Case File 4: Marfan’s Syndrome

Joey Bradley is a 16 year old basketball star at Homefield High. He and two younger sisters are being evaluated at a genetics clinic to determine if they have Marfan’s syndrome. The Bradley family’s doctor had noticed that Joey and his sisters all had a particularly tall, thin body type along with very long fingers and near-sightedness, which are all characteristics of Marfan’s syndrome.

At the genetics clinic, information is gathered about Joey’s family history. Joey’s mother had a similar body type to himself and his sisters. She died several years ago because of a heart problem while swimming laps at a local pool. This also fits with having Marfan’s syndrome because certain heart conditions are common in individuals affected with this condition. Other family members as diagrammed below were described as having characteristics of Marfan’s.

The Bradley family history and physical examinations of Joey and his sisters provide enough evidence for the siblings to be diagnosed with Marfan’s syndrome without needing a genetic test.

1) What pattern of inheritance is seen in this family: Dominant, Recessive or X-linked recessive?

2) What are the genotypes of Joey, his siblings and parents?

Joey would like some information about Marfan’s syndrome.

3) Can you find an appropriate website or information on the internet to give him?

It is 20 years later. Joey is married and thinking of starting a family. His wife does not have Marfan’s syndrome and there is no history of it in her family.

4) What is the chance that they will have children affected with Marfan’s syndrome?

Bonus Question: Which American president from the 1800s is rumored to possibly have Marfan’s syndrome?
Case File 4: Answers

Question 1
ANSWER: Dominant Inheritance

EXPLANATION:
- Both males and females have the condition which makes X-linked inheritance very unlikely.
- There are many affected people in the family which makes recessive inheritance very unlikely.
- Dominant inheritance is the best fit.

Key points about classic Dominant inheritance:
- Many individuals are affected across many generations
- Both males and females are affected
- Affected individuals have affected parents

Question 2
ANSWER:
Joey: Dd
Affected sisters: Dd
Unaffected brother: dd
Mother: Dd
Father: dd

EXPLANATION:
- Let ‘D’ be the non-functioning (mutant) allele associated with Marfan’s. Let ‘d’ be the functional (normal) allele.
- We know that Joey, his sisters, and most likely his mother all have Marfan’s. This means they must each have one non-functioning ‘D’ allele.
- We know that Joey and his sisters must have inherited a functional (‘d’) allele from their unaffected father. Therefore Joey and his sister’s genotypes are ‘Dd’
- Joey’s mother must have inherited her non-functional ‘D’ allele from her father with Marfan’s. She would have inherited a functional ‘d’ allele from her unaffected mother. Therefore Joey’s mother’s genotype is also ‘Dd’.
- Joey’s brother and father are unaffected. They must have two functional (‘d’) alleles each and no non-functional ‘D’ alleles. Therefore their genotypes are ‘dd’.

Question 3
There are many good resources available that provide information articles and access to support groups for individuals and families affected by Marfan’s syndrome. Examples of some are:
- The Canadian Marfan Association’s website: www.marfan.ca
- The Your Genes Your Health website: www.yourgenesyourhealth.org

Question 4
ANSWER: There is a 1/2 or 50% chance of Joey and his wife having a child with Marfan’s syndrome

EXPLANATION:
- Joey’s genotype is Dd.
- His wife does not have Marfan’s therefore her genotype must be dd.
- Joey’s wife will always pass down a functional ‘d’ allele to her children
• Joey has a 50% (1/2) chance of passing down his functional ‘d’ allele and a 50% (1/2) chance of passing down his non-functional ‘D’ allele. This is diagrammed in the table below:

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<tbody>
<tr>
<td>D</td>
<td>Dd (Affected)</td>
<td>Dd (Affected)</td>
</tr>
<tr>
<td>d</td>
<td>Dd (Unaffected)</td>
<td>Dd (Unaffected)</td>
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</tbody>
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½ of their children will be Dd and therefore affected with Marfan’s. ½ of their children will be dd and not affected with Marfan’s.

**Bonus answer:**
While impossible to confirm and much debated, Abraham Lincoln has been suggested to have Marfan’s syndrome. Long after his death, some doctors speculated that he showed some features of Marfan’s syndrome based on their physical observations from portraits as well as descriptions of his body type in the literature.

**Reference:**