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Genetic Testing and a Real World Case of Lynch Syndrome

Paige Montanaro – Cell and Molecular Biology

Sponsor: Dr. Joel Chandlee

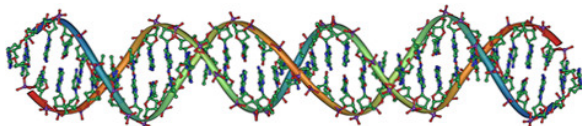


Introduction

In recent years, advancements in genetic testing methods have revolutionized the medical field by enhancing the ability to identify persons with an inherited predisposition to cancer. This project was designed to investigate the methods used in genetic testing as well as demonstrate the importance of implementing genetic tests to individuals who appear to be at risk for cancer.

Methods

To complete this project, I did research on the methods used in genetic testing, specifically for those with an inherited predisposition to cancer. To supplement this research, a real world case study was evaluated. An individual who underwent the Myriad myRisk® genetic test and presented with a mutation known as Lynch Syndrome was studied. I produced an inclusive research paper that highlights the positive impact of genetic testing by dissecting the pathogenesis of Lynch Syndrome at a molecular level.



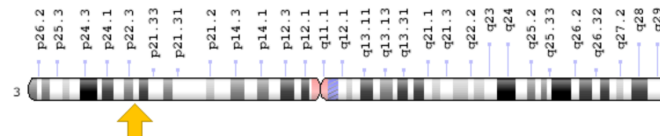
Results

Genetic Testing

Type of Test	Test For	Example
Cytogenetic	Chromosomal Abnormality	Fluorescence in-situ hybridization
Biochemical	Protein Function	Tandem mass spectrometry
Molecular	DNA sequence	Comparative genomic hybridization

Classification of the 3 main types of genetic testing (1).

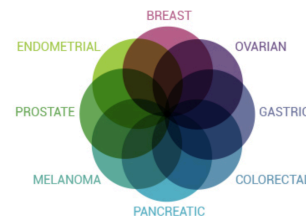
Geneticists search for specific markers within the human genome that are indicators of an inherited cancer syndrome.



Cytogenetic location of MLH1 gene on chromosome 3 3p22.2 (3).

Case Study

The patient underwent the Myriad myRisk® test, which uncovered a genetic mutation in her genome classified as Lynch Syndrome. Testing revealed the mutation **c.113del (p.Thr45Glnfs*5)**. This mutation results in premature truncation of the MLH1 protein.

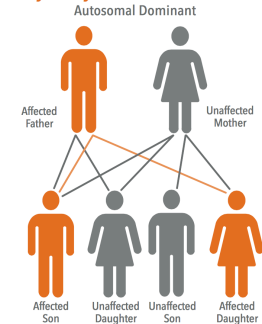


The Myriad myRisk® Hereditary Cancer Test is a 28-gene panel that identifies an elevated risk for 8 cancers (2).

Lynch Syndrome

Lynch syndrome, or hereditary non-polyposis colon cancer (HNPCC), is an autosomal dominant disease that leads to colon cancer as well as increased risk to many other cancers due to the inactivation or altered function of integral DNA mismatch repair genes.

Lynch Syndrome Inheritance



Pattern of inheritance in Lynch Syndrome families

- Mismatch repair genes mutated in LS: MLH1, MSH2, MSH6, PMS2, EPCAM
- 3-5% of colorectal cancers arise from LS
- One of the most common hereditary cancer syndromes
- Increased risk of many abdominal and reproductive cancers

Acknowledgements

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Sources

1. "Genetic Diagnosis and Testing in Clinical Practice." *Clinical Medicine and Research*
2. "Myriad MyRisk® |." *Myriad Genetics*, myriad.com/managed-care/myriad-myrisk/.
3. "MLH1 Gene - Genetics Home Reference." *U.S. National Library of Medicine, National Institutes of Health*