

Medical Genetics Short Answer Question Exam

Part 1

1.

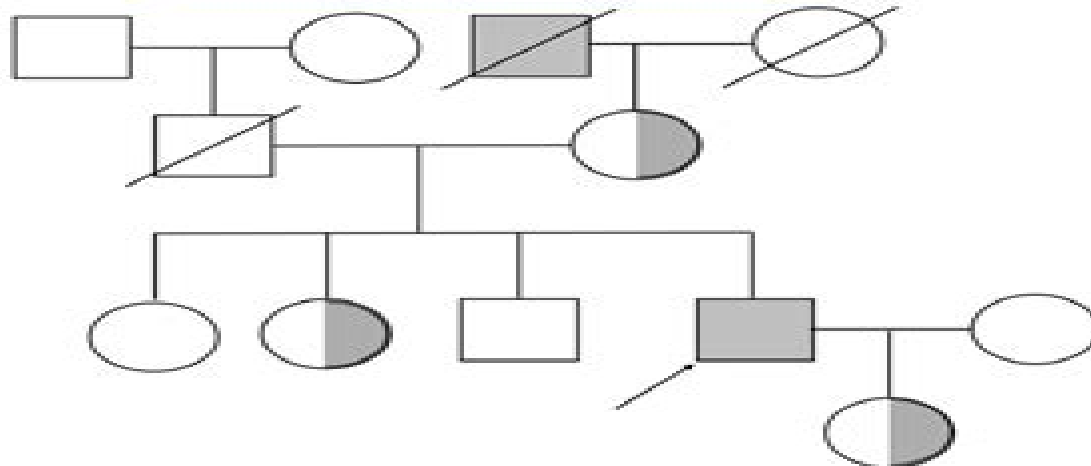
- a) Midline 1 (*MID1*)
- b) The gene has 11 aliases - OS; FXY; OSX; OGS1; XPRF; BBBG1; GBBB1; MIDIN; RNFS9; ZNFXY; TRIM18. These are names that were previously used for the gene but were unapproved. The *MID1* protein belongs to the Tripartite protein family (TRIM) which is why it has an alias known 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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AW Rasmussen



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- 3 Lethality and Interaction of Genes
- 4 Genetics of Quantitative Traits QTs
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- 5 Genetics of Quantitative Traits
- 2 Biometrical Approach
- 6 Genetics of

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Table of Contents Genetics Short Answer Questions With Answers

1. Understanding the eBook Genetics Short Answer Questions With Answers
 - The Rise of Digital Reading Genetics Short Answer Questions With Answers
 - Advantages of eBooks Over Traditional Books
2. Identifying Genetics Short Answer Questions With Answers
 - Exploring Different Genres
 - Considering Fiction vs. Non-Fiction
 - Determining Your Reading Goals
3. Choosing the Right eBook Platform
 - Popular eBook Platforms
 - Features to Look for in an Genetics Short Answer Questions With Answers
 - User-Friendly Interface
4. Exploring eBook Recommendations from Genetics Short Answer Questions With Answers
 - Personalized Recommendations
 - Genetics Short Answer Questions With Answers User Reviews and Ratings
 - Genetics Short Answer Questions With Answers and Bestseller Lists

5. Accessing Genetics Short Answer Questions With Answers Free and Paid eBooks
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 - Genetics Short Answer Questions With Answers Budget-Friendly Options
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 - Highlighting and Note-Taking Genetics Short Answer Questions With Answers
 - Interactive Elements Genetics Short Answer Questions With Answers
8. Staying Engaged with Genetics Short Answer Questions With Answers
 - Joining Online Reading Communities
 - Participating in Virtual Book Clubs
 - Following Authors and Publishers Genetics Short Answer Questions With Answers
9. Balancing eBooks and Physical Books Genetics Short Answer Questions With Answers
 - Benefits of a Digital Library
 - Creating a Diverse Reading Collection Genetics Short Answer Questions With Answers
10. Overcoming Reading Challenges
 - Dealing with Digital Eye Strain
 - Minimizing Distractions
 - Managing Screen Time
11. Cultivating a Reading Routine Genetics Short Answer Questions With Answers
 - Setting Reading Goals Genetics Short Answer Questions With Answers
 - Carving Out Dedicated Reading Time
12. Sourcing Reliable Information of Genetics Short Answer Questions With Answers
 - Fact-Checking eBook Content of Genetics Short Answer Questions With Answers
 - Distinguishing Credible Sources
13. Promoting Lifelong Learning

- Utilizing eBooks for Skill Development
- Exploring Educational eBooks

14. Embracing eBook Trends

- Integration of Multimedia Elements
- Interactive and Gamified eBooks

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