

Before the Test: Informed Consent and Collecting Samples

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Learning Objectives:

1. Identify ACMG recommendations for obtaining informed consent
2. Explain the benefits, limitations and utility of genomic testing in a family-appropriate manner
3. Identify parent and provider resources for informed consent
4. Describe challenges and solutions in collecting parent samples
5. Discuss challenges and solutions to implementing genomic medicine in a NICU setting

2012 ACMG Policy Statement

- GS/ES is not recommended before the legal age of majority except for:
 - Phenotype-driven clinical diagnostic uses;
 - Circumstances in which early monitoring or interventions are available and effective; or
 - Institutional review board–approved research.
- A clear distinction should be made between clinical and research-based (genetic) testing
 - Patients should be informed as to whether individually identifiable results may be provided to databases, and they should be permitted to opt out of such disclosure.
- Patients (and families) should be counseled regarding the potential benefits and risks of GS/ES, the limitations of such testing, potential implications for family members, and alternatives to such testing.
- Pretest counseling should include a discussion of the expected outcomes of testing, the likelihood and type of incidental results that may be generated, and the types of results that will or will not be returned
 - Secondary findings? Pharmacogenomic results?
- Patients should be informed of policies regarding re-contact of referring physicians as new knowledge is gained about the significance of particular results.

What is the role of Genetics?

Genetic testing is a blood test that looks at your child's **DNA**



Goals of genetic testing:

- Identify spelling changes in DNA that could explain your child's medical concerns
- **WHY** does your child have this medical history
- May provide information to guide your child's medical management

No genetic test can identify every DNA change

- Exome/genome have a 30-40% diagnostic rate

Genetic Variants

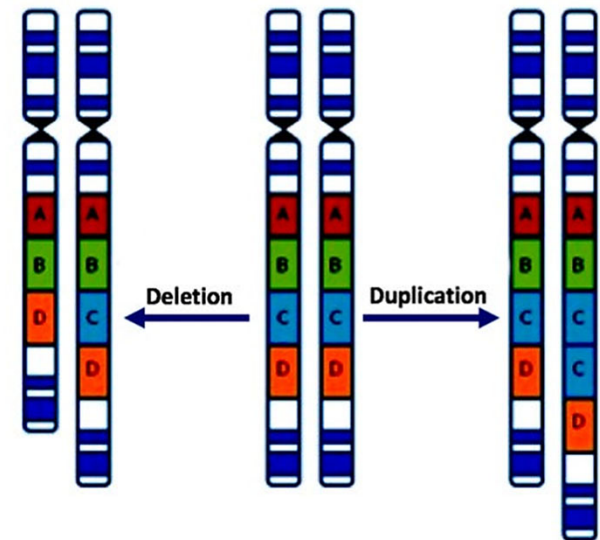
We all have spelling changes within our DNA

- These are called **sequence variants**

Person 1	A	T	C	G	C	A	G
Person 2	A	T	C	G	C	C	T

We all have differences in our total amount of DNA

- These are called **copy number variants (CNVs)**. This is either:
 - **Missing DNA**
(chromosomal deletion)
 - **Extra DNA**
(chromosomal duplication)



Genetic Testing

Genetic testing may be **targeted** or **broad**

- **Targeted** genetic testing looks at **certain genes** known to be associated with a specific health concern
- **Broad** genetic testing (“exome” or “genome”) looks at **all your child's genes**
It looks for an explanation for:
 - Your child’s current health concerns
 - Other possible growth and development differences that may present in childhood

Types of results from genetic testing



- **Genetic explanation** for child's history

- **Incidental findings** (see next page)

- **Unexpected results**



- Parent found to be unrelated to child
 - Non-paternity
 - Non-maternity
- Biological relationship between parents (such as being cousins)

Incidental Findings

- With **broad** genetic testing, there is the possibility of detecting an **incidental finding**
 - An **unexpected genetic diagnosis** that does not explain the patient's current concerns or history
 - Most adult-onset conditions are not included
 - **The lab can filter out these findings if you do not want to know about them**

Possible test results

- **Informative/Positive**



- The genetic test identified a pathogenic variant
- This DNA change provides us with a diagnosis

- **Negative**



- The genetic test did not identify any pathogenic variants
- Genetic testing was not informative for your child in (Month/Year)

- **Inconclusive genetic test results**



- The genetic test did not identify a clear-cut answer
- Additional testing or screening may be recommended for your child or for you (their parent)

Inheritance

Any DNA variant can be:

- **New ("*de novo*")**

- New variant in person tested

OR

- **Inherited**

- Same (or related) variant in parent

DNA variants can be passed through families in different patterns

- We'll review a specific inheritance pattern once we have a DNA change to follow thru your family

Privacy

Genetic information is private - *you choose who to tell about a diagnosis*

There are laws that protect you and your child from being discriminated against based on a genetic diagnosis:

- **HIPAA:** The Health Insurance Portability and Accountability Act
 - Protects sensitive health information from disclosure without your consent
- **GINA:** Genetic Information Nondiscrimination Act
 - Protects against employment and health insurance discrimination
 - Has some exceptions such as:
 - Long term care insurance
 - Disability Insurance
 - Life Insurance
 - US Military and Federal Employees
 - Indian Health Insurance
 - Employers with fewer than 15 employees
- **ACA:** Affordable Care Act
 - Protects against discrimination based on pre-existing conditions

Parent resources for understanding ES/GS/rGS

- Exome & Genome Sequencing - Informed Consent video

- [Exome/Genome Sequencing Informed Consent - NICU](#)
- By Sara Zoran, CGC

- Unique 'DNA sequencing' guide

- [https://rarechromo.org/media/information/Other%20Topics/DNA%20sequencing%20\(whole%20genome%20and%20exome\)%20FTNW.pdf](https://rarechromo.org/media/information/Other%20Topics/DNA%20sequencing%20(whole%20genome%20and%20exome)%20FTNW.pdf)



Provider resources for ES/GS consenting best practices

- ACMG policy statements and practice guidelines
 - <https://www.acmg.net/ACMG/Advocacy/Policy-Statements.aspx?WebsiteKey=6e814a8c-3077-4552-ba39-f7fcacff42d6>
 - <https://www.acmg.net/ACMG/Medical-Genetics-Practice-Resources/Practice-Guidelines.aspx>
- Baby Badger network resources
 - <https://geneticsinwisconsin.wisc.edu/baby-badger-network/>
 - BBN ECHO training sessions from 2024, 2025

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Parental samples

- Strongly recommended for any broad testing
 - Provide a familial reference for the millions of DNA variants that each of us have
- Provide familial data that helps with variant interpretation
 - Is this DNA change *de novo*?
 - Are these variants in *trans*? for recessive conditions
- Provide key information for other family members
 - Who else in the family should we be testing?
 - What is the recurrence likelihood for this condition?

Parental samples

- Some parents are interested in providing samples, others aren't. Others are unavailable.
 - By obtaining buccal or saliva samples we can lower the burden of needing to register, blood draw, etc
 - Commonly cited reasons for not providing samples include:
 - Timeliness
 - Forgetfulness
 - Emotional burden
 - Privacy concerns

Parental samples

- Have collection kits readily available
 - Ensure that you're obtaining a validated sample type for the lab you're sending
 - Exomes: buccal, saliva, blood
 - Genomes: blood, occasionally saliva, occasionally buccal
- Label collection kit for each parent
 - Be present for sample collection
 - Coordinate with bedside team to remind parents to provide samples

Common challenges

- Ideally, you complete consent and sample collection in one visit with a family
 - Highly unlikely but you can set yourself up for success
 - Prepare by creating consenting materials/resources
 - Have any consent documents (that require parents' signatures) printed
 - Have buccal/saliva collection kits on hand
- Family may need multiple check-ins
 - Decision fatigue
 - Emotional burden

Lab specific requirements for consent

What requirements does the lab have?

- Familiarize yourself with the test requisition form (TRF) prior to sending out the analysis
- Does it require a signature? From whom?
 - Parent/legal guardian?
 - Ordering provider?

PATIENT TEST SELECTION		
STANDARD DIAGNOSTIC PGnome ☐ PATIENT ONLY Test Code 7000 FAMILY ☐ DUO Test Code 8000 ☐ TRIO Test Code 8001 ☐ OTHER Specify _____ Complete a Comparator Test Requisition form for each family member. Clinical information is REQUIRED for each comparator for accurate interpretation. ☐ Include family/comparator demographics (name, DOB, ID#, and relationship) on the proband report.	SECONDARY (ADDITIONAL) FINDINGS Details can be found in the PGnome Healthcare Provider Statement. Options for reporting of Secondary Findings are to be marked below. ☐ OPT IN: GUIDELINE RECOMMENDED GENES TRIO (WITH PARENTS) ONLY ☐ OPT IN: PG DISCOVERY OPTIONAL SECONDARY FINDINGS ADD-ON Details can be found in the PGnome Healthcare Provider Statement. Marking one or both of the options below will add an additional \$590 USD to your order. ☐ OPT IN: OTHER PREDISPOSITIONS / DIAGNOSES ☐ OPT IN: CARRIER STATUS	COMMENTS OR SPECIAL INSTRUCTIONS: ☐ RE-ANALYSIS Re-analysis will be completed with original secondary finding selections unless otherwise specified. Changes to secondary finding opt ins or structure of the analysis (additional comparators) may result in additional charges. See our website for full re-analysis policy. Test Code _____ Desired secondary findings (choose opt ins at left) _____
RAPID DIAGNOSTIC PGnome ☐ PATIENT ONLY Test Code 14000 ☐ FAMILY - DUO, TRIO, ETC Codes Duo-14001/Trio-14002 Complete a Comparator Test Requisition form for each family member. Clinical information is REQUIRED for each comparator for accurate interpretation.	NO SECONDARY FINDINGS AVAILABLE FOR RAPID PGNAME ☐ Include family/comparator demographics (name, DOB, ID#, and relationship) on the proband report.	
HEALTH SCREEN PGnome ☐ PATIENT ONLY Test Code 9000 Includes carrier status.	SECONDARY (ADDITIONAL) FINDINGS Details can be found in the PGnome Healthcare Provider Statement. Options for reporting of Secondary Findings are to be marked below. ☐ OPT IN: GUIDELINE RECOMMENDED GENES ☐ OPT IN: OTHER PREDISPOSITIONS / DIAGNOSES	
I have provided informed consent to my patient. My patient has had the opportunity to ask questions. Please indicate patient preferences for secondary findings on the PGnome® Test Requisition Form.		
HEALTHCARE PROVIDER SIGNATURE	PRINTED NAME	DATE

financially responsible for any and all amounts as indicated on the explanation of benefits issued by my health insurance plan. If my insurance provider sends a payment directly to me for services performed by GeneDx on my behalf, I agree to endorse the insurance check and forward it to GeneDx within 30 days of receipt as payment towards GeneDx's claim for services rendered.

By signing this form: (i) I acknowledge that I have read or have had read to me the GeneDx Informed Consent document, and understand the information regarding genetic testing; (ii) I have had the opportunity to ask questions about the testing, the procedure, the risks, and the alternatives; (iii) I authorize GeneDx to perform genetic testing as ordered; (iv) I understand that, for tests that evaluate data from multiple family members concurrently, test results from these family members may be included in a single comprehensive report that will be made available to all tested individuals and their healthcare providers; (v) if at any time I or my provider provide an email address or mobile phone number at which I may be contacted, I consent to receiving email or text messages from GeneDx; and (vi) I understand that this consent applies to all future communications unless I request a change in writing.

☐ **Secondary Findings Opt-out.** Check this box if you do not wish to receive ACMG secondary findings (Full Exome Sequencing and Genome Sequencing Tests ONLY; not for Xpanded® or Slice tests).

☐ **New York Retention Opt-in.** By checking this box, I confirm that I am a New York State resident, and I give permission for GeneDx to retain any remaining sample longer than 60 days after the completion of testing, and to be used as a de-identified sample for test development and improvement, internal validation, quality assurance, and training purposes. Otherwise, New York law requires GeneDx to destroy my sample within 60 days, and it cannot be used for test development studies.

☐ **Patient Research Opt-out.** Check this box if you wish to opt out of being contacted for research studies.

☐ **Health Information Exchange Opt-in.** Check this box if you reside in CA, FL, MA, NV, NY, RI, and VT and wish to opt-in to my health information to be shared for Health Information Exchange participation.

☐ **Health Information Exchange Opt-out.** Check this box if you reside in any other US state or territory and wish to opt-out of participation in Health Information Exchange.

Signature of Patient/Legal Guardian (required)		Date
Signature of Relative A/Legal Guardian	Relative A Relationship to Patient	Date
Signature of Relative B/Legal Guardian	Relative B Relationship to Patient	Date

Lab specific requirements for consent

☐ Include family/comparator demographics (name, DOB, ID#, and relationship) on the proband report.	☐ OPT IN: OTHER PREDISPOSITIONS / DIAGNOSES ☐ OPT IN: CARRIER STATUS
RAPID DIAGNOSTIC PGnome	
☐ PATIENT ONLY Test Code 14000	☐ NO SECONDARY FINDINGS AVAILABLE FOR RAPID PGNAME
☐ FAMILY - DUO, TRIO, ETC Codes Duo-14001/Trio-14002 Complete a Comparator Test Requisition form for each family member. Clinical information is REQUIRED for each comparator for accurate interpretation.	☐ Include family/comparator demographics (name, DOB, ID#, and relationship) on the proband report.
HEALTH SCREEN PGnome	
SECONDARY (ADDITIONAL) FINDINGS	

By signing this form: (i) I acknowledge that I have read regarding genetic testing; (ii) I have had the opportunity to perform genetic testing as ordered; (iv) I understand family members may be included in a single comprehensive report any time I or my provider provide an email address or GeneDx; and (vi) I understand that this consent applies:

- ☐ **Secondary Findings Opt-out.** Check this box if you **DO NOT** want to receive secondary findings (ONLY; not for *Xpanded*[®] or Slice tests).
- ☐ **New York Retention Opt-in.** By checking this box, I agree to allow my sample longer than 60 days after the completion of testing for validation, quality assurance, and training purposes for test development studies.
- ☐ **Patient Research Opt-out.** Check this box if you **DO NOT** want to participate in research.
- ☐ **Health Information Exchange Opt-in.** Check this box if you **DO** want to participate in Health Information Exchange participation.
- ☐ **Health Information Exchange Opt-out.** Check this box if you **DO NOT** want to participate in Health Information Exchange.

Signature of Patient/Legal Guardian (required)

Simple solutions

- The patient should be actively encouraged to be part of the conversation and be given the opportunity to ask questions throughout the conversation.
- Healthcare professionals should attempt to elicit the patients' views and questions. In addition to closed questions such as *"Do you have any questions?"*, open questions may be particularly effective, e.g. *"What questions do you have?"*, *"What are your thoughts about that?"*, and *"What else can I tell you about?"*.
- Healthcare professionals should use effective communication and listening techniques to identify and respond to any queries or concerns. These include question asking (e.g. *"What are your thoughts about that?"*), active listening (e.g. summarising what the patient has said) and ceding the floor.
- Healthcare professionals will need to have sufficient knowledge to talk about potential secondary findings that may arise from genome sequencing with patients.
- Healthcare professionals will also need to have sufficient knowledge to answer patients' questions relating to genetics and inheritance patterns, or at a minimum know where to direct patients if they are unsure.

Sanderson et al, 2019

Summary

- Consenting patients/families to rGS can present many logistical and emotional barriers
- Multiple check-ins gives families time to process information being shared and provides multiple opportunities for them to share their concerns, questions, etc
 - Benefits your therapeutic relationship with family

Summary

- By agreeing to any genetic testing (including rGS), a family is agreeing to possible inconclusive and unexpected test results
- Are they also agreeing to:
 - Incidental findings?
 - Secondary findings?
 - Data sharing? and/or Research consent?
 - Parental samples?

Literature Reviewed

1. ACMG Board of Directors. Points to consider for informed consent for genome/exome sequencing. Genet Med. 2013 Sep;15(9):748-9. doi: 10.1038/gim.2013.94. Epub 2013 Aug 22. PMID: 23970068.
2. Sanderson SC, Lewis C, Patch C, Hill M, Bitner-Glindzicz M, Chitty LS. Opening the "black box" of informed consent appointments for genome sequencing: a multisite observational study. Genet Med. 2019 May;21(5):1083-1091. doi: 10.1038/s41436-018-0310-3. Epub 2018 Oct 1. PMID: 30270361; PMCID: PMC6752270.