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Authoring and Peer Review

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Medical Genetics Summaries (MGS) for pharmacogenetics is a freely available collection of articles describing how genetics plays a role in an individual's response to drugs or predisposition to disease. The structured format of each summary makes accessing information such as genetic testing or therapeutic recommendations quick and easy to use. Medical Genetics Summaries use authoritative sources, are guideline-driven and actionable, and are subject to an extensive review process as described below.

Editorial Oversight

The editors of Medical Genetics Summaries advise on subject matter, guide the project through developments in the field, provide final approval prior to the publication of each summary, and assist in recruiting reviewers and in resolution of key issues which may be raised during the review process.

Selection of Topics

The selection of topics for new MGS chapters is guided by two factors. First, the author consults the FDA's "Table of Pharmacogenomic Biomarkers in Drug Labeling" to select new drugs that have not yet been covered in MGS. Second, to prioritize the order of new MGS chapters, the author checks the Genetic Testing Registry (GTR) for drug response records which contain information about genetic testing, but lack summary information about the drug response. After a new MGS chapter is released to the production site, an excerpt from the chapter is displayed in the relevant GTR drug response record. Additional reciprocal links between MGS, GTR, and MedGen are also added.

Structured Format

Each MGS drug response chapter follows a structured format. Each summary has one drug section, but may have one or more gene sections, depending on how many genetic factors have been identified.

1. Introductory paragraphs detail the drug and its uses, how the genetic variants influence an individual's response to the drug, and displays dosing recommendations from the FDA and practice guidelines from authoritative professional societies.
2. The drug section begins with a description of the drug, the drug class, its mechanism of action, the indications for its use, and common side effects. This is followed by a discussion on the factors which influence the drug response.
3. The gene section reviews important facts about the gene — what role it plays in the drug metabolism or action, and the nature of the gene variants and how they impact the drug response. The common or clinically significant variants are then discussed, including their prevalence across different ethnic populations.

4. "Genetic Testing" section is a key part of the summary. Here, the summary clearly describes the genetic testing options that are available, linking to genetic test providers listed in GTR.
5. "Therapeutic Recommendations based on Genotype" excerpts clinically actionable information, e.g. dosing recommendations from the FDA drug label; and therapeutic recommendations from pharmacogenetic societies such as CPIC, CPNDS and DPWG and medical societies, such as ASCO, ACMG, NCCN.
6. Nomenclature table provides information about the different terms used for genetic variants. Terms that are commonly used in the literature and historic terms are linked to the official HGVS terms and rs identifiers when available.
7. Expert reviewers are acknowledged, and information about previous versions of the summary is given.

Writing Process

Each summary is written by our in-house senior medical writer, who is an MD. All phases from authoring to production are tracked in an internal ticket management system. To create the first draft of a summary:

1. The author consults the most recent FDA drug label for the drug. To gain a better understanding for the context of the drug use and impact of genetic factors, the author will use NIH resources and other clinical sites, such as UpToDate.
2. The author then identifies key guidelines and primary papers, using PubMed Clinical Queries, PubMed, CPIC and PharmGKB.
3. Finally, the author searches PubMed for the most recent publications — to both find content that has not yet been cited by guidelines, and to identify external reviewers who are actively involved in research.

Internal Review

Each summary undergoes internal review involving one or two NCBI staff members. Once the author has finalized the first draft of a summary, it is submitted for internal review, along with key supporting guidelines (e.g., FDA drug label, key guidelines). The internal reviewers perform the first round of expert review, using track changes to ask questions and make suggestions and corrections. Because this process occurs in a ticket management system, all versions of the document and comments from the author and reviewers are documented.

External Review

Following internal review, each summary goes through a scientific peer-review process involving between 2 to 9 experts from outside NCBI. Typically, the external review includes at least one individual who is a member of CPIC, and a clinical specialist, experienced in prescribing the drug and has published papers about its use. Expert reviewers comments are tracked so that the evolution of the summary can be seen, and after the summary is released to production, all versions of the summary are stored in the document management system.

Finalizing the Summary

Once all the review comments are reconciled, the summary is copyedited in-house and released to production.

Updates

Summaries are scheduled to be updated every 2 years. An earlier update is triggered by an update to guidelines from which excerpts have been taken for the summary. The internal reviewers decide whether the nature of the updates is minor or major. All minor updates undergo internal review and copy editing, and when published —

a link to the previous version of the summary is made available. For major updates, the summary is sent out for external review.

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