

Medical Genetics Short Answer Question Exam

Part 1

1.

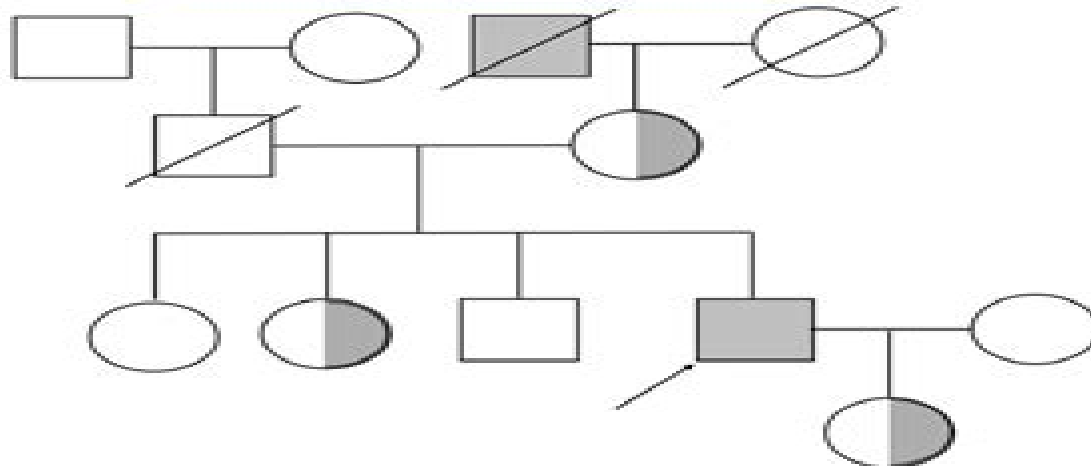
- a) Midline 1 (*MID1*)
- b) The gene has 11 aliases - OS; FXY; OSX; OGS1; XPRF; BBBG1; GBBB1; MIDIN; RNFS9; ZNFXY; TRIM18. These are names that were previously used for the gene but were unapproved. The *MID1* protein belongs to the Tripartite protein family (TRIM) which is why it has an alias known as TRIM18.
- c) Xp22.2
- d) *MID1* is located on the negative strand and is transcribed from right to left (indicated by arrows on diagram in Genome Browser). Genes located on the forward strand are transcribed from left to right and genes located on the reverse strand are transcribed from right to left, as RNA polymerase synthesises the strand in a 5' to 3' direction.
- e) NP_000372.1
- f) The *MID1* gene is transcribed from exon 1
- g) The *MID1* gene is expressed in many tissues, with lowest expression in the bone marrow, pancreas (RPKM 0.3) and spleen and high expression in the urinary bladder (RPKM 2.3), gall bladder and prostate (RPKM 2.5). This implies how organs with higher expression of the variant may be more severely affected by the disease caused by mutations in the *MID1* gene.

[MID1 midline 1 \[Homo sapiens \(human\)\] - Gene - NCBI \(nih.gov\)](#)

[MID1 Gene - GeneCards | TRIM18 Protein | TRIM18 Antibody](#)

[Human hg38 chrX:10445556-10833654 UCSC Genome Browser v413](#)

2. Opitz G/BBB is inherited as an X-linked genetic condition as the *MID1* gene is located on the X-chromosome. The condition is likely to be X-linked recessive. This is because in females who have two copies of the X chromosome, one altered copy of the gene causes less severe/no functions. [Opitz G/BBB syndrome: MedlinePlus Genetics](#)



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