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## GeneReviews Resource Materials

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Note: These resource materials, provided by the editors of *GeneReviews*, include the following:

- Educational Materials, intended for individuals of varying experience with clinical genetics and designed to clarify genetics concepts presented in *GeneReviews* chapters – as the *GeneReviews* glossary clarifies our use of genetic counseling and testing terms
- Resources for Genetics Professionals

*GeneReviews* Resource Materials will be updated as needed, and new materials added as they are completed. Reader comments and suggestions are [welcome](#). — The Editors

## Educational Materials

### Genetic Testing: Current Approaches (revised 6-18-20)

Introductory Information

Detailed Information for Clinicians Ordering Genetic Tests

References

## Resources for Genetics Professionals

### Limitations of Clinical Exome Sequencing: Genetic Disorders Not Reliably Diagnosed

Genetic Disorders Caused by Imprinting Errors and Uniparental Disomy Not Detectable by Sequence Analysis (3-14-17)

Genetic Disorders Caused by Nucleotide Repeat Expansions and Contractions (revised 10-20-22)

Genes with Highly Homologous Gene Family Members or a Pseudogene(s) (3-8-18)

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## **Epigenetic Signature Analysis: An Introduction (revised 6-30-22)**

An introduction to epigenetic signature analysis and comparison of gene-specific genome-wide methylation analysis and epigenetic modifications to specific regions of the genome

## **Mosaicism (10-27-22)**

The occurrence within a single individual or tissue of two or more cell lines with a different genetic or chromosomal composition, involving somatic cells, germline cells, and/or tumor cells. Implications for molecular genetic testing and genetic counseling are discussed.

## **Founder Variants by Population: Genes and Disorders (revised 12-7-23)**

Pathogenic variants observed at high frequency in specific populations due to the presence of the variant in a single ancestor or small number of ancestors

- [Acadian \(5-26-22\)](#)
- [Afrikaner \(8-11-22\)](#)
- [Amish \(revised 12-7-23\)](#)
- [Apache \(revised 1-12-23\)](#)
- [Ashkenazi Jewish \(revised 10-20-22\)](#)
- [Bedouin \(8-18-22\)](#)
- [Bukharan Jewish \(9-29-22\)](#)
- [Choctaw \(revised 6-1-23\)](#)
- [Costa Rican \(6-30-22\)](#)
- [Cree and/or Ojibway \(revised 6-1-23\)](#)
- [Druze \(revised 9-28-23\)](#)
- [Faroe Islander \(revised 1-12-23\)](#)
- [Finnish \(revised 5-4-23\)](#)
- [Georgian Jewish \(9-29-22\)](#)
- [Gitxsan \(8-11-22\)](#)
- [Hutterite \(revised 12-1-22\)](#)
- [Inuit \(revised 8-24-23\)](#)
- [La Réunion Island \(revised 10-19-23\)](#)
- [Lumbee \(revised 1-12-23\)](#)
- [Māori \(1-12-23\)](#)
- [Mennonite \(6-1-23\)](#)
- [Métis \(6-16-22\)](#)
- [Navajo \(revised 11-3-22\)](#)
- [Newfoundland and Labrador \(revised 8-17-23\)](#)
- [Pueblo \(9-29-22\)](#)
- [Romani \(8-11-22\)](#)
- [Sephardic Jewish \(1-12-23\)](#)
- [Yup'ik \(revised 6-30-22\)](#)
- [Zuni \(8-11-22\)](#)

## **Direct-to-Consumer Testing (6-13-19)**

## **The Undiagnosed Diseases Network (3-15-18)**

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