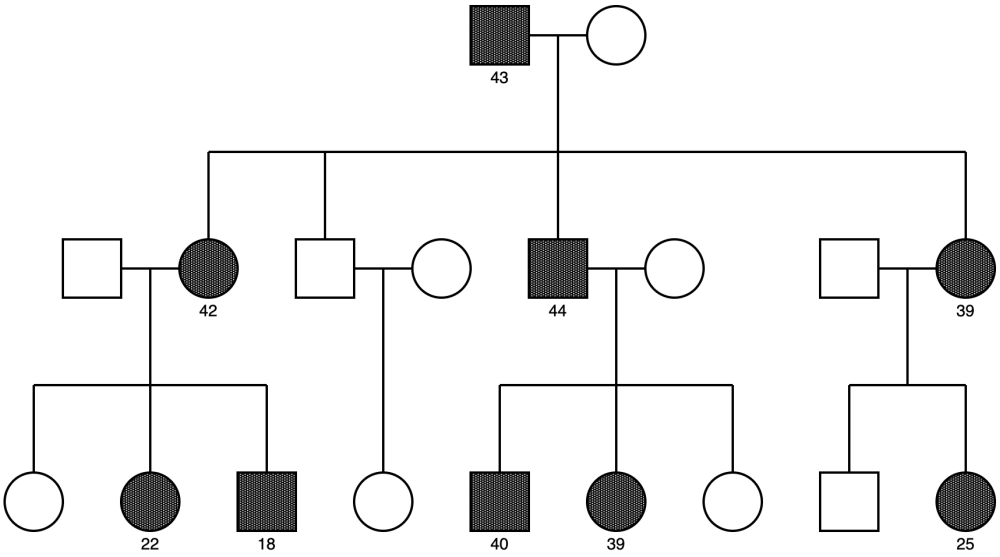
BIOL3120 Problem set 2

1. **1. Someone who is 75% mosaic for a mutation has had which of the following occur during their fertilization/development:**

   a) A germline mutation before fertilization
   b) A somatic mutation before fertilization
   c) A germline mutation after fertilization
   d) A somatic mutation after fertilization



2. This pedigree above shows a family with members affected with a dominant neurodegenerative condition. Numbers represent the age of onset for affected people. **What phenomenon can be observed in this family?**

3. A couple, neither of whom is affected, has a child with cystic fibrosis. The mother subsequently becomes pregnant with a new partner, a man who is unaffected himself and has unaffected parents but has an affected sister (they met at a cystic fibrosis charity event).

   What is the likelihood that this pregnancy is affected with cystic fibrosis?

4. **Which detection method(s) are appropriate for detecting copy number variants (CNVs) smaller than 2Mb?**

   (note, multiple answers can be selected - correct answers will gain you marks, incorrect answers will lose you marks, but your overall mark for this question can't be negative).
   a) Microarray
   b) Sanger sequencing
   c) Next generation sequencing
   d) Triplet-repeat primed PCR
   e) MLPA
   f) Karyotype

5. Assume eye colour is a simple mendelian trait, with the brown eye allele dominant, and the blue eye allele recessive (in reality it's more complex). In a certain random mating population, 9% of people have blue eyes. In this population:

   a) What proportion of brown-eyed persons would be heterozygous for blue eyes?

   b) What would be the predicted frequency of blue-eyed children among offspring of parents, both of whom are brown-eyed?

6. In a population of interest, 1 in 4000 women are affected with haemophilia A, an X-linked recessive condition. Assuming this population is mating randomly and available treatments mean haemophilia does not affect individual fitness.

   a) What is the carrier frequency in this population for Haemophilia A?

   b) What is the expected frequency of men affected with Haemophilia A?