

GDA Score

The DISGENET Score (S) for GDAs is computed according to:

$$S = C + M + I + L$$

where:

$$C = \begin{cases} 0.6 & \text{if } N_{\text{sources}_i} > 2 \\ 0.5 & \text{if } N_{\text{sources}_i} = 2 \\ 0.3 & \text{if } N_{\text{sources}_i} = 1 \\ 0 & \text{otherwise} \end{cases}$$

- N_{sources_i} is the number of **curated** sources supporting a GDA
- $i \in \text{CGI, CLINGEN, GENOMICS ENGLAND, CTD, PSYGENET, ORPHANET, UNIPROT}$

$$M = \begin{cases} 0.2 & \text{if } N_{\text{sources}_j} > 0 \\ 0 & \text{otherwise} \end{cases}$$

- N_{sources_j} is the number of **animal models** sources supporting a GDA
- $j \in \text{RGD, MGD, and CTD (rat and mouse)}$

$$I = \begin{cases} 0.1 & \text{if } N_{\text{sources}_k} > 0 \\ 0 & \text{otherwise} \end{cases}$$

- N_{sources_k} is the number of **inferred** sources supporting a GDA
- $k \in \text{HPO, CLINVAR, GWASCAT, GWASDB}$

$$L = \begin{cases} 0.1 & \text{if } N_{\text{pubs}} > 9 \\ N_{\text{pubs}} \times 0.01 & \text{if } N_{\text{pubs}} < 9 \end{cases}$$

- N_{pubs} is the number of publications supporting a GDA in the sources **LHGDN** and **BEFREE**