A Silver Standard Corpus of Human Phenotype-Gene Relations

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health and biomedical informatics

Motivation

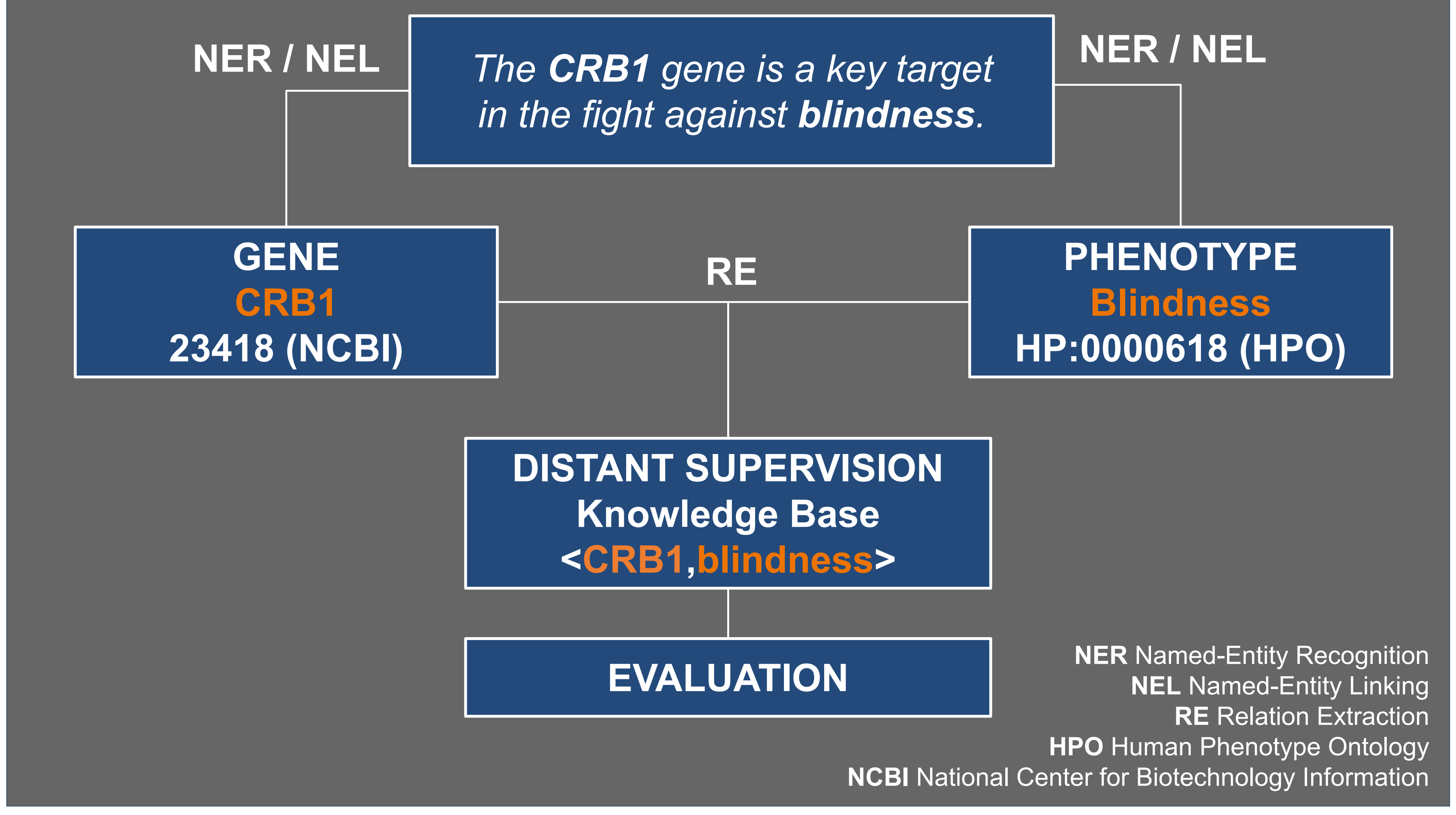
Detect the **origin of phenotypic abnormalities** and their **associated diseases** through relations expressed in biomedical literature, using Relation Extraction tools.

Relation Extraction tools require an annotated corpus and, to the best of our knowledge, there is **no corpus available** annotated with human phenotype-gene relations.

Goal

This paper presents the **Phenotype-Gene Relations (PGR)** corpus, a silver standard corpus of human phenotype and gene annotations and their relations.

Methodology



NER Named-Entity Recognition
NEL Named-Entity Linking
RE Relation Extraction
HPO Human Phenotype Ontology
NCBI National Center for Biotechnology Information

Results

Table 1. Corpus statistics. The *Known* relations are relations that are in the knowledge base and the *Unknown* relations are relations that are not yet identified or that do not exist.

Query	Abstracts	Annotations		Relations		
		Phenotype	Gene	Known	Unknown	Total
1 (10/12/2018)	1712	5676	13835	1510	2777	4283
2 (11/03/2019)	2657	9553	23786	2480	5483	7963

2 PubMed Queries
2 Named-Entity Recognition Tools:
- Minimal Named-Entity Recognizer (MER)
- Identifying Human Phenotypes (IHP)

1 HPO Knowledge Base of Gold Standard Relations
8 Curators

Table 2. The number of *Known* and *Unknown* relations selected for the test set, the number of true positives, false negatives, false positives and true negatives, and the evaluation metrics for the *Known* relations.

Relations		Marked Relations				Metrics		
Known	Unknown	True Positive	False Negative	False Positive	True Negative	Precision	Recall	F-Measure
77	143	67	86	10	57	87.01	43.79	58.26

87.58%
Inter-curator Agreement

Impact on Deep Learning

BO-LSTM System

A deep learning system that is used to extract and classify relations via long short-term memory networks along biomedical ontologies.

BioBERT Application

A pre-trained biomedical language representation model for biomedical text mining based on the **BERT** architecture.

Table 3. Precision, recall, and F-measure of the co-occurrence baseline, BO-LSTM, and BioBERT.

Method	Precision	Recall	F-Measure
Co-occurrence	35.00	100.00	51.85
BO-LSTM	69.23	42.00	52.28
BioBERT	78.95	58.44	67.16

Adaptability for the creation of other RE silver standards.

github.com/lasigeBioTM/PGR

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