Identifying CYP4V2 mutation in Bietti’s crystalline dystrophy patient

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Abstract: A patient with Bietti’s crystalline dystrophy had their DNA extracted to be screened for a mutation in CYP4V2. The extraction of the DNA from the patient allowed the lab to locate the point mutation in the gene. Using PCR (polymerase chain reaction), exons 1 through 11 were amplified and then genotyped to see exactly where the point mutation occurs. After sequencing the patient’s DNA, a mutation on exon 9 (a combined sequence since both exons are short) was found. The mutation is expected to be a point mutation with C-T (cytosine – thymine) on nucleotide c1198.

Introduction

- Bietti’s crystalline dystrophy (BCD) is a disease that is found in the retina and/or cornea of the eye.
- BCD is characterized by yellow/white crystallization in the retina/cornea, and is thought to be caused by complex lipids localized in these regions.
- BCD is an autosomal recessive disease.
- The end stage of BCD is blindness.
- The early symptoms include: decline in central vision, night blindness, and gradual constriction of the visual field.
- The negative effects caused by BCD are associated with gene mutations in CYP4V2.
- CYP4V2 is closely studied when researching BCD because of mutations in the gene that causes the enzyme to not be functional.

Methods & Materials

1. PCR (Polymerase chain reaction)
   PCR amplifies a strand of DNA and creates many copies of that sequence.

2. Gel electrophoresis
   Gel electrophoresis is a method that separates DNA fragments based on size.

3. DNA purification
   DNA purification is the process of removing contaminants from DNA.

4. Genotyping
   Genotyping is the process of determining the genetic makeup of an individual.

Results

Bietti’s crystalline dystrophy (BCD) is caused by mutations in CYP4V2, which causes the illness. On exon 9 a C>T homozygous point mutation was found. The amino acid change is arginine to cysteine at nucleotide 400 (pR400C).

Conclusion

The patient analyzed by the lab has a mutation that is consistent with a diagnosis of BCD. The results from the sequence of exon 9 have shown that in fact the patient does have BCD because of a mutation at CYP4V2. There have been 60 other mutations found on the other exons (1 through 11), and the identification of this mutation has shown that there are other point mutations that can cause the illness.