

# Sharing / Reporting Genomic Results and Data

# Outline

- 1) FDA MedWatch Reports** (*Any Medical Event*)
- 2) PrecisionFDA** (*Raw Data*)
- 3) NIH ClinGen GenomeConnect** (*Genetic Reports*)

# FDA MedWatch Reporting

## *(Step #1: Report a Problem)*

The screenshot shows the FDA MedWatch homepage. At the top is the FDA U.S. Food & Drug Administration header with a search bar and menu. Below is a breadcrumb trail: Home / Safety / MedWatch: The FDA Safety Information and Adverse Event Reporting Program. The main heading is 'MedWatch: The FDA Safety Information and Adverse Event Reporting Program'. Below this are social media share buttons and a 'Subscribe to Email Updates' button. A left sidebar contains links: 'MedWatch: The FDA Safety Information and Adverse Event Reporting Program', 'Subscribe to MedWatch Safety Alerts', 'Medical Product Safety Educational Resources', 'Medical Product Safety Information', and 'Reporting Serious Problems to FDA'. The main content area starts with a paragraph: 'MedWatch, the FDA's medical product safety reporting program for health professionals, patients and consumers.' Below this is a row of three buttons: 'Report a Problem' (highlighted with a red box and an arrow from the sidebar), 'Safety Information', and 'Stay Informed'. Another arrow points from the sidebar to the 'Report a Problem' button. Below the buttons is a paragraph: 'MedWatch receives reports from the public and when appropriate, publishes safety alerts for FDA-regulated products such as:'. This is followed by a bulleted list of product categories. On the right, there is a 'Content current as of: 12/27/2023' section and a 'Regulated Product(s)' list including Biologics, Cosmetics, Dietary Supplements, Drugs, Medical Devices, Radiation-Emitting Products, and Medical Food/Beverage. A 'Topic(s)' section lists 'Recalls'.

U.S. FOOD & DRUG ADMINISTRATION

Search Menu

Home / Safety / MedWatch: The FDA Safety Information and Adverse Event Reporting Program

### MedWatch: The FDA Safety Information and Adverse Event Reporting Program

Subscribe to Email Updates

Share Post LinkedIn Email Print

MedWatch: The FDA Safety Information and Adverse Event Reporting Program

Subscribe to MedWatch Safety Alerts

Medical Product Safety Educational Resources

Medical Product Safety Information

Reporting Serious Problems to FDA

MedWatch, the FDA's medical product safety reporting program for health professionals, patients and consumers.

**Report a Problem** Safety Information Stay Informed

MedWatch receives reports from the public and when appropriate, publishes safety alerts for FDA-regulated products such as:

- **Prescription and over-the-counter medicines**
- **Biologics** such as blood components, blood/plasma derivatives and gene therapies.
- **Medical devices** such as hearing aids breast pumps, and pacemakers.
- **Combination products** such as pre-filled drug syringe, metered-dose inhalers and nasal spray.
- **Special nutritional products** such as dietary supplements, medical foods and infant formulas.
- **Cosmetics** such as moisturizers, makeup, shampoos, hair dyes and tattoos.
- **Food** such as beverages and ingredients added to foods.

Content current as of: 12/27/2023

**Regulated Product(s)**

- Biologics
- Cosmetics
- Dietary Supplements
- Drugs
- Medical Devices
- Radiation-Emitting Products
- Medical Food/Beverage

**Topic(s)**

- Recalls

- While collecting information for recalls and/or reversal of FDA Approval may be a goal, FDA MedWatch can be used for products that are **not** FDA approved.
- **In fact, most of my submissions would fall in that category.**

# FDA MedWatch Reporting

## *(Step #2: Consumer / Patient)*

### MedWatch Online Voluntary Reporting Form



#### Welcome

If this is a medical emergency, please call 911.  
If you have a mental health crisis, please call 988.

Health professionals, consumers and patients can voluntarily report observed or suspected adverse events for human medical products to FDA. Voluntary reporting can help FDA identify unknown risk for approved medical products. Reporting can be done through our online reporting portal or by downloading, completing and then submitting FDA Form 3500 (Health Professional) or 3500B (Consumer/Patient) to MedWatch: The FDA Safety Information and Adverse Event Reporting Program.

While not mandatory, FDA encourages reporters to provide their contact information in case FDA needs to gather more information. Note that reporters can request, within the report, FDA not release their contact information to the manufacturer.



# FDA MedWatch Reporting

## *(Step #3: Complete Report)*



### **Please Note:**

- If I have something important to report, then my expectation is that I can eventually submit a successful report. It is also possible to resume an incomplete submission.
- *However, your report might be accepted, and it may be helpful to save information for a re-submission.*
- You should be notified if your report is accepted. **However, if the report is not accepted, then I don't believe that you will be notified (and you won't receive feedback to improve the submission).**
- **If the report is accepted, then a de-identified version of the report should become available in the MAUDE database (next slide).**

# FDA MAUDE Database (for Accepted Reports)

The screenshot shows the FDA MAUDE Database website. At the top is the U.S. Department of Health & Human Services header. Below it is the FDA U.S. Food & Drug Administration logo. A navigation bar contains links for Home, Food, Drugs, Medical Devices, Radiation-Emitting Products, Vaccines, Blood & Biologics, Animal & Veterinary, Cosmetics, and Tobacco Products. A search bar with a 'SEARCH' button is located in the top right. The main heading is 'MAUDE - Manufacturer and User Facility Device Experience'. Below this is a breadcrumb trail: FDA Home > Medical Devices > Databases. A text box explains that the MAUDE database houses medical device reports submitted to the FDA by mandatory reporters (manufacturers, importers and device user facilities) and voluntary reporters (health care professionals, patients and consumers). A 'Learn More' link and a 'Disclaimer' link are provided. The 'Search Database' section contains several input fields: Product Problem (dropdown), Product Class (dropdown), Event Type (dropdown), Manufacturer (text), Model Number (text), Report Number (text), Brand Name (text), and Product Code (text). There are also date pickers for 'Date Report Received by FDA (mm/dd/yyyy)' with a range from 11/01/2023 to 11/30/2023. At the bottom of the search section are links for 'Go to Simple Search', a 'Records per Report Page' dropdown set to 10, a 'Clear Form' link, and a 'Search' button. On the right side, there is a section titled 'Other Databases' with a list of links: 510(k)s, De Novo, CDRH Export Certificate Validation (CECV), CDRH FOIA Electronic Reading Room, CFR Title 21, CLIA, Device Classification, FDA Guidance Documents, Humanitarian Device Exemption, Medsun Reports, Premarket Approvals (PMAs), Post-Approval Studies, Postmarket Surveillance Studies, Radiation-Emitting Products, Radiation-Emitting Electronic Products Corrective Actions, Recalls, Registration & Listing, Standards, Total Product Life Cycle, and X-Ray Assembler.

U.S. Department of Health & Human Services

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**FDA** U.S. FOOD & DRUG ADMINISTRATION

SEARCH

Home Food Drugs Medical Devices Radiation-Emitting Products Vaccines, Blood & Biologics Animal & Veterinary Cosmetics Tobacco Products

## MAUDE - Manufacturer and User Facility Device Experience

FDA Home Medical Devices Databases

The MAUDE database houses medical device reports submitted to the FDA by mandatory reporters<sup>1</sup> (manufacturers, importers and device user facilities) and voluntary reporters such as health care professionals, patients and consumers.

[Learn More](#) [Disclaimer](#)

### Search Database

Help Download Files

Product Problem

Product Class

Event Type  Manufacturer

Model Number  Report Number

Brand Name  Product Code

Date Report Received by FDA (mm/dd/yyyy)  11/01/2023 to  11/30/2023

[Go to Simple Search](#) 10 Records per Report Page [Clear Form](#)

#### Other Databases

- 510(k)s
- De Novo
- CDRH Export Certificate Validation (CECV)
- CDRH FOIA Electronic Reading Room
- CFR Title 21
- CLIA
- Device Classification
- FDA Guidance Documents
- Humanitarian Device Exemption
- Medsun Reports
- Premarket Approvals (PMAs)
- Post-Approval Studies
- Postmarket Surveillance Studies
- Radiation-Emitting Products
- Radiation-Emitting Electronic Products Corrective Actions
- Recalls
- Registration & Listing
- Standards
- Total Product Life Cycle
- X-Ray Assembler

- I will go through some examples in subsequent slides.



# Mayo/Helix GeneGuide (MW5093889)

The screenshot shows the FDA's MAUDE Adverse Event Report for the MAYO CLINIC GENEGUIDE HELIX; GENETIC VARIANT DETECTION AND HEALTH RISK ASSESSMENT SYSTEM. The report is dated 02/08/2019 and describes a malfunction. The event description includes a patient's account of receiving a false negative result for a cystic fibrosis carrier test. The report is redacted with several (b)(6) and (b)(4) codes. The FDA logo and navigation links are visible at the top.

**U.S. FOOD & DRUG ADMINISTRATION**

**MAUDE Adverse Event Report: MAYO CLINIC GENEGUIDE HELIX; GENETIC VARIANT DETECTION AND HEALTH RISK ASSESSMENT SYSTEM**

[FDA Home](#) [Medical Devices](#) [Databases](#)

**CDRH SuperSearch**

510(k) | DeNovo | Registration & Listing | Adverse Events | Recalls | PMA | HDE | Classification | Standards  
CFR Title 21 | Radiation-Emitting Products | X-Ray Assembler | Medsun Reports | CLIA | TPLC

**MAYO CLINIC GENEGUIDE HELIX; GENETIC VARIANT DETECTION AND HEALTH RISK ASSESSMENT SYSTEM** [Back to Search Results](#)

**Device Problem** Incorrect, Inadequate or Imprecise Result or Readings (1535)  
**Patient Problem** No Known Impact Or Consequence To Patient (2692)  
**Event Date** 02/08/2019  
**Event Type** malfunction  
**Event Description**  
I think the link will be removed from the page; however, in terms of providing info to the fda, there is add'l info available here: (b)(6) I also uploaded data to my personal genome project page on (b)(6) 2019. (b)(6) I think my concern is technically not an error, but i think it is important for consumers: i am a cystic fibrosis carrier (as define by clinvar, the cfr2 database, etc.) but my variant was not above those listed as being checked in my report. However, if you checked my raw data (which i had to pay extra to receive, and i only received a gvcf file, rather than the more typical fastq+bam+vcf combination that i would prefer), then you could see the variant call for my cystic fibrosis variant. So, it is possible to determine this from the data generated, even though my report said i was not a cystic fibrosis carrier (which i think may be confusing for some customers, and that is why i submitted a report). The company specifically advertises cystic fibrosis testing. Fda safety report id# (b)(4)

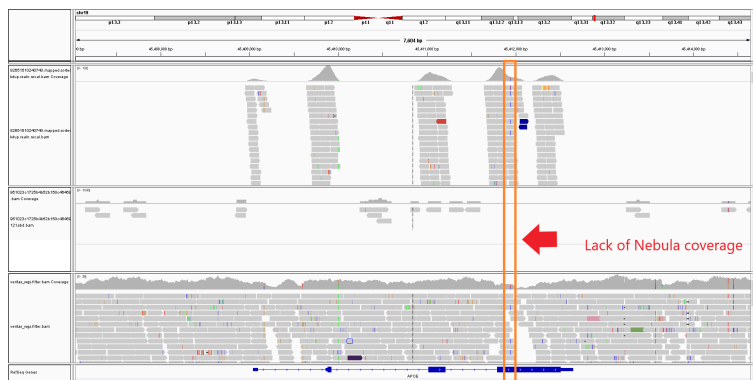
[Search Alerts/Recalls](#)

[New Search](#) | [Submit an Adverse Event Report](#)

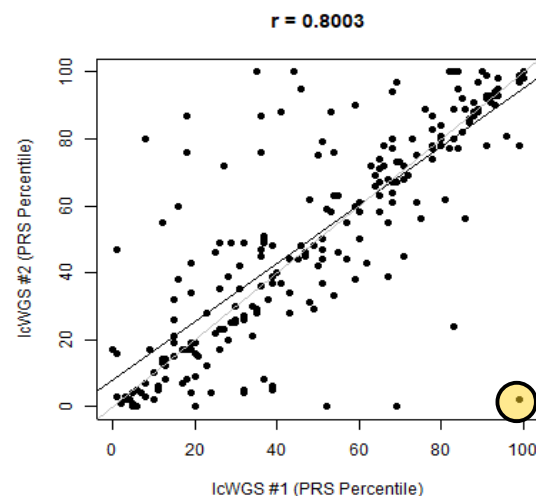
- I am a cystic fibrosis carrier, and this this was designed to test for variants causing this disease. However, I received a false negative result.
  - [Link to Associated Blog Post](#)
- For most examples, I am not sure if there are more recent changes (since original report, in this case in 2019).
  - However, this particular product has been discontinued.
- In the example above, you can see highlighted examples of de-identification. I am not sure about other formatting changes (such as a lack of capitalization within the sentences).

# Nebula low-coverage Whole Genome Sequencing (IcWGS) (MW5093887)

Inaccurate APOE Imputation  
(I am **E3/E4**, not **E3/E3**)



PRS Concerns  
(including replicate discordance)



Replicate  
PRS percentile  
varies from 2%  
and 99%

- [Link to Associated Blog Post](#)
  - The scatterplot of PRS percentiles shown above can be found on [this GitHub subfolder](#).
- In general, I am not sure about more recent changes (after 2019), and **my understanding is that Nebula currently emphasizes on higher coverage Whole Genome Sequencing** (such as 30x or higher, instead of ~0.5x or lower).
- *Other Polygenic Risk Score (PRS) Discussions:*
  - PRS results from other sources (described in [this blog post](#)).
  - I am attempting to apply iCARE breast cancer PRS to samples from myself [here](#).

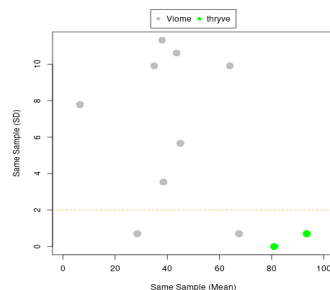


# Viome

## (MW5106218)

- There are multiple metagenomic posts. I have raw data for re-analysis from other companies, but not Viome.
- However, I believe that the outline of most relevant topics is provided in [this blog post](#).
- I also submitted an [FDA Regulatory Misconduct](#) report related to describing being “approved” by the FDA, where you can see a draft for the submitted content [here](#).
- Based in part upon what I show in the next slide (for a different company), **I did not take any action based upon recommendations from these reports.**
  - However, I believe **discordance in results from replicate samples collected at the same** is still noteworthy.
  - The [blog post](#) used for the dietary table on the left was also created for **supplement recommendations** (where I only noted variation in recommendations, without testing for adverse events).
- I have not collected more recent samples, so I am not sure what changes may have occurred after these 2021 reports.

### Higher Variation for Replicate Viome Scores (with more total provided scores)



### Viome Variation and Discordance from Current Helpful Dietary Changes or Preferences

#### Viome "Foods to Avoid":

	Stool 1a	Stool 1b	Stool 2	Stool 3	Stool 4
Vegetables to Avoid	Bell Pepper Broccoli Brussels Sprouts Cabbage Mustard Greens Tomato	Bell Pepper Tomato	Bell Pepper Sauerkraut Tomato	Bell Pepper Tomato	Bell Pepper Cucumber Tomato
Proteins and Fats to Avoid	Almonds Chicken Egg Yolk Pistachios	Almonds Pistachios	Kefir (Cow Milk) Yogurt (Cow Milk, Plain)	Almonds Pistachios	Shrimp (Domestic)
Fruits and Grains to Avoid	None	None	Barley Blueberry	None	Watermelon
Other Food Items to Avoid	None	None	Coffee	Turmeric	None

**I do drink tea instead of coffee**, since coffee can irritate my eyes (and, at least to some extent, my stomach).

# Vitagene (MW5092056)

In other words, Vitagene recommended that I take 7 supplements:

Reported Adverse Event  
encountered after taking *L-Theanine*

*Zinc* supplement may have contributed to  
headache (at dosage available for generic)?  
...I did not continue to take additional tablets.

- *Bromelain Quercetin Complex (500 mg)*: Lifestyle (Joint health and Digestive health)
- *Probiotics (40 billion CFU)*: **Genetics (31%, risk of Overweight, Hormonal support, Eczema, Allergies and Blood pressure health, based upon 103 variants, all reported to have "Fair" research quality)**, Lifestyle (Everyday stress and Digestive health), and Goals (Everyday stress and Overweight)
- *Vitamin D (2000 IU)*: **Genetics (59% Vitamin D Levels, Eczema and Joint health, based upon 36 variants, all reported to have "Fair" research quality)**, Lifestyle (Everyday stress), and Goals (Everyday stress)
- *Theanine (200 mg)*: Lifestyle (Everyday stress), and Goals (Everyday stress)
- *Iron Free Multivitamin (10 Multi)*: Lifestyle (Energy and Nutrient intake levels)
- *Zinc (15 mg)*: **Genetics (50% Overweight, based upon 52 variants, all reported to have "Fair" research quality)** and Goals (Overweight)
- *Chromium (200 mcg)*: **Genetics (83% Hormonal support, Overweight and Blood Sugar Health, based upon 203 variants, all reported to have "Fair" research quality)** and Goals (Overweight)

- [Related Blog Post](#)

- I have not submitted new samples or checked for updates from the company after this post in 2019.
  - However, I believe from a source other than Vitagene, I see that there is a warning that I noted in the blog post: *"If you are currently taking prescription antidepressants such as MAOIs or SSRIs, consult your physician before taking this product."*
  - I was taking (and am taking) an SSRI.

# Probabilistic Language from NIH All of Us Pharmacogenomics Report

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**Important! Genetic information is really just one piece of the puzzle.**

- It won't tell us if a medicine will definitely work.
- It won't tell us if a medicine will definitely cause side effects or won't work at all.
- It won't tell us exactly how much medicine someone should take.
- It only applies to medicines that you eat, drink, or inject. It doesn't apply to medicines that are rubbed on your skin or used in your eyes or ears.

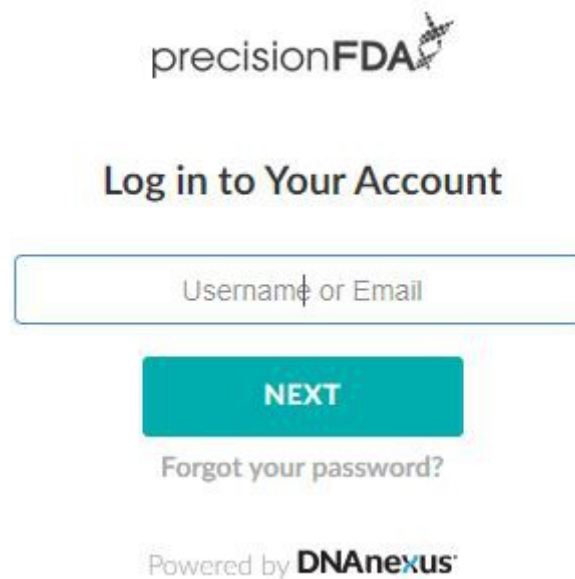
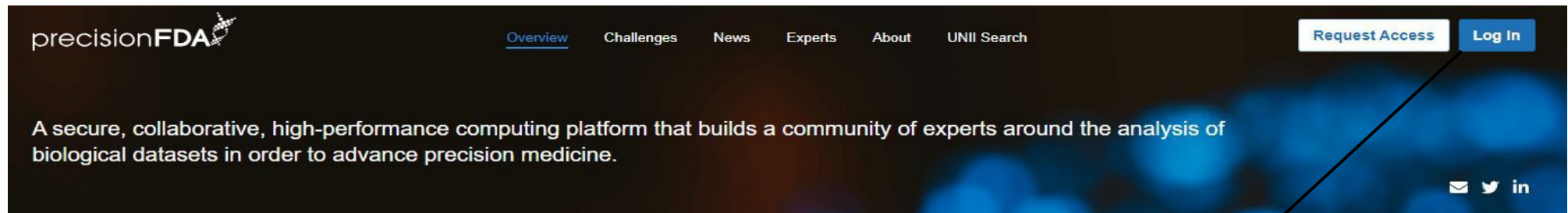
**If your doctor has prescribed medicine for you, keep taking it.** It can be dangerous to stop taking a medicine, or to change the dose or timing of it, without first asking your doctor.

- I will revisit this report in a later slide.
  - However, briefly, you can see my report uploaded [here](#).

# Outline

- 1) FDA MedWatch Reports (*Any Medical Event*)
- 2) **PrecisionFDA** (*Raw Data*)
- 3) NIH ClinGen GenomeConnect (*Genetic Reports*)

# PrecisionFDA Controlled Access Data Sharing

The image shows the login form for PrecisionFDA. At the top is the 'precisionFDA' logo. Below it is the heading 'Log in to Your Account'. There is a text input field with the placeholder text 'Username or Email'. Below the input field is a teal button with the text 'NEXT'. Underneath the button is a link that says 'Forgot your password?'. At the bottom of the form is the text 'Powered by DNAnexus'.

# Experiences with Data Uploads

- There are web-based and command line options for data upload.
- At one point in time, I believe both worked for my account.
- However, most recently I believe the **web-interface** was *more successful* in being able to upload a **VCF** file with **variant calls**.
- For larger files (such as BAM alignment or FASTQ reads from Whole Genome Sequencing), I uploaded the data to Google Cloud and another developer created a URL Import function (**url-fetcher**):  
<https://precision.fda.gov/home/apps/app-F0pyzk000GBvX7qVG137gV5Z-1>
  - So, I think this experience may match others. **Either way, I found this custom app to be very helpful.**
  - That said, please note that there will probably be some **cost to uploading data to a Google Cloud bucket**. If you could directly upload to PrecisionFDA, then that should be free.



# Outline

- 1) FDA MedWatch Reports (*Any Medical Event*)
- 2) PrecisionFDA (*Raw Data*)
- 3) **NIH ClinGen GenomeConnect** (*Genetic Reports*)

*...also includes some public data sharing with personal GitHub repository.*

# ClinGen

The screenshot shows the ClinGen website homepage. At the top is a dark blue navigation bar with links: Data Sharing Resources, GenomeConnect, Events, Contact, and Dashboard. Below this is a lighter blue header with the ClinGen logo and a search bar. The main content area has a large heading 'Explore the clinical relevance of genes & variants' followed by a paragraph about ClinGen's mission. Below this is a search bar with the placeholder text 'Enter a gene symbol or HGNC ID (Examples: ADNP, HGNC:15766)'. A secondary navigation bar lists various resources like 'All Curated Genes', 'Gene-Disease Validity', etc. The main content area features a large teal banner for 'ClinGen is defining the clinical relevance of genes and variants' and several smaller boxes for 'ClinGen Guidance and Recommendations for Monogenic Disease Nomenclature', 'File Downloads and APIs', and 'Volunteer to Curate'.

My understanding is that **“GenomeConnect”** provides options for **individual** genetic/genomic data sharing.

As of 1/ 1/2024, **“Data Sharing Resources”** provides information for **clinicians** and **laboratories** (for genetic/genomic data sharing).

# BRCA1 ClinGen Screenshot

## Gene-Disease Validity

Gene	Disease	MOI	Expert Panel	Classification	Report & Date
BRCA1	Fanconi anemia, complementation group S MONDO:0054748	AR ⓘ	Hereditary Cancer GCEP <a href="#">↗</a>	Definitive	<a href="#">05/14/2020</a>
BRCA1	breast-ovarian cancer, familial, susceptibility to, 1 MONDO:0011450	AD ⓘ	Breast/Ovarian Cancer GCEP <a href="#">↗</a>	Definitive	<a href="#">09/13/2017</a>

## Dosage Sensitivity

Gene	Disease	Working Group	HI Score & TS Score	Report & Date
BRCA1	breast-ovarian cancer, familial, susceptibility to, 1 MONDO:0011450	Dosage Sensitivity WG <a href="#">↗</a>	3 (Sufficient Evidence for Haploinsufficiency)	<a href="#">09/23/2021</a>
BRCA1		Dosage Sensitivity WG <a href="#">↗</a>	0 (No Evidence for Triplosensitivity)	<a href="#">09/23/2021</a>

## Clinical Actionability

Gene	Disease	Report	Working Group	Assertions	Report & Date
BRCA1	breast-ovarian cancer, familial, susceptibility to, 1 MONDO:0011450	Hereditary Breast and Ovarian Cancer	Pediatric Actionability WG <a href="#">↗</a>	Pediatric N/A - Insufficient evidence: early rule-out ⓘ	<a href="#">09/15/2021</a>
		Hereditary Breast and Ovarian Cancer	Adult Actionability WG <a href="#">↗</a>	Adult Definitive Actionability ⓘ	<a href="#">01/28/2014</a>
BRCA1	hereditary breast ovarian cancer syndrome MONDO:0003582	Hereditary Breast and Ovarian Cancer	Pediatric Actionability WG <a href="#">↗</a>	Pediatric N/A - Insufficient evidence: early rule-out ⓘ	<a href="#">09/15/2021</a>
		Hereditary Breast and Ovarian Cancer	Adult Actionability WG <a href="#">↗</a>	Adult Definitive Actionability ⓘ	<a href="#">01/28/2014</a>

## Variant Pathogenicity

Gene	Disease	Expert Panel	Classification	Date
BRCA1	breast-ovarian cancer, familial, susceptibility to, 1 MONDO:0011450	ENIGMA BRCA1 and BRCA2 VCEP <a href="#">↗</a>	<div>Pathogenic 10</div> <div>Likely Pathogenic 1</div> <div>Uncertain Significance 1</div> <div>Benign 8</div>	<a href="#">Evidence</a> <a href="#">Evidence</a> <a href="#">Evidence</a> <a href="#">Evidence</a>

- You can see annotations from multiple sources are provided in ClinGen.
  - I believe that ClinGen “*Variant Pathogenic*” expert panel classifications are also submitted to ClinVar (with 3-star evidence).
- Please be aware that the ClinGen “*Variant Pathogenicity*” annotations from ENIGMA are **on-going**. For example, there are more than 20 variants for BRCA1.
  - In general, I believe BRCA Exchange is also a useful source of information.
  - I believe ENGIMA is also used as the source for clinical significance annotations for BRCAExchange.
  - However, in ClinGen, you can see additional information available for c.135-1G>T (versus c.5212G>A, for example).

# CFTR ClinGen Screenshot

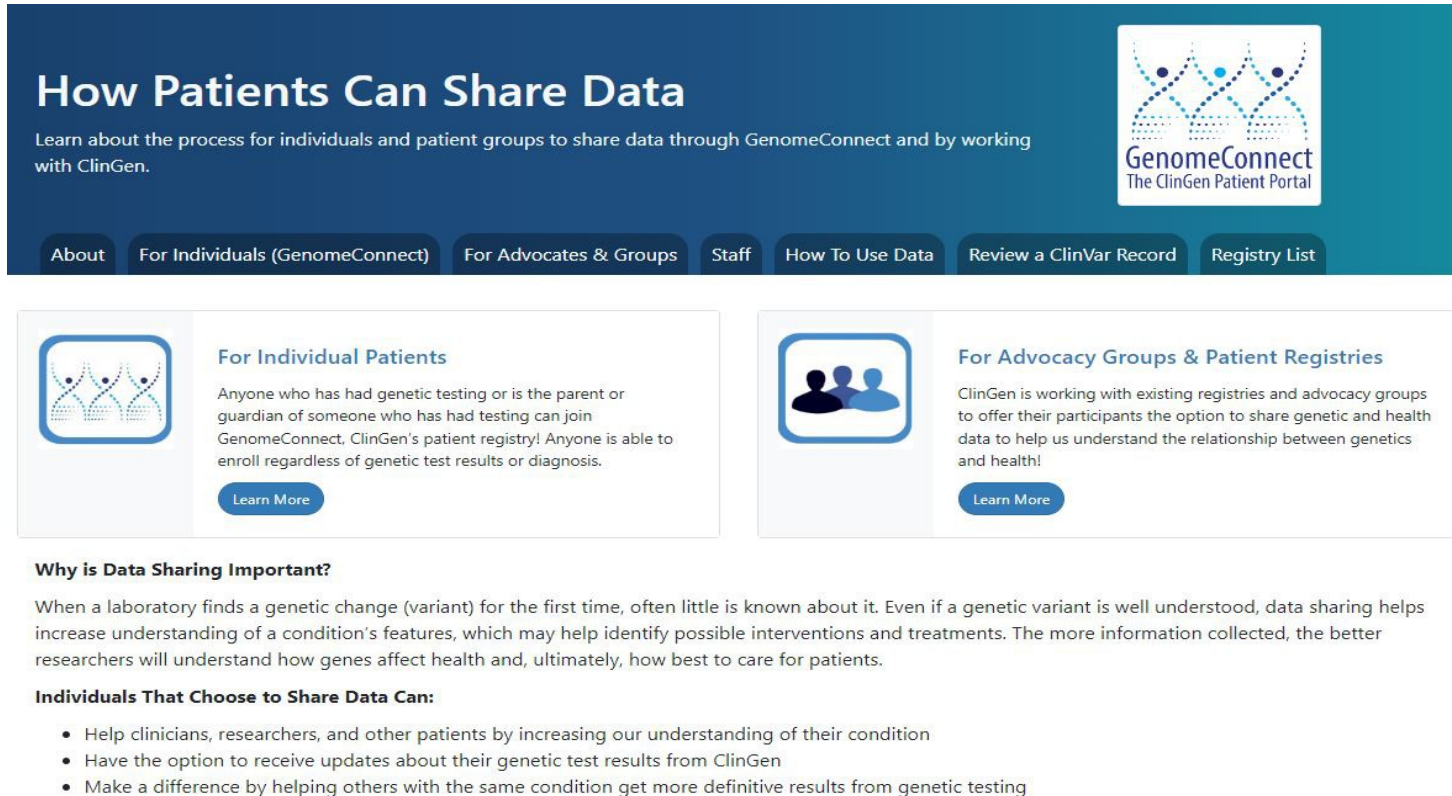
The screenshot displays the CFTR ClinGen page with the following sections:

- Top Navigation:** CFTR logo, "View Gene Facts" button, and statistics for Gene-Disease Validity (0), Dosage Sensitivity (1), Clinical Actionability (0), Variant Pathogenicity (0), and CPIC / PharmGKB High Level Records (2 / 3). A "Follow Gene" star icon is also present.
- Tabbed Interface:** Curation Summaries, Status and Future Work (selected), External Genomic Resources, and ClinVar Variants.
- Dosage Sensitivity Section:**
  - Group By: Activity (selected), Gene-Disease Pair
  - Table with columns: Gene, Disease, Working Group, HI Score & TS Score, Report & Date.
  - Row 1: CFTR, cystic fibrosis (MONDO:0009061), Dosage Sensitivity WG, 30 (Gene Associated with Autosomal Recessive Phenotype), 08/22/2016.
- Pharmacogenomics - CPIC Section:**
  - Table with columns: Gene, Drug, CPIC Level, Date Accessed, CPIC Clinical Guidelines.
  - Row 1: CFTR, ivacaftor, Level A, 01/01/2024, Guideline.
  - Row 2: CFTR, ataluren, Level C, 01/01/2024, Provisional.
- Pharmacogenomics - PharmGKB Section:**
  - Table with columns: Gene, Drug, Highest Level of Evidence, Last Curated, Information.
  - Row 1: CFTR, ivacaftor, Level 1A, 03/24/2021, View.
  - Row 2: CFTR, ivacaftor / lumacaftor, Level 1A, 03/24/2021, View.
  - Row 3: CFTR, ivacaftor / tezacaftor, Level 1A, 03/24/2021, View.

- Again, you can see annotations from multiple sources is provided in ClinGen.
  - If you click the ClinVar link, you can see individual variants with a condition listed as “cystic fibrosis.”
- However, **I don't see direct evidence for CFTR causing cystic fibrosis** (which I would guess should be from within some similar categories as BRCA1 on last slide, even if this is *predictive* for autosomal recessive inheritance instead of a *risk* that can be described autosomal dominant).
  - Based upon assistance from ClinGen / GenomeConnect support, I believe this is because **a ClinGen expert panel evaluation has not been submitted**.



# GenomeConnect for Individual Data Sharing (Screenshot from ClinGen)



The screenshot shows the 'How Patients Can Share Data' section of the GenomeConnect website. The header includes the title 'How Patients Can Share Data' and a sub-header 'Learn about the process for individuals and patient groups to share data through GenomeConnect and by working with ClinGen.' The header also features the GenomeConnect logo and a navigation bar with links: 'About', 'For Individuals (GenomeConnect)', 'For Advocates & Groups', 'Staff', 'How To Use Data', 'Review a ClinVar Record', and 'Registry List'.

**How Patients Can Share Data**  
Learn about the process for individuals and patient groups to share data through GenomeConnect and by working with ClinGen.

**For Individual Patients**  
Anyone who has had genetic testing or is the parent or guardian of someone who has had testing can join GenomeConnect, ClinGen's patient registry! Anyone is able to enroll regardless of genetic test results or diagnosis.  
[Learn More](#)

**For Advocacy Groups & Patient Registries**  
ClinGen is working with existing registries and advocacy groups to offer their participants the option to share genetic and health data to help us understand the relationship between genetics and health!  
[Learn More](#)

**Why is Data Sharing Important?**  
When a laboratory finds a genetic change (variant) for the first time, often little is known about it. Even if a genetic variant is well understood, data sharing helps increase understanding of a condition's features, which may help identify possible interventions and treatments. The more information collected, the better researchers will understand how genes affect health and, ultimately, how best to care for patients.

**Individuals That Choose to Share Data Can:**

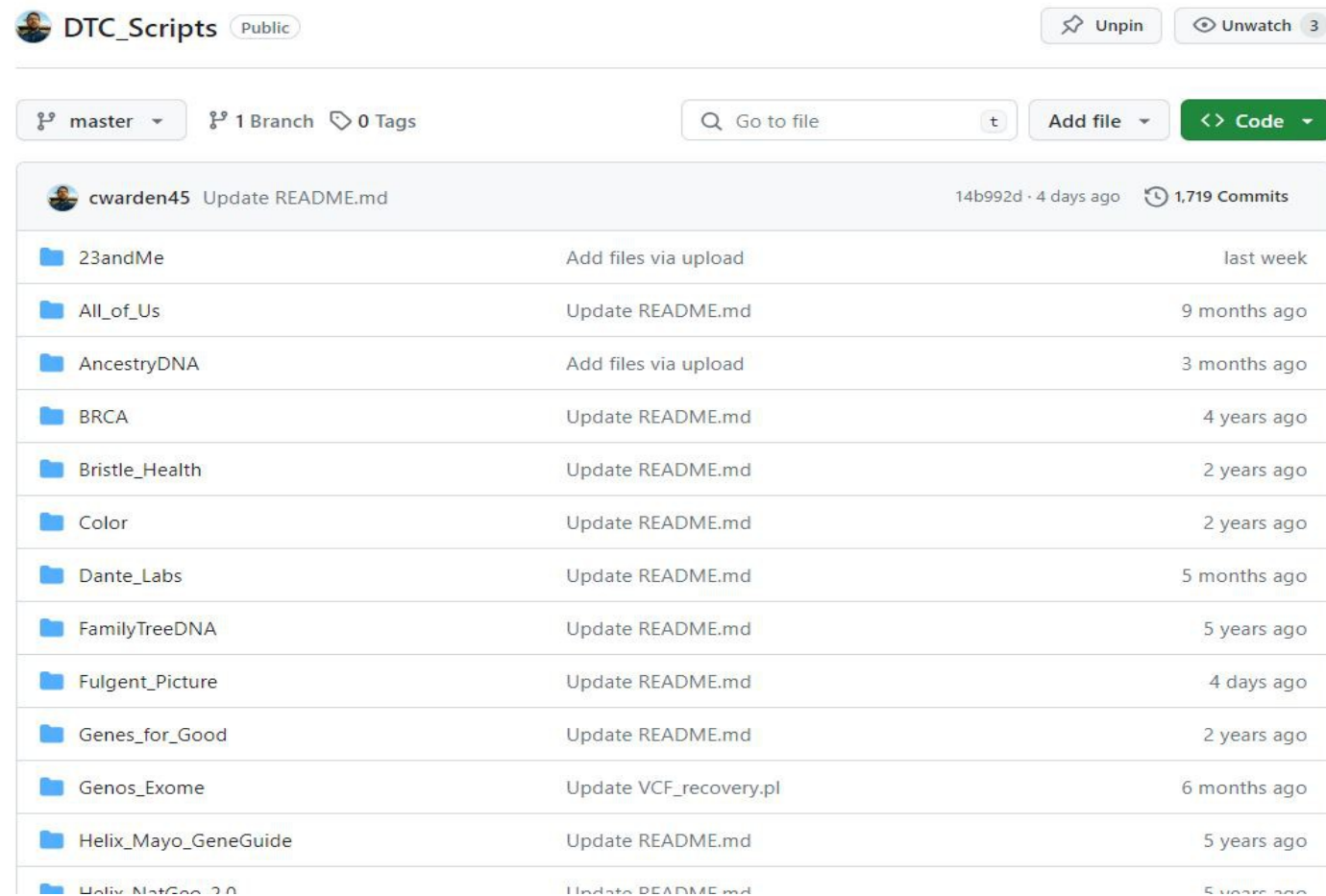
- Help clinicians, researchers, and other patients by increasing our understanding of their condition
- Have the option to receive updates about their genetic test results from ClinGen
- Make a difference by helping others with the same condition get more definitive results from genetic testing

- More recently, I have encountered difficulties uploading additional data to my Personal Genome Project page (for [hu832966](#)).
- I can upload most of the similar content to a [GitHub repository](#) (and/or Google Cloud Buckets).
  - This is described in more detail on the next slide.
- However, that is less systematic data sharing than the Personal Genome Project (or PrecisionFDA, etc).
- **So, I was hoping that this could be an alternative from the NIH.**

# DTC\_Scripts GitHub Page

## (Re-Analysis Code, Reports, and Small Datasets)

- Veritas Whole Genome Sequencing Code and Results can be found at the top level.
- However, “subfolders” (shown right) can be used to access content related to other companies or organizations.
- **I have also uploaded these slides** (available here)



DTC\_Scripts Public

Unpin Unwatch 3

master 1 Branch 0 Tags

Go to file t Add file >> Code

cwarden45 Update README.md 14b992d · 4 days ago 1,719 Commits

23andMe	Add files via upload	last week
All_of_Us	Update README.md	9 months ago
AncestryDNA	Add files via upload	3 months ago
BRCA	Update README.md	4 years ago
Bristle_Health	Update README.md	2 years ago
Color	Update README.md	2 years ago
Dante_Labs	Update README.md	5 months ago
FamilyTreeDNA	Update README.md	5 years ago
Fulgent_Picture	Update README.md	4 days ago
Genes_for_Good	Update README.md	2 years ago
Genos_Exome	Update VCF_recovery.pl	6 months ago
Helix_Mayo_GeneGuide	Update README.md	5 years ago
Helix_NatGeo_2.0	Update README.md	5 years ago



# NIH *All of Us* Pharmacogenomics Example

## Medicine (Pharmacogenomics)

I expanded the details when [creating the uploaded PDF](#), which includes more details of what was tested and limitations in the results.

There is a warning that **These medicines MAY BE impacted by your genetics**. Likewise, there are the following warnings (**bold font** added by me):

- "It **won't** tell us if a medicine will definitely work."
- "It **won't** tell us if a medicine will definitely cause side effects or won't work at all."
- "It **won't** tell us exactly how much medicine someone should take."

So, I think is good in terms of communicating some limitations in predictive power. However, it might have been nice if I could learn more about the variation in risk estimates (for absolute and relative risk).

In terms of a short summary:

Gene	Alleles / Type	Status	Possible Affected Medications
CYP2C19	*1/*1	Normal Metabolizer	
DPYD	*1/*1	Normal Metabolizer	
G6PD	B	Normal	
NUDT15	*1/*1	Normal Metabolizer	
SLCO1B1	*1/*15	Decreased Function ( <b>may</b> increase your risk of developing muscle pain.)	simvastatin (Zocor®)
TPMT	*1/*1	Normal Metabolizer	
UGT1A1	*1/*1	Normal Metabolizer	

... ..

- In general, you can see summaries and re-analysis of results [on the GitHub page](#).
- The above text is an example of me **parsing and reformatting** some content from my *All of Us* report.

# GenomeConnect Homepage



# Additional GenomeConnect Notes

- I used "*Other disease or health condition*" for myself, in order to complete registration.
  - In general, you can **find other participants** based upon diagnosis, gene, and/or geographic location.
- I do not have experience with my own variants, but my understanding is that GenomeConnect is capable of providing **free variant updates**.
  - However, please be aware that the timeline for updates is not certain.
  - Also, as explained by GenomeConnect staff, "*most participants will not receive updates and GenomeConnect does not identify all genetic updates.*"

# Additional GenomeConnect Notes

## (...continued)

- Based upon Savatt et al. 2018, my understanding is that GenomeConnect helps meet the goal for ClinGen to assist with *"[supporting] data sharing."*
  - Savatt et al. 2018 also describes potential benefits to adding and/or refining variant information for classifications in ClinVar.
- I believe that you can use *clingen@clinicalgenome.org* or *info@genomeconnect.org* to ask for assistance, which I found to be very helpful.

Thank You Very Much  
for Your Interest!