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Genetic testing registry

The Genetic Testing Registry aims to simplify searching by allowing users to browse through subcategories or utilize their browser's Find function. The registry itself does not contain individual test results but provides information on available tests, their indications, accuracy, and usefulness. Benefits for various stakeholders include expanding publicly accessible data, identifying knowledge gaps, facilitating collaborations among test providers, understanding trends in genetic testing, and streamlining billing processes. The National Institutes of Health developed the registry due to its role as a primary Federal agency for conducting and supporting medical research. The Genetic Testing Registry is integrated with other NIH resources to cater to researchers, clinicians, consumers, and patients. Its intended audience includes healthcare providers and researchers initially, with potential future features catering to patients and the general public. The registry's overarching goal is to advance public health and research into genetic conditions. Key functions include encouraging test providers to share information about their tests' scientific basis and utility, providing an informational resource for locating laboratories offering specific tests, and facilitating data-sharing for research and new discoveries. The Genetic Testing Registry (GTR) is an online tool that provides comprehensive information on genetic and infectious disease tests. It enables healthcare providers, researchers, and others to navigate the rapidly evolving landscape of genetic testing. The Genetic Testing Registry (GTR) is integrated with other National Institutes of Health databases and resources. Test providers can contribute test information to GTR, including U.S. and international clinical and research laboratories. Individuals from a laboratory can register tests using the 'Groups' function, allowing multiple individuals to submit lab and test data for a single laboratory. Research tests can also be registered in GTR, with instructions provided on how to enroll and get started. Somatic tests can be registered in GTR, with test targets identified as germline or somatic. Custom panels can be registered as separate tests, with options for large panels with all possible genes or phenotype grouping. Laboratory information must be entered first before submitting a test for a GTR ID. The review process takes two to three business days, and once approved, test information can be submitted. Test information becomes available on the public site within 24-48 hours after submission. GTR aims to bring transparency to genetic testing by ensuring that a test is only registered once. Laboratories performing at least part of the test should register it, with send-out tests not requiring registration. Individuals can access their account in the GTR Submission Portal for step-by-step instructions. Q: Help! I lost my login information for myNCBI account (to access GTR). A: Click "login/sign in" in the upper right-hand corner; Below the blue "sign in" button, click the link “Forgot NCBI username or password?”. Follow the instructions to get your username or reset your password. For more help, please contact NCBI staff at tryhd@ncbi.nlm.nih.gov. Q: What information is in scope for GTR? A: The GTR contains information about clinical and research tests for Mendelian disorders and drug responses as well as somatic/cancer tests. Labs that offer whole exome sequencing and whole genome sequencing can be found by searching Laboratory services or via links from the home page and elsewhere. Q: What determines the order of search results? A: Relevance of the search term and completeness of the submitted data determine the order within lists of results. Q: How can I learn how to use GTR? A: Get started by viewing GTR Tutorials and reading the How to use GTR help page (both accessible from the Home page). Learn how to quickly find tests, compare labs, filter your results, find GeneReviews, and set preferences. Search the help pages for the information you need. Q: Starting from GeneReviews, how do I find tests in GTR? A: New links to testing information in GTR are located along the right-hand side of GeneReview summaries, labeled ‘Tests in GTR by Gene’ and ‘Tests in GTR by Condition’. Q: How can I find a test that is no longer in GTR? A: Laboratories can remove tests from GTR at any time. When a laboratory removes a test from their test menu, that test will not display in GTR anymore. Q: How can I find previous versions of a test? A: GTR test accessions have a total of 9 digits; ‘GTR’ prefix followed by 9 digits and include the unique GTR ID (GTR000). If the GTR ID is less than 6 digits, replace digit with 0. Q: What has been the level of submitter participation in GTR thus far? A: For a current count of all labs and tests registered in GTR, click here. Q: How is the COVID-19 pandemic affecting lab accreditations and renewals? A: Due to the COVID-19 pandemic, some entities are experiencing delays with their protocols (e.g., inspections), and lab certifications and accreditations remain valid past the due date until the process is completed. For other entities or state agencies with specific accreditation inquiries, kindly refer directly to them for updates on their status. Q. What does "classic" view represent? A. In April 2024, GTR introduced a new viewer for public website access to clinical and research genetic tests. The previous "classic" view will remain accessible via a link at the top of the page, labeled "Classic view of this page." Tests related to microbe detection will only be displayed in classic view. Q. How are labels for clinical categories assigned to tests? A. Please consult the documentation provided on Clinical test categories. Q. What action would the National Institutes of Health (NIH) take if test submitters fail to review submitted information at least once annually? A. Submitters agree to adhere to a code of conduct, requiring them to review and update their submissions as necessary at least once a year. The NIH's National Center for Biotechnology Information sends an annual reminder about this obligation. If this stipulation is not honored by a test submitter, information more than one year old will be labeled out of date. NIH reserves the right to take further action, including removing outdated submissions from the GTR if the information becomes two years beyond the last annual review. Q. What steps would the National Institutes of Health (NIH) take if it discovers submitted test information is inaccurate or misleading? A. Submitters agree to abide by a code of conduct, stipulating that they uphold the integrity of the GTR through submissions of accurate and non-misleading information. If this condition is not met by a submitter, NIH may take action, which includes requesting that submitters amend incorrect or misleading information in its sole discretion. If submitters fail to respond to such requests, NIH may remove relevant submissions from the GTR. Q. What would happen if marketing materials claim NIH endorses a test? A. Submitters agree to abide by a code of conduct, prohibiting them from making explicit or implicit claims that their listed tests in the GTR have been approved or endorsed by NIH, the Department of Health and Human Services, or the U.S. Government. If this stipulation is not honored, NIH may take action, including requesting submitters revise their marketing materials to delete such claims or remove relevant submissions from the GTR in its sole discretion. Marketing materials can refer to the fact that test information is provided within the GTR. Q. How will NIH ensure the quality of test information provided to the Genetic Testing Registry? A. NIH does not verify the content of submitted information by test providers. Submitters are solely responsible for the content and quality of their submissions to the GTR. However, NIH acknowledges its critical role in recognizing and addressing potential inaccuracies or misinformation within the registry. NIH clarifies that it does not endorse tests or verify accuracy of GTR information The National Institutes of Health (NIH) has implemented measures to inform users about the limitations of the Genetic Testing Registry (GTR). The National Center for Biotechnology Information (NCBI), which manages the GTR, checks for administrative errors during submission and requires submitters to follow a code of conduct. A prominent disclaimer on the GTR homepage states that NIH does not independently verify information submitted to the registry. To improve transparency, NCBI assigns unique accession numbers to each test, allowing for uniform references in scientific publications and facilitating third-party evaluations. The registry also links published reviews to referenced tests and provides access to external resources such as professional practice guidelines and studies supporting or refuting claims made by submitters. Users can report incorrect information to NCBI staff. NIH has engaged stakeholders to shape the GTR, including defining its scope, improving navigation, enhancing usability, and identifying important data elements. The registry's initial phase focuses on single-gene tests for heritable mutations, pharmacogenomic tests, and multiplex panels and arrays. Future phases will add tests for somatic mutations, infectious agents, and direct-to-consumer genetic tests. NIH has considered the needs of healthcare providers as the primary audience for the initial phase, with plans to expand the audience in future phases. Certain data elements, such as test price and patent information, have been excluded from the initial phase. The development of the Genetic Testing Registry (GTR) was a collaborative effort between test developers, manufacturers, and healthcare providers. This collaboration ensured that the GTR's utility would be optimized through continued stakeholder interaction. Stakeholders provided input through various means, including: - A Request for Information (RFI) in June 2010, which garnered 68 public comments. - A Public Stakeholder Meeting on November 2, 2010, resulting in 17 public comments and meeting discussion. - Two Federal Register notices in July and November 2011, collecting a total of 24 public comments. - Consultations with two GTR clinical advisory groups, consisting of four meetings with NIH experts and six meetings of the medical genetics working group. - Meetings with government agencies interested in genetic testing, such as the Food and Drug Administration and the Centers for Medicare & Medicaid Services. - In-person meetings or teleconferences with stakeholder groups, totaling 19 meetings. - Presentations and discussions at professional organization meetings, conducted seven times. - Comments submitted through "Contact GTR," receiving 95 responses. The National Institutes of Health collaborates with other agencies in the Department of Health and Human Services to streamline data submission for test providers. The Genetic Testing Registry was designed by the NIH's National Center for Biotechnology Information, incorporating extensive input from stakeholders, including clinicians and test providers. The Paperwork Reduction Act applies to the GTR because it collects standardized information from 10 or more respondents within a 12-month period. This act requires two comment periods for public feedback, with comments submitted on July 27, 2011, and November 23, 2011, being made available for reading. The Paperwork Reduction Act was passed in 1980 to ensure that federal agencies obtain Office of Management and Budget clearance before requesting most types of information from the public. According to the PRA regulation, data collection from at least 10 respondents occurs within a 12-month timeframe. The relevant OMB regulation can be found at 5 CFR 1320, while additional information on the Patient Registries Act is available through the Department of Health and Human Services. GeneTests has been replaced by GeneReviews in NIH support, with its website now redirecting to GTR or NCBI's Bookshelf. GeneReviews are fully incorporated into GTR, as described elsewhere. For inquiries regarding UnitedHealthcare’s Molecular Pathology Reimbursement Policy, contact Provider Services at 877-842-3210 or BeaconLBS at 800-377-8808. Further information can be found on the UHC website: The Rare and Inherited Disorders policy outlines genomic tests for rare and inherited diseases, while the Cancer policy covers eligibility criteria and ordering procedures. The latter is divided into five categories: Adult Solid Tumours, Neurological Tumours, Sarcomas, Haematological Cancer, and Paediatric Tumours.