APEX Order Guide for Inpatient and Ambulatory Staff

GENOME SEQUENCING

BRIEF TEST DESCRIPTION:

Genome sequencing interrogates nearly the entire genomic sequence, including both coding and non-coding regions. Genome sequencing allows for analysis beyond the exome within known areas of interest, offering improved detection of pathogenic variants across a broader genomic landscape. It is particularly useful for individuals with complex or atypical presentations, suspected monogenic disorders, or negative/inconclusive results from prior genetic testing. This comprehensive method enables detection of single nucleotide variants, small insertions and deletions (indels), exon-level and broader copy number variants, mitochondrial variants, and some repeat disorders. (PMIDs: 21946919, 26733501, 30072248).

THINGS TO CONSIDER:

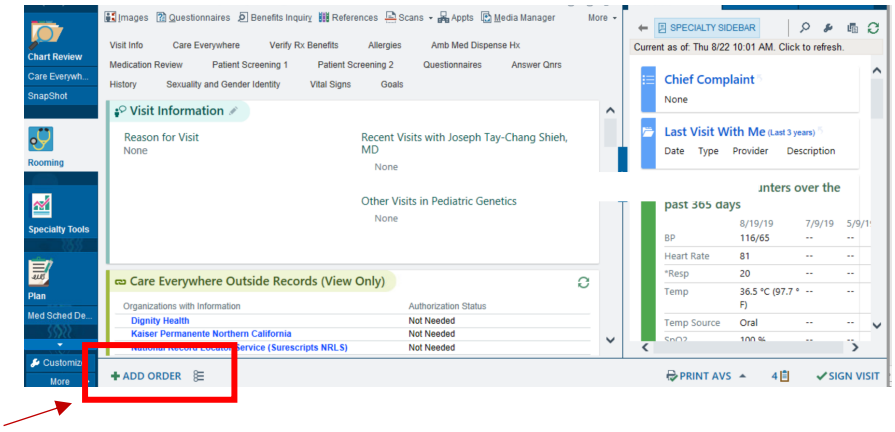
- Click the "STAT" button for expedited turnaround AKA Rapid Genome Sequencing. Expected turnaround time is around 2 weeks.
- The test is phenotypically driven. Include all relevant diagnoses and clinical information under "Indication for test/Diagnosis."
- For saliva samples, select "Secretion" under "Specimen Type" and follow the prompts. The laboratory will ship saliva kits to the address on file unless instructed otherwise. Kits can also be requested via email at GML@ucsf.edu
- Use Smartphrase: UCSFexomegenomeconsent in your provider note to document informed consent. Refer to the link below for Informed Consent guide and short video to assist your consent process.
- For specific panel request performed on an exome/genome backbone, include the test name, laboratory, and any relevant links or genes. Recommended to select “No” for Secondary Findings, in these cases.
- For family member blood draws, order Family Member Blood Draw for GML (LAB4253).
- Results will be automatically released to the patient’s MyChart Test Results unless a 5-day delay was specified.

LINKS TO ASSIST CONSENTING:

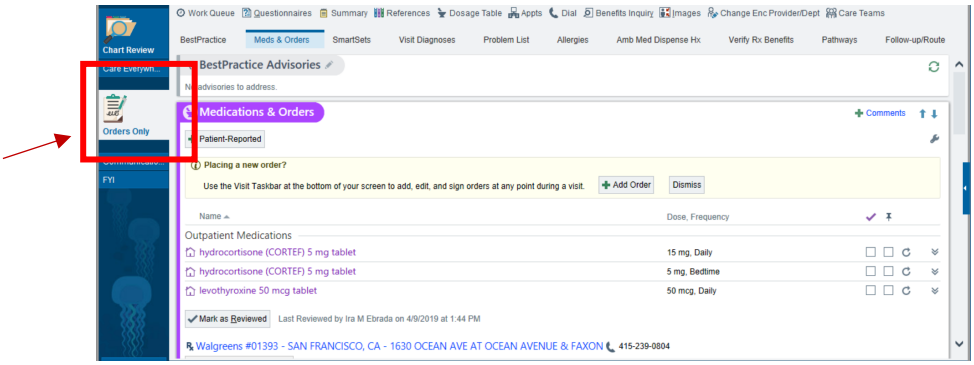
- Link to Frequently Asked Questions for Exome/Genome Sequencing
- Link to One Pager Consent guide for providers
- Link to Consent Video for parents/patients
- Link to Secondary Findings list from American College of Medical Genetics
- Link to Genetic Information Nondiscrimination Act (GINA)

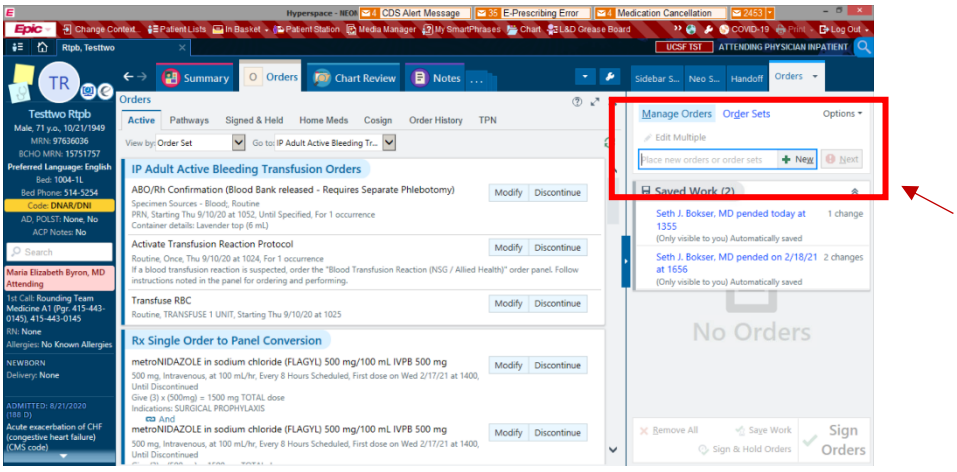
STEP 1:

- OUTPATIENT/AMBULATORY SETTING: In the Encounter – Click on **"+Add Order".**



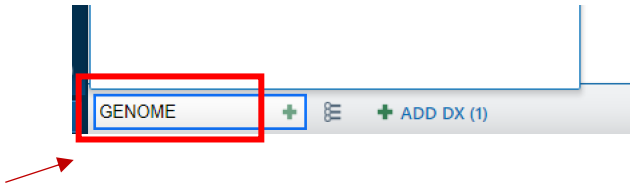
- You can also do an "**Orders Only**" encounter and add the order.





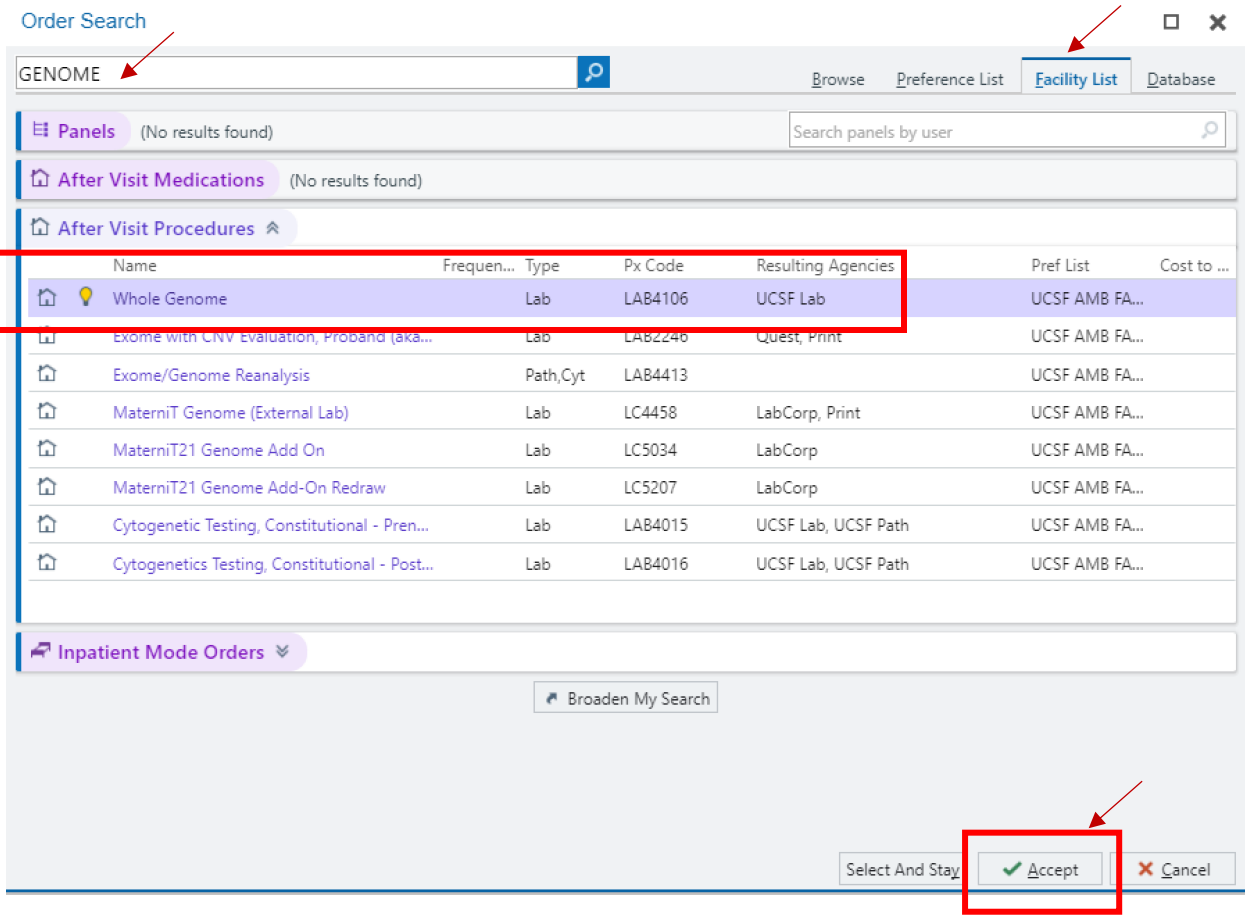
STEP 2:

- Type the word "**GENOME**" in the bottom left "**+ADD Order" or in the "Manage orders"** Box.



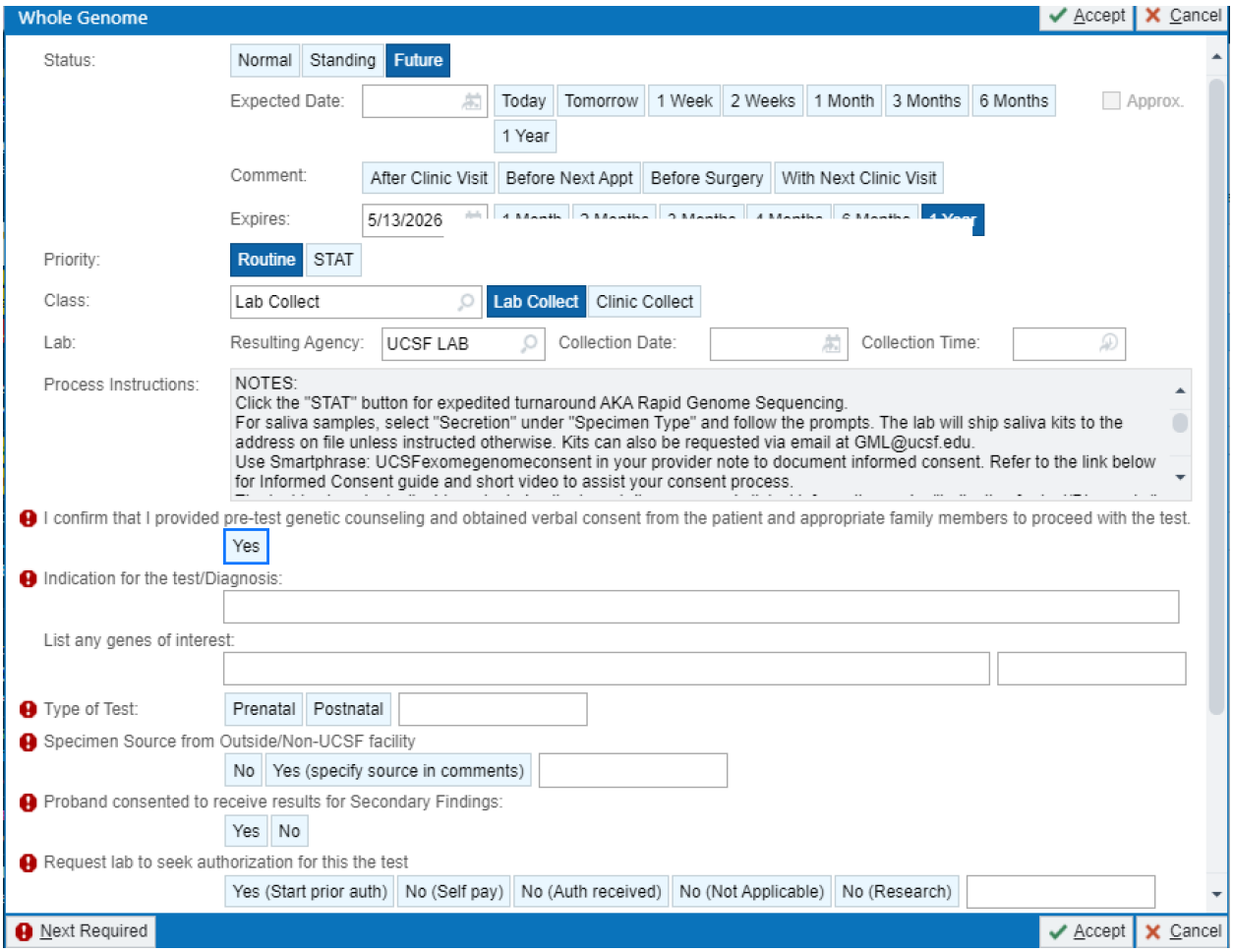
STEP 3:

- Select "**Whole Genome" or** type the lab code **"Lab 4106"**.
- If you cannot see it, try going under **"Facility List"** and searching for the test with the terms above. You can choose to broaden the search as needed. Please note that if you are using a "preference list," this test may not appear, as availability can vary depending on the clinical context or ordering setting.
- Then hit **"Accept".**

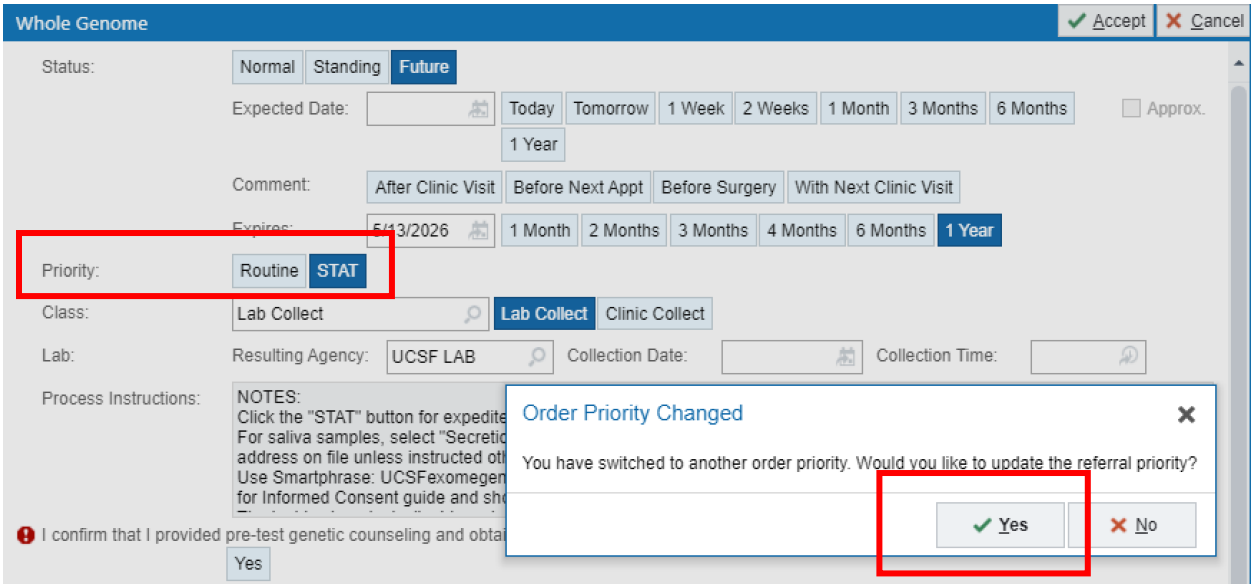


STEP 4:

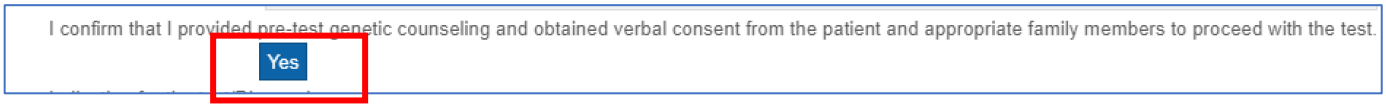
- A pop-up window will appear with instructions and suggestions below.
- All required fields must be completed.



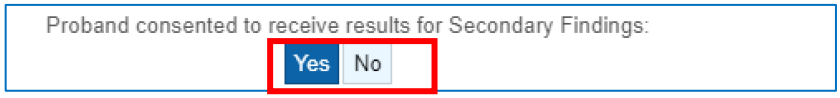
- ***STAT Orders:*** Select **"STAT"** and **"Yes"** if you want **Rapid Genome Sequencing** with turnaround time of 7-14 days and "Routine" is around 30 days.



- ***Test Consent:*** Physician statement of consent required to order this test. Consider using dot phrase **"UCSFexomegenomeconsent"** in your note to document consent in patient chart. No paper or pdf consent is required for UCSF or affiliated patients.



- ***Secondary Findings:*** Select "Yes" if the proband has consented to receive secondary findings; this is the default selection. The test screens for the current ACMG-recommended secondary findings gene list. Additional family members will only receive results in this category if the proband is found to have a reportable variant. 3-4% cases will have a positive secondary finding.

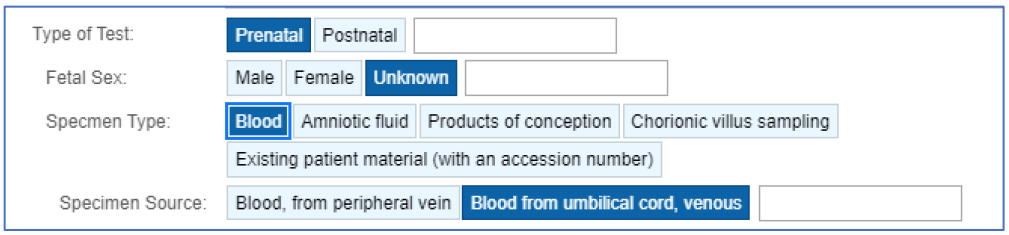


- ***Test Consent Support:*** Use the provided links and consent support materials to ensure patient and/or family is appropriately consented. For assistance, contact the laboratory at GML@ucsf.edu or consult with Peds or Adult Genetics.

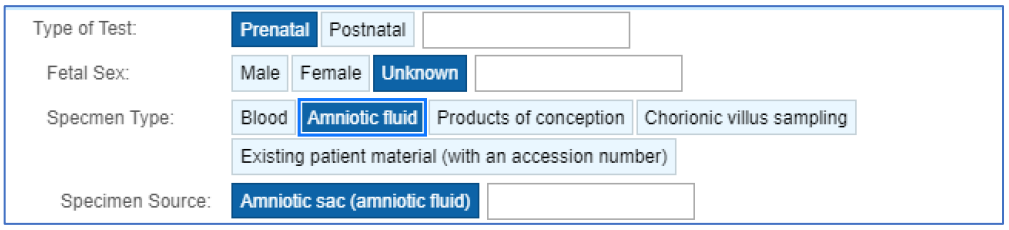
- ***Indication for test:*** Carefully enter clinical terms, as this is a phenotype-driven test. Include all relevant diagnoses and clinical details under "Indication for Test/Diagnosis." Specify any genes or panels of interest, and include the test name or a link, if available.

- ***Prenatal Orders – Sample types:*** GML can perform genome sequencing on a variety of prenatal specimen types, including umbilical cord blood, amniotic fluid, products of conception (POCs), chorionic villus samples (CVS), or DNA extracted from other sources. Please follow the specific prompts for each sample type during the order process.

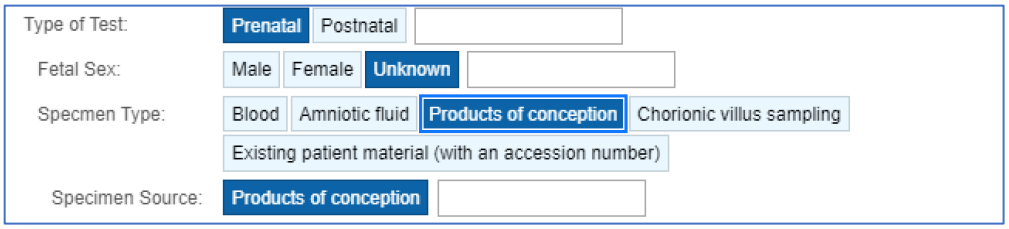
Blood:



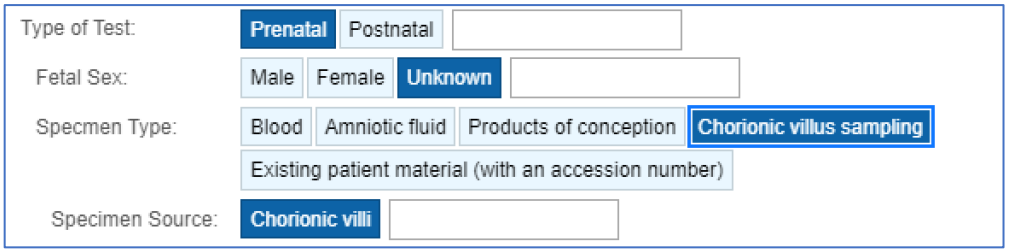
Amniotic fluid:



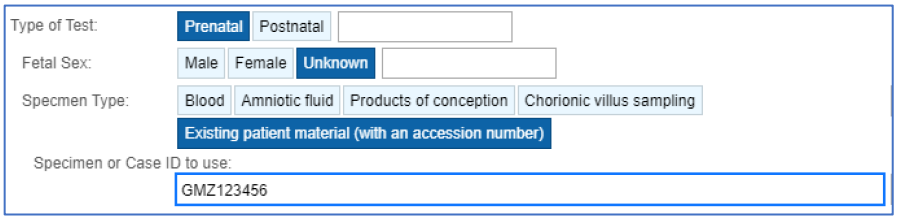
Product of Conception (POC):



Chorionic Villi Sampling (CVS):



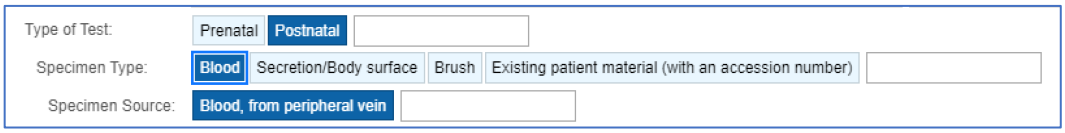
Existing DNA:



- ***Postnatal Orders – Sample types*:** GML can perform genome sequencing on a variety of postnatal specimen types, including blood, saliva, buccal, or DNA extracted from other sources. Please follow the specific prompts for each sample type during the order process*. GML will send the kits out to the patient address on file. If a different address is requested, please include details in the "Comments" section.*

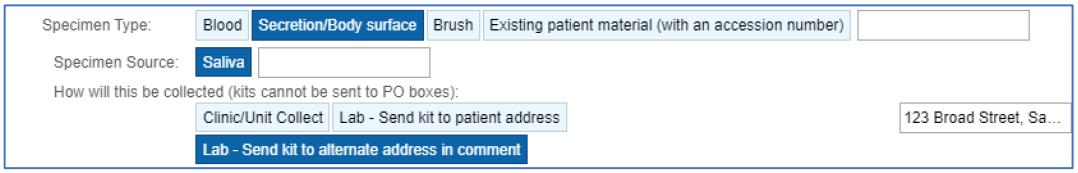
Blood:

- Specimen type: "**Blood**". Then select **"Blood, from peripheral vein".**



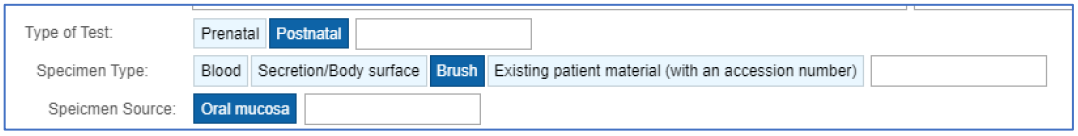
Saliva:

- Specimen type: "**Secretion/Body surface**". Then select "**Saliva**".



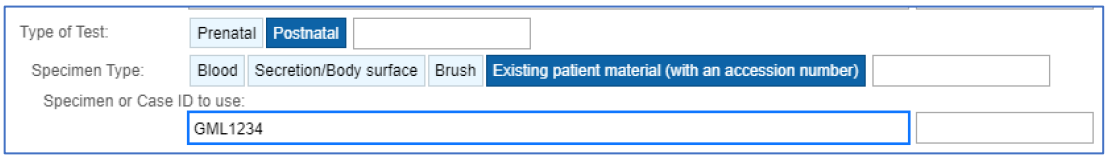
Buccal:

- Specimen type: "**Brush**". Then select "**Oral mucosa**".

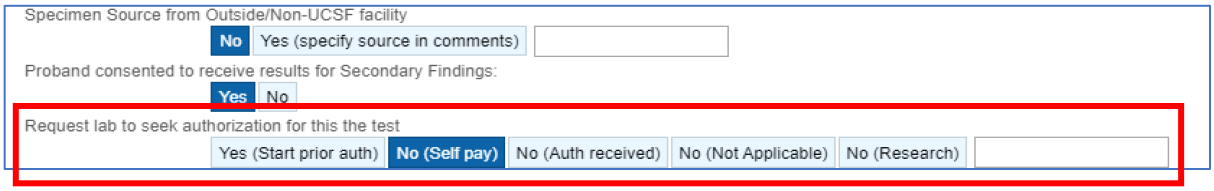


Existing DNA:

- Add the details of the known sample in the lab. Include accession numbers, GML#s from past test/sample or other test details.



- ***Authorization:*** You can select any option, as applicable:



  - **Yes (Start prior auth) -** Select this option when you want UCSF Genomics lab (GML) to request prior authorization for you. GML will start insurance authorization on behalf of the ordering provider and will share updates via In-basket in APEX.

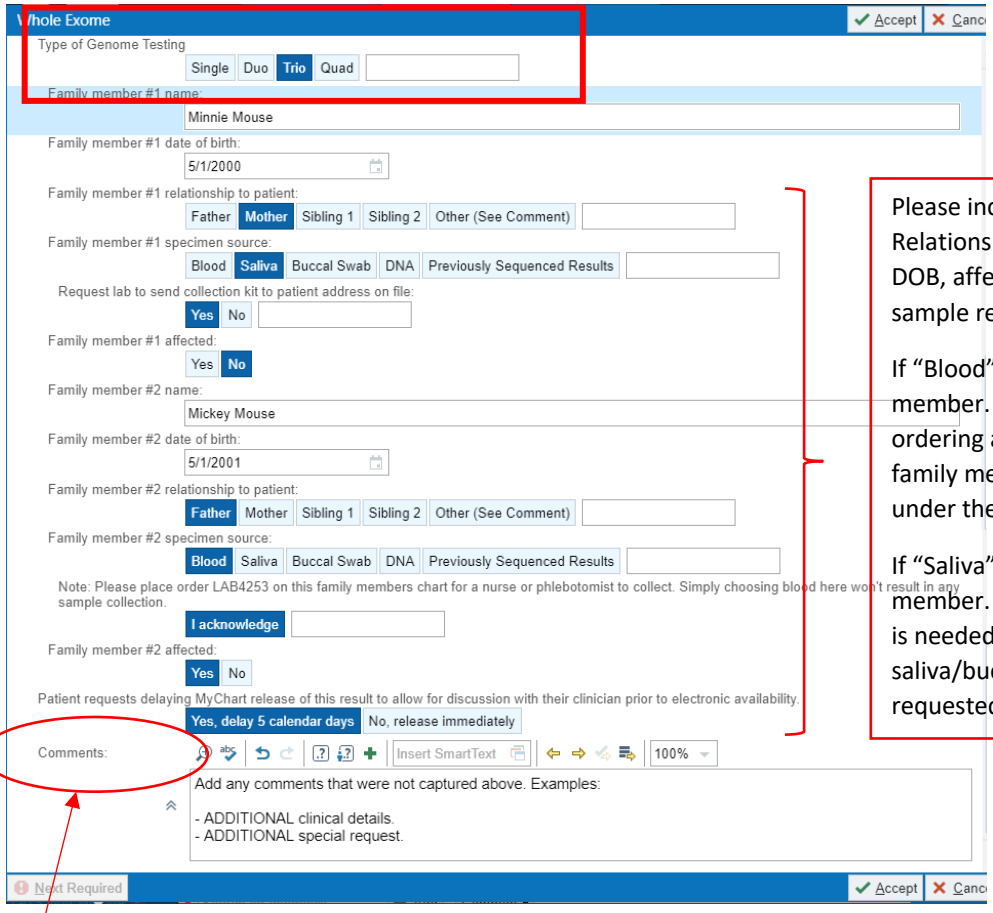
  - **No (Self-pay) –** Lab offers self-pay prices. Contact GML@ucsf.edu for current price details.

  - **No (Auth received) –** Select this option if authorization has already been obtained. Include prior authorization details in the Comments section.

  - **No (auth not required) –** Select this option if authorization for the test is not required or not a covered benefit.

  - **No (research) –** Select this option if the test is paid through research/COA/ZZ account. Include details in the "Comments" section.

- *Type of Test* - Order the test as **Singleton/Duo/Trio/Quad.** Family members are added to aid in variant interpretation, assess inheritance, and increase diagnostic yield.
    - **Singleton –** sequencing "proband only". Your index patient.
    - **Duo –** sequencing "proband" and one family member (usually one available parent or a first degree relative).
    - **Trio** – sequencing "proband" and two family members (usually both biological parent).
    - **Quad –** sequencing "proband" and three family members (usually both parents and one affected sibling or family member).



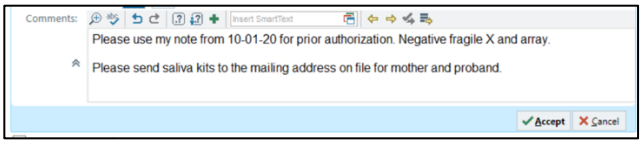
Please include Family Member's Relationship to Proband, Full Name, DOB, affected status, and type of sample requested.

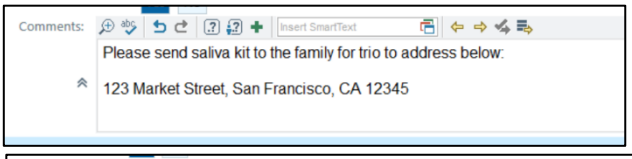
If "Blood" is preferred for a family member. Follow directions for ordering a blood draw for each family member (Separate order under the parent's MRN, LAB4253).

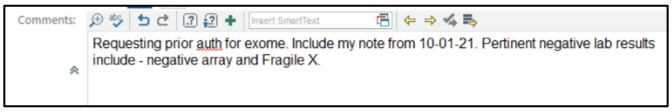
If "Saliva" is preferred for a family member. No MRN or separate order is needed. The lab can send saliva/buccal kits to address on file if requested at no additional cost.

Comments: Include any details that will be relevant for testing/coordination/billing. See examples below:

- Specific requests
- Confirm the mailing address for saliva samples (if different from APEX)
- Include relevant prior authorization details

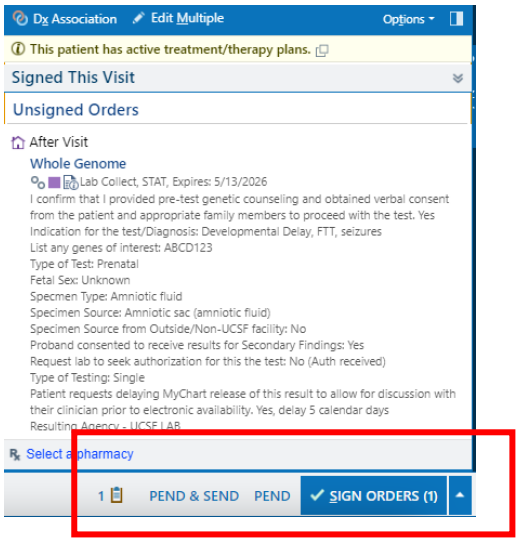




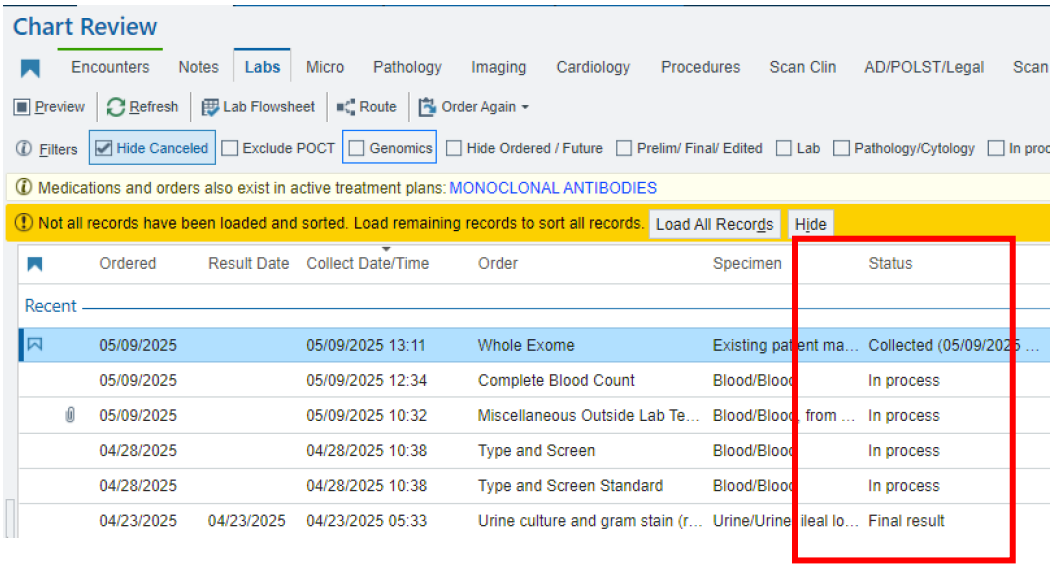


STEP 5:

- Sign or Pend the order after associating diagnoses.



Once the orders are signed, the order will appear under “Labs” in “Chart Review”.



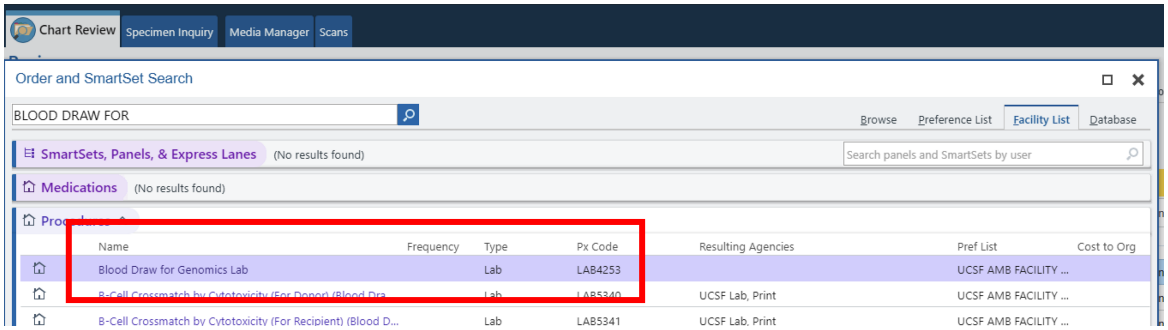
The status of the test will be updated automatically in APEX.

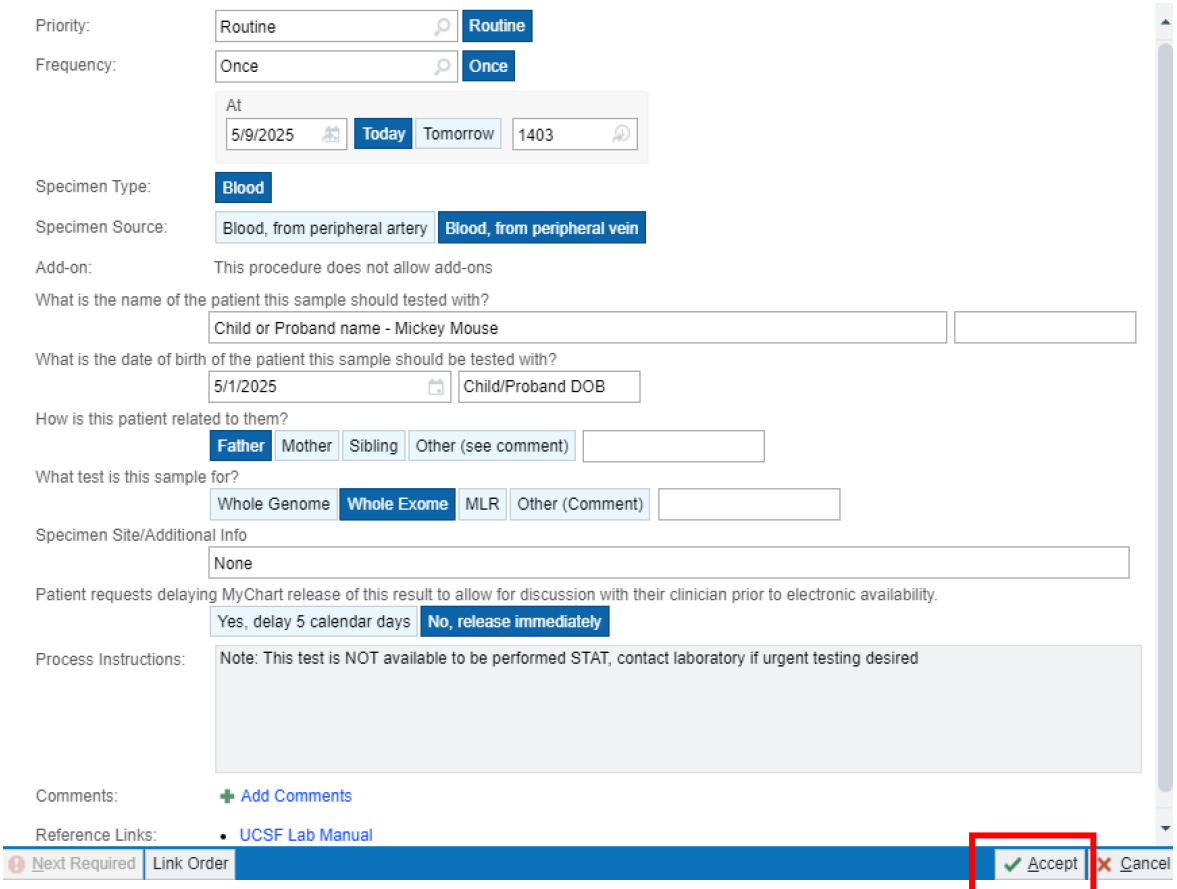
1. Needs to be collected
   - Orders placed. Sample Needs to be collected.
2. Collected (DATE, TIME)
   - Samples collected. In transit to the lab performing the test.
3. In process
   - Samples and test order received by the lab. Test is in progress.
4. Final Result
   - Test completed. Results available.
5. Edited Result – FINAL
   - Test needed amendment/edits. New report available.

STEP 8: Separate order for **blood draw for a Family member.**

Use this order if:

- You are ordering a duo/trio/quad genome test requesting a blood draw on a family member.
- Each Family Member will require a UCSF medical record number.
- Type "**Blood draw**" in the search box. Select **"Blood Draw for Genomics Lab"**
- Include the proband/index patient/primary patient's Full name and DOB, so that we can link the family member with the proband or index patient.
- Complete the order details and sign the order.





Prior Authorization:

- All genome orders placed in APEX in the outpatient setting will be processed by the GML lab for prior authorization.
- The ordering provider will be notified of the **authorization status via "In-Basket"** message via APEX.
- Depending on the insurance plan details, deductible and other factors, a peer-to-peer or letter of medical necessity might be required for appeal. Physician collaboration is requested to complete this step after initial determination.
- Self pay options also available on request. Contact lab for current pricing details.

For any questions or requests, please contact the Genomics Medicine Laboratory (GML) at:

UCSF Genomic Medicine Laboratory (GML)
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Phone: (415) 502-3560
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Email: GML@ucsf.edu
https://genomics.ucsf.edu/ucsf-genomic-medicine-laboratory