclopia	8928	<i>FOXH1</i>	13.22
HP:0009914	Cyclopia	84976	<i>DISP1</i>	13.22
HP:0009914	Cyclopia	7546	<i>ZIC2</i>	13.22
HP:0009914	Cyclopia	7050	<i>TGIF1</i>	13.22
HP:0009914	Cyclopia	6997	<i>CRIPTO</i>	13.22
HP:0009914	Cyclopia	6496	<i>SIX3</i>	13.22
HP:0009914	Cyclopia	4838	<i>NODAL</i>	13.22
HP:0009914	Cyclopia	2619	<i>GAS1</i>	13.22
HP:0003798	Nemaline bodies	4703	<i>NEB</i>	13.03
HP:0100245	Desmoid tumors	324	<i>APC</i>	12.68
HP:0000871	Panhypopituitarism	8928	<i>FOXH1</i>	12.68
HP:0000871	Panhypopituitarism	84976	<i>DISP1</i>	12.68
HP:0000871	Panhypopituitarism	7546	<i>ZIC2</i>	12.68
HP:0000871	Panhypopituitarism	7050	<i>TGIF1</i>	12.68
HP:0000871	Panhypopituitarism	6997	<i>CRIPTO</i>	12.68
HP:0000871	Panhypopituitarism	6496	<i>SIX3</i>	12.68
HP:0000871	Panhypopituitarism	4838	<i>NODAL</i>	12.68
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