Summary Annotations Help File

The set of files comprised of **summary_annotations.tsv**, **summary_ann_alleles.tsv**, **summary_ann_evidence.tsv** and **summary_ann_history.tsv** contain ClinPGx's summary annotations (a.k.a. clinical annotations) and associated information. These annotations are manually created by the ClinPGx curators to provide an evidence-rated, genotype- or allele-based summary of the literature evidence annotated in ClinPGx for an association between a genetic variant and a drug. Please refer to the ClinPGx website for more information about summary annotations, and how they are assigned a level of evidence based on scores.

Description of Files:

- **summary_annotations.tsv**: Contains all of the meta-data about each summary annotation.
- **summary_ann_alleles.tsv**: Contains the genotype- or allele-based annotation text and CPIC-assigned allele function, if available.
- **summary_ann_evidence.tsv**: Contains information about each supporting annotation (variant annotation, guideline annotation, label annotation) for every summary annotation.
- **summary_ann_history.tsv**: Contains the history of the summary annotation, including the creation date and the dates of changes or updates to the annotation.
- **LICENSE.txt**: The ClinPGx license for using ClinPGx data, including summary annotations.
- **CREATED_xxxx-xx-xx.txt**: This file indicates the date that all files in this group were created from the database.
- **README.pdf** file: This document.

A description of the fields in each file follows.

summaryl_annotations.tsv:

- ***SummaryAnnotation ID***: The unique ClinPGx ID number for the annotation.
- ***Variant/Alleles***: Variant rsID from dbSNP or the allele names.
- ***Gene***: HGNC gene symbol.
- ***Level of Evidence***: Levels 1A-4 with 1A being the highest level of evidence; more information found on ClinPGx.
- ***Level Override***: Description of whether the level of evidence assigned based on the summary annotation score was changed by ClinPGx curators. Options: Yes (plus reason), No.
- ***Level Modifiers***: Description of extra information used when assigning level of evidence. Options: VIP Tier 1, rare variant.
- ***Score***: Summary annotation score calculated from supporting annotations; more information found on ClinPGx.
- ***Phenotype Category***: Association phenotype. Options: toxicity, efficacy, dosage, metabolism/PK, PD, other.
- ***PMID Count***: The number of PMIDs with variant annotations used to support the summary annotation.
- ***Evidence Count***: Number of annotations supporting the summary annotation, including variant annotations, guideline annotations and drug label annotations.
- ***Drug(s)***: Drugs associated with the variant/allele.
- ***Phenotype(s)***: Phenotypes in the variant/allele-drug association. For example, if the association was found in patients with a particular phenotype (disease), or if the variant/allele-drug combination causes a particular phenotype.
- ***Latest History Date***: The date of creation of the summary annotation or the last time it was updated.

- ***URL***: ClinPGx webpage for the summary annotation.
- ***Specialty Population***: Description of a specialty population (e.g. 'Pediatric') in any supporting variant annotation.

Example row from summary_ann_alleles.tsv file:

Summary Annotation ID	Variant/Haplotypes	Gene	Level of Evidence	Level Override	Level Modifiers	Score	Phenotype Category	PMID Count	Evidence Count	Drug(s)	Pher
1447954390	rs75039782	CFTR	3	Yes: Level of evidence set to 3. Ataluren is a drug for the treatment of Duchenne muscular dystrophy caused by a nonsense mutation and not indicated in CF treatment.	Rare Variant; Tier 1 VIP	4	Other	2	2	ataluren	Cysti Fibro

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summary_ann_alleles.tsv:

- ***Summary Annotation ID***: The unique ClinPGx ID for the annotation.
- ***Genotype/Allele***: The genotype or allele associated with the clinical phenotype in the next column.
- ***Annotation Text***: The summary annotation for the given genotype or allele.
- ***Allele Function***: The CPIC allele function, if it has been assigned; more information found on ClinPGx.

Example rows from summary_ann_alleles.tsv file:

Summary Annotation ID	Genotype/Allele	Annotation Text	Allele Function
613979022	CC	May be less likely to have improved left ventricular ejection fraction after carvedilol treatment.	

1183615480	*3	Patients carrying the CYP2D6*3 allele in combination with another no function allele may have decreased metabolism of carvedilol as compared to patients carrying two normal function alleles. This annotation only covers the pharmacokinetic relationship between CYP2D6 and carvedilol and does not include evidence about clinical outcomes. Other genetic and clinical factors may also influence carvedilol metabolism.	No function

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summary_ann_evidence.tsv:

- ***Summary Annotation ID***: The unique ClinPGx ID for the annotation.
- ***Evidence ID***: The unique ClinPGx ID for the annotation supporting the summary annotation, including variant annotations, guideline annotations and drug label annotations.
- ***Evidence Type***: The type of supporting annotation. Options: Variant Annotation (Drug), Variant Annotation (Phenotype), Variant Annotation (Functional Assay), Guideline Annotation, Label Annotation.
- ***Evidence URL***: ClinPGx webpage for the supporting annotation.
- ***Evidence PMID***: If the supporting annotation is a variant annotation, the PMID the variant annotation is based on; otherwise, blank.
- ***Evidence Summary***: Variant annotation text or description of the guideline or label.
- ***Study Parameter Used for Scoring***: The ID of the Study Parameters object used to determine score, only applicable to Variant Annotations
- ***Evidence Score***: The score of the supporting annotation.

Example row from summary_ann_evidence.tsv file

Summary Annotation ID	Evidence ID	Evidence Type	Evidence URL	Evidence PMID	Evidence Summary	Evidence Score
449717935	1449717924	Variant Drug Annotation	https://clinpgx.org/variantAnnotation/1449717924	30136624	Genotype GG is associated with increased response to Opioid anesthetics, Other general anesthetics or volatile anesthetics as compared to genotypes	3

AA + AG.

summary_ann_history.tsv:

- ***Summary Annotation ID***: The unique ClinPGx ID number for the annotation.
- ***Date (YYYY-MM-DD)***: The date of the history event.
- ***Type***: The type of the history event. Options: Create, Update, Note, Correction.
- ***Comment***: The comment entered by the ClinPGx curator describing the action taken on the summary annotation; this field may be blank.

Example row from summary_ann_history.tsv file:

Summary Annotation ID	Date (YYYY-MM-DD)	Type	Comment
1450931822	2021-01-29	Update	Edited phenotype descriptions to include CPIC 'no recommendation'.

It is important to understand that summary annotations are created from literature that has been curated by ClinPGx. There may be more literature in the public domain to support or contradict an association that is not in the ClinPGx database. ClinPGx manually curates high profile literature but does not contain curated literature from every domain-based journal, or all of PubMed. ClinPGx reviews evidence from curated literature in non-regular intervals and re-evaluates the evidence strength for each association as more literature becomes available.