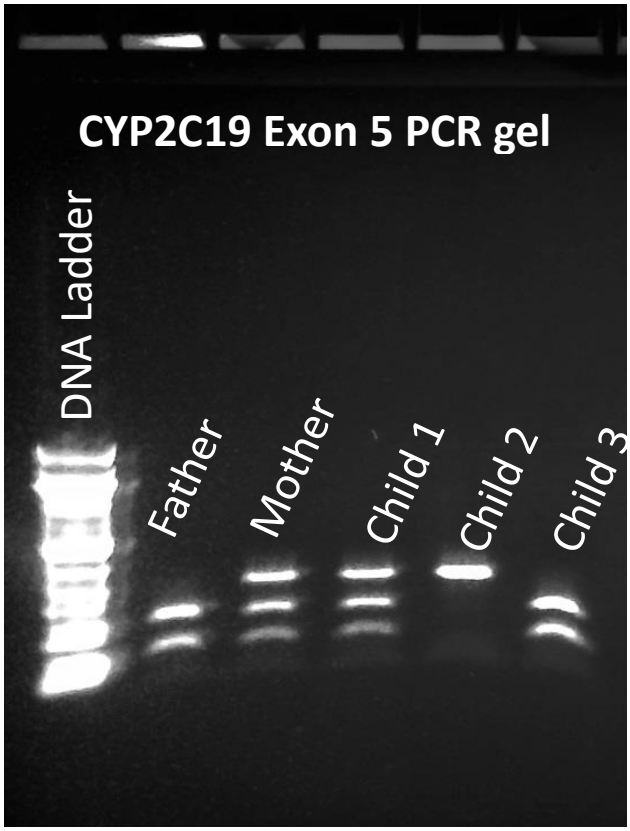
TEACHING THE GENOME GENERATION

Mendelian Genetics Activity – **Interpreting Gels**
Teacher Information

This student exercise compliments the laboratory activities in TtGG. In particular this short exercise uses the same gene test/assay (CYP2C19 exon) that students genotype through PCR and subsequent restriction digest. The goal of this activity is to get students to think about Mendelian inheritance from two parents to a group of their children. The example gel below shows a father and mother with the assumption that these are male and female biological parents. Three children are displayed. Students need to demonstrate that the bands on the gel are the parental alleles. In the case below the father is homozygous and a fast metabolizer; the mother is heterozygous (moderate metabolizer) with one allele of the gene/PCR product cut into the two smaller bands and the other allele uncut at ~320 bp. Child 1 is heterozygous and must have gotten the uncut/larger allele (320 bp) from the Mother and the smaller cut allele (two bands) from the Father. Child 2 is homozygous (poor metabolizer): there is a paternity issue here in Child 2; there is no allele from the Dad. Child could be from a different biological Father or could be adopted. Child 3 is homozygous (fast metabolizer) getting a similar allele from both the Mother and the Father. The student version follows this teacher version.



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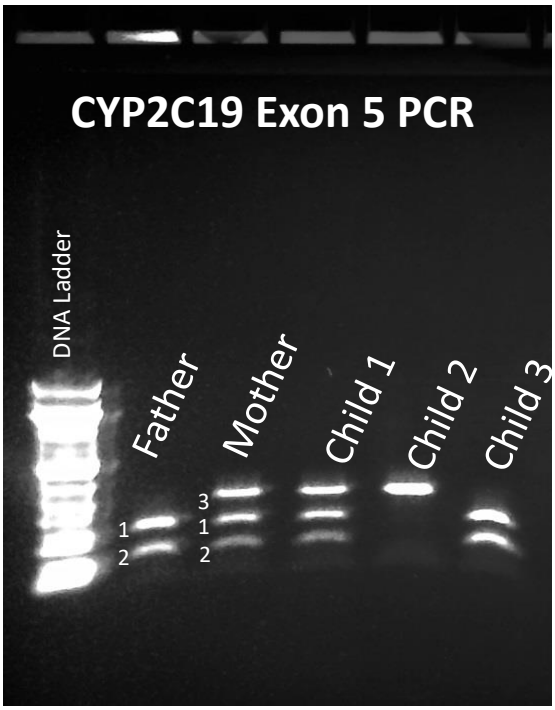
Introduction

In the laboratory you may be investigating DNA sequence variation in the human CYP2C19 gene.
In the laboratory, the presence of the G or A base is tested by amplifying the DNA sequence using Polymerase Chain Reaction (PCR), followed by restriction digestion of a ~320 bp PCR product;

- When G is present in two copies (one from each parent) then the PCR product digests into two bands (~110 bp and ~220 bp), bands 1 & 2 below.
- Heterozygous individuals (G/A, one from each parent) yield three bands; one parental band (G) gets digested in to two, one parental band (A) does not get digested, leaving a ~320 bp band, bands 1,2,3 below.
- In a homozygous AA individual (one A from each parent) no digestion occurs because the Restriction Enzyme (Sma I in this case) does not 'see' the proper DNA sequence to make a cut/digest.

In this exercise you need to explain the outcome of genotyping a family of five individuals shown on the gel to the right. There are two biological parents, biologically male and female. The parent's bands on the gel are given numbers 1-3. Using information from the introduction above and your knowledge of genetics explain how each child 1, 2, and 3 can end up with the genotype shown on the gel.



A. What is the Genotype of:
   Child 1:
   Child 2: _____
   Child 3:

B. What alleles (represented by numbered bands) did each child get from each parent?
   Child 1:
   Child 2:
   Child 3: _____

C. Is there anything unusual about the results on the gel?