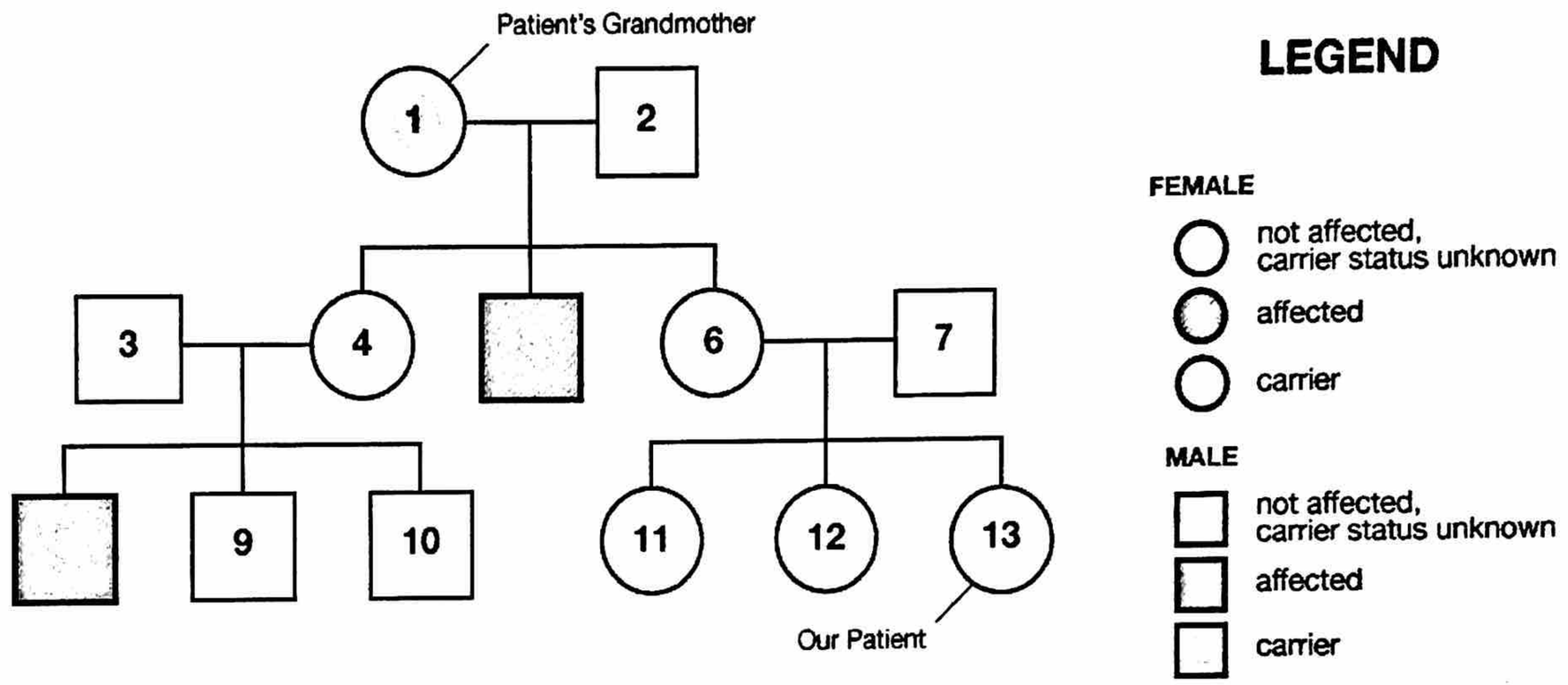
GENETICS LAB

Scenario One - Our patient seeks genetic counseling

Kayla has a family history of Duchenne muscular dystrophy (DMD), which is a sex-linked trait. The patient wishes to know her risk of having an affected child. Her grandmother was a known carrier. DMD is an inherited disorder that involves progressive muscle weakness. It affects approximately 1 in 3500 male births worldwide and is inherited in an X-linked recessive pattern.

Click here to learn more about DMD

The following pedigree was drawn up by the genetic counselor. Our patient, Kayla, is #13.



What are the relationships between:

1 and 2

1 and 5

1 and 10

6 and 7

4 and 5

5 and 8

8 and 10

1 and 3

What are the chances her mother was a carrier?

What are the chances our patient is a carrier?

What are the chances she passes the syndrome to a male child?

Testing would establish her status as either a carrier or a noncarrier.

If she is a carrier what are the chance of her having an affected child?

If she is not a carrier what are the chance of her having an affected child?

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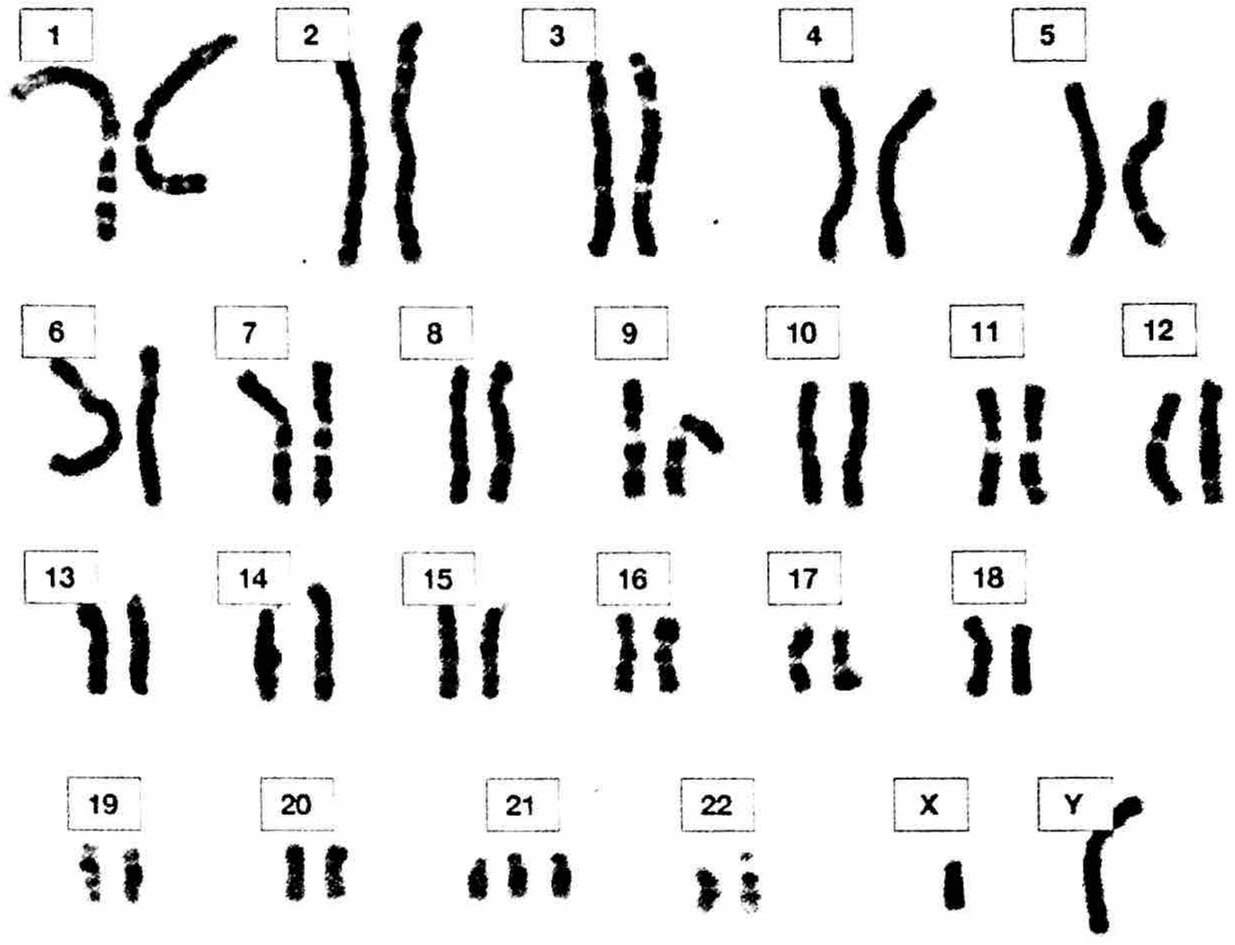
Scenario Two - Our patient seeks genetic counseling

Emily, a forty year old pregnant woman has an amniocentesis which provides the karyotype below.



Using the karyotype above answer the following questions:

What is the gender of the fetus? gender

Are there any chromosomal abnormalities? yes/no

What would this chromosomal combination result in? syndrome name

GO TO CHART >