

Answer Key

Part 1

Some issues include age of consent and competence (autonomy) and issue of secondary diagnosis from genetic testing such as finding a mutation in another gene that was not the initial reason to sequence the genome.

- I agree with the mother (or disagree with the daughter) and would do the test because the mother has her daughter's best interest and wants to know if she will develop breast cancer like herself
- I agree with the daughter (or disagree with the mother) because she has the right to her own privacy and if she is competent and understands the consequences then she can make her own decisions.
- Tell the mother that her daughter can wait till she is older to get tested so that if she does carry the mutation, then there are reasonable steps to follow.

Part 2

1. Point mutation of A to G at nucleotide #126
2. Inversion of TGAGA (#334-338) to AGAGT

Part 3

Gene #: 1, 5, 6, 7, 8, 10, and 11

Part 4

Patient 1

- Gene 1: 3 fold increase in proliferation
- Gene 5: BRCA1 mutation
- Gene 6: About 9 fold increase in invasion. Cells are invasive (tumour more likely spread)
- Gene 9: No difference between variant and normal (variation)
- Gene 12: increase in angiogenesis

Patient 2

- Gene 7: no difference but tendency to increase (somewhat invasive) or can say large error bars (not valid) or can say mutation has no different effect and could be a genetic variation
- Gene 8: reduction in the number of dead cells (sign of cancer)
- Gene 9: 1.5 fold increase in proliferation
- Gene 12: High expression of protein marker = increase in angiogenesis

Part 5

Patient 1 → Drug A or E to target proliferation, Drug D to target HER2-positive status, and enroll in clinical trial to test Drug II to prevent high rate of metastasis

Patient 2 → Drug B to target angiogenesis, Drug E to target cell division