#### Part 1

1.

- a) Midline 1 (MVD1)
- b) The gene has 11 aliases OS; FXY; OSX; OGS1; XPRF; BBBG1; GBBB1; MIDIN; RNF59; 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 prostrate (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 | TRI18 Protein | TRI18 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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