

# Proposal of genes for TREAT-panel

## Contact details from person submitting the proposal

Name:

Organization:

Email address:

## Details on the gene suggested for inclusion on the TREAT-panel

Gene name:

Associated diseases:

HGNC symbol for gene:

Orphanet-ID of diseases:

## Treatment (mandatory)

Please describe available treatment with supporting references (PMID):

## Please propose scoring according the TREATpanel selection criteria

### Disease onset

| X | Score | Definition                                                                                              |
|---|-------|---------------------------------------------------------------------------------------------------------|
|   | 2     | Predominantly paediatric onset of disease                                                               |
|   | 1     | Spectrum of onset across age groups, difficult to predict onset/limited knowledge about natural history |
|   | 0     | Mainly adult onset (>18 years)                                                                          |

### Disease severity

| X | Score | Definition                                      |
|---|-------|-------------------------------------------------|
|   | 2     | Most likely to cause significant health problem |
|   | 1     | Spectrum of severity, difficult to predict      |
|   | 0     | Not causing significant health problem          |

### Penetrance

| X | Score | Definition                       |
|---|-------|----------------------------------|
|   | 2     | Penetrance > 80%                 |
|   | 1     | Intermediate penetrance (20-80%) |
|   | 0     | Low penetrance (< 20%)           |

### Clinical validity

| X | Score | Definition                                                                                            |
|---|-------|-------------------------------------------------------------------------------------------------------|
|   | 2     | Known pathogenic variants with clear genotype-phenotype correlation                                   |
|   | 1     | Genes with known pathogenic variants and partial genotype/phenotype correlation (as in ultrarare dis) |
|   | 0     | Genes with only benign or variants of unknown significance, no established geno/phenotype correlation |

Additional comments: