Pre-Submission Communications with URGenT Program Staff

Please provide the following preliminary information and supporting data as instructed by the relevant Funding Opportunity Announcement (FOA) you plan to submit your proposal to:

PAR-22-030: Translational Efforts to Advance Gene-based Therapies for Ultra-Rare Neurological and Neuromuscular Disorders (U01 - Clinical Trial Optional)

PAR-22-028: Ultra-Rare Gene-based Therapy (URGenT) Network Resource Access (X01, Clinical Trial Not Allowed)

This information will enable us to determine your fit and eligibility for the proposed FOA and to provide you with further guidance on program scope, goals, and developing objectives.

Note: This form must be completed *six weeks* before application receipt date. Potential applicants interested in discussing an application with program staff are strongly encouraged to do so six weeks before receipt date. Any correspondence within six weeks of receipt date may only be completed by email.

Section 1: Submitter Information

Submitter Name:

Institution:

PI Name and Institution (if different from submitter):

Primary Contact email address:

Team Management [List major collaborators and their expertise]:

Section 2: Proposed Study Information

Do you plan to apply to X01 URGenT FOA?

☐ Yes ☐ No

OR

Do you plan to apply to U01 URGenT FOA?

☐ Yes ☐ No

Expected Date of Submission:

Draft Title of Proposal:

Clinical Syndrome/Disorder:

Type of Proposed Therapy: Choose One

If *Other*, please explain:

Section 3: Patient and Disease Information

Disease Prevalence:

Estimated number of patients with same genetic diagnosis:

Estimated number of patients with same genetic change:

Predicted Disease Trajectory:

☐ Rapidly Progressing

☐ Slowly Progressing

Section 4: Genetics

Gene Name:

Gene ID:

Pathogenic Human Genetic Variant:

Transition point mutation:

☐ C → T; ☐ G → A; ☐ A → G; ☐ T → C

Transversion point mutation:

☐ A → C; ☐ C → A; ☐ G → C; ☐ T → A

☐ A → T; ☐ C → G; ☐ G → T; ☐ T → G

☐ Deletion
☐ Duplication
☐ Copy Number Loss
☐ Copy Number Gain
☐ Insertion
☐ Insertion and Deletion
☐ Other:

Predicted Consequence of Genetic Change:

- ☐ Gain of Function
- ☐ Dominant Negative
- ☐ Partial Loss of Function
- ☐ Total loss of function
- ☐ Unknown

Section 5: Information to Support Proposed Application

Background and Supporting Data: (With a focus on explaining how the data meets the entry criteria for the PAR of interest; you may include relevant data and key citations that support your data)

Specific Aims:

Research Strategy:

Proposed budget by year:

Budget justification:

Have you had prior Regulatory Interactions/feedback?

☐ Yes ☐ No

Is there availability of patient-derived cell lines?

☐ Yes ☐ No ☐ Not Applicable

Please explain, if applicable:

Is there availability of animal model(s)?

☐ Yes ☐ No ☐ Not Applicable

Please explain, if applicable:

Intellectual Property (Briefly explain your freedom to operate):

Is there any other relevant information you would like to add?