

**ACMG SECONDARY FINDINGS PANEL NOMINATION FORM**

**(to Add or Remove a Gene)**

**Submit completed forms to:** [**acmg@acmg.net**](file:///C:\Users\mlyon\AppData\Local\Microsoft\Windows\INetCache\Content.Outlook\SHNNWHNG\acmg@acmg.net)

**SUBMITTER INFORMATION**

|  |  |
| --- | --- |
| Date | Click here to enter text. |
| Name | Click here to enter text. |
| Email | Click here to enter text. |
| Address | Click here to enter text. |
| Title | Click here to enter text. |
| Phone number | Click here to enter text. |
| Co-sponsoring organization (if applicable) | Click here to enter text. |

**BACKGROUND**

**As you submit a nomination, please keep in mind the goals of the Secondary Findings Gene Panel:**

* **Genes should be medically actionable. “Actionability” means that a medical or surgical intervention is available that has demonstrated effectiveness to alter the course of the disease process itself.**
* **Genes should have a clear phenotype associated with deleterious mutations. The phenotype(s) and penetrance will need to be described and at least one of the phenotypes should have serious medical implications.**
* **A gene may be associated with more than one phenotype. The actionability of a gene is in relation to a particular phenotype.**

**GENE, CONDITION(S) AND INTERVENTION(S)**

Please complete the following section about the gene you are nominating for adding/removing and include references to support the information you provide. If the gene is associated with more than one phenotype, please consider the most severe phenotype for answering questions related to actionability.

|  |  |  |  |
| --- | --- | --- | --- |
| **Add or Remove?** | **Gene Symbol** | **Gene Name** | **OMIM#** |
| Choose an item. | Click here to enter text. | Click here to enter text. | Click here to enter text. |
| **Phenotype(s)/Condition(s)** caused by pathogenic variants in this gene that are associated with an alternative clinical manifestation that has low morbidity (e.g., is not life threatening) and therefore would not be reported in a secondary finding list. Only the phenotype with the most significant morbidity will be reported on subsequent pages. | | | **OMIM Phenotype #** |
| 1. Click here to enter text. | | | Click here to enter text. |
| 2.Click here to enter text. | | | Click here to enter text. |
| 3.Click here to enter text. | | | Click here to enter text. |

|  |  |
| --- | --- |
| **PHENOTYPE (Consider the phenotype with the most significant morbidity)** | **OMIM Phenotype #** |
| Click here to enter text. | Click here to enter text. |

**CLINICAL FEATURES**

|  |  |
| --- | --- |
| How is the diagnosis routinely made clinically? | Click here to enter text. |
| How frequently would diagnosis be missed in the context of routine clinical care by a general pediatrician/internist? | Click here to enter text. |
| Is the condition included in the consensus panel for newborn screening? | Click here to enter text. |
| Fraction of patients with this phenotype attributable to this gene | Click here to enter text. |
| Range in age of onset | Click here to enter text. |
| Average age of onset | Click here to enter text. |
| Prevalence (Across all ethnic groups in the US) | Click here to enter text. |
| Is it more common in any specific ethnic groups? If so, which one and how frequent? | Click here to enter text. |
| Penetrance (provide age specific penetrance, if available) | Click here to enter text. |
| Please indicate any references to support the information about clinical features | Click here to enter text. |

**MOLECULAR GENETIC FEATURES**

|  |  |
| --- | --- |
| Inheritance Mode (e.g., AD, AR, X- linked, mitochondrial) | Click here to enter text. |
| Frequency of spontaneous mutations (common/rare) | Click here to enter text. |
| Types of mutations (e.g. frameshift, nonsense) known to have deleterious impact | Click here to enter text. |
| Are there well characterized founder mutations? (if so, please list) | Click here to enter text. |
| List curated databases to classify variants | Click here to enter text. |
| Please indicate any references to support the information about molecular genetics | Click here to enter text. |

**CLINICAL GENETIC TESTING**

|  |  |
| --- | --- |
| What fraction of mutations are identifiable on routine exome sequencing? | Click here to enter text. |
| Is there an available confirmatory, orthogonal diagnostic test? | Click here to enter text. |
| If so, what is the test? | Click here to enter text. |
| How readily available is the confirmatory diagnostic test at most medical centers? | Click here to enter text. |
| What does the test entail (blood, imaging, biopsy)? | Click here to enter text. |
| How sensitive is the confirmatory diagnostic test? | Click here to enter text. |
| How specific is the confirmatory diagnostic test? | Click here to enter text. |
| Please indicate any references to support the information about clinical testing | Click here to enter text. |

**ACTIONABILITY**

|  |  |
| --- | --- |
| **1. Severity**(What is the nature of the morbidity for an individual carrying a clearly deleterious allele in this gene? Consider the most severe outcome for those gene-disorder pairs with multiple outcomes.) | |
| Risk of sudden death? (Y/N) | Click here to enter text. |
| Reasonable possibility of death or major morbidity? (Y/N) | Click here to enter text. |
| Modest morbidity? (Y/N) | Click here to enter text. |
| Please describe the morbidity or threat to health related to this phenotype (If any) | Click here to enter text. |
| **2.** **Likelihood of disease**: What is the lifetime percent chance that if a deleterious mutation is present the morbidity noted above will materialize? Click here to enter text. | |
| **3. Intervention (Prevention or Treatment):** Only specific medical or surgical interventions known to ameliorate risk will be considered (e.g. we do not consider general “lifestyle” and behavioral changes that are generally recommended to individuals in many contexts). | |
| Are there effective and available interventions that will mitigate or ameliorate the threat of the outcome above? (Y/N) | Click here to enter text. |
| Please describe the intervention(s) to ameliorate this phenotype and their effectiveness | Click here to enter text. |
| Please indicate any references to support improved outcomes for patients treated with the indicated intervention | Click here to enter text. |