

Knowledge Burst | Before the Test: Informed Consent and Collecting Samples

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Learning Objectives

1. Identify the ACMG recommendations for obtaining informed consent prior to performing genomic testing
2. Discuss 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 for genomic testing



ACMG recommendations

- 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.



ACMG recommendations continued

- 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.



ACMG recommendations continued

- 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? Trinucleotide repeats?
- Patients should be informed of policies regarding re-contact of referring physicians as new knowledge is gained about the significance of particular results.



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Best practices for being a science communicator

- Share big ideas first, fill in with details
 - 'need to know vs nice to know'
- Practice 'teach back' method to verify comprehension
- Analogies can be helpful
 - But be wary of taking them too far
- Watch for non-verbal cues
 - Families are often emotionally overburdened and have information overload
 - Attempt to lessen decision fatigue for parents
- Validate the uncertainty



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

DOI: [10.1038/s41436-018-0310-3](https://doi.org/10.1038/s41436-018-0310-3)



Goals of genetic testing

- Identify spelling changes in DNA that could explain your child's medical concerns
- Answer **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

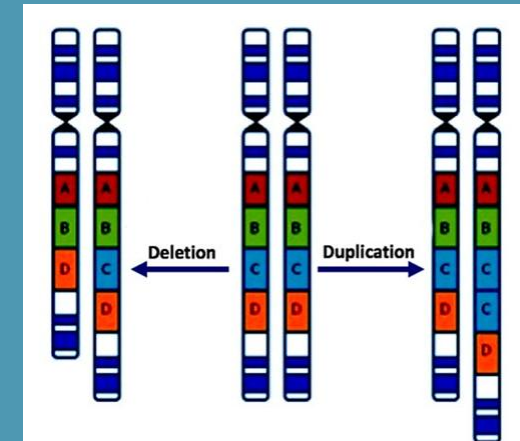


Basic background information

- No one has 'perfect' DNA
- We all have DNA changes
 - Misspellings
 - Missing or extra pieces
 - Inappropriate repeats
- This is a broad genetic test (genome) will identify your baby's DNA changes
 - The impact of these DNA changes may be significant, unimportant, or unknown

Person 1

Person 2



Categories of test results

Expected: DNA explanation for child's medical history

Unexpected:

- Familial:
 - Non-parentage found to be unrelated to child
 - Non-paternity, non-maternity
 - Parents are related by blood
 - Consanguinity
- Medical:
 - Incidental:
 - Secondary: as defined by ACMG (adult onset predispositions that have associated medical management)
 - Other: pharmacogenomic, mitochondrial DNA variants, trinucleotide repeat disorders



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Patient Resources

Exome & Genome Sequencing - Informed Consent video

–https://mediaspace.wisc.edu/media/Exome%20%20Genome%20Sequencing%20-%20Informed%20Consent/1_puirsf9q

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)



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Provider Resources

- ACMG policy statements and practice guidelines

Lee K, et al, ACMG SF v3.3 list for reporting of secondary findings in clinical exome and genome sequencing: A policy statement of ACMG). Genet Med. 2025 Aug;27(8):101454. PMID: 40568962;
Green RC, et al; ACMG recommendations for reporting of incidental findings in clinical exome and genome sequencing. Genet Med. 2013 Jul;15(7):565-74. doi: 10.1038/gim.2013.73. Epub 2013 Jun 20. Erratum in: Genet Med. 2017 May;19(5):606. PMID: 23788249;

- Baby Badger Network resources

–<https://geneticsinwisconsin.wisc.edu/baby-badger-network/>
BBN ECHO training sessions



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Parental Samples: Benefits

- 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: Difficulties

- 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: Best Practices

Have collection kits readily available

- Ensure that you're obtaining a validated sample type.
 - Exomes: buccal, saliva, blood
 - Genomes: blood, occasionally saliva, occasionally buccal

Label collection kit for each parent

- Be present for sample collection (if possible)
- Coordinate with bedside team to remind parents to provide samples



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?

