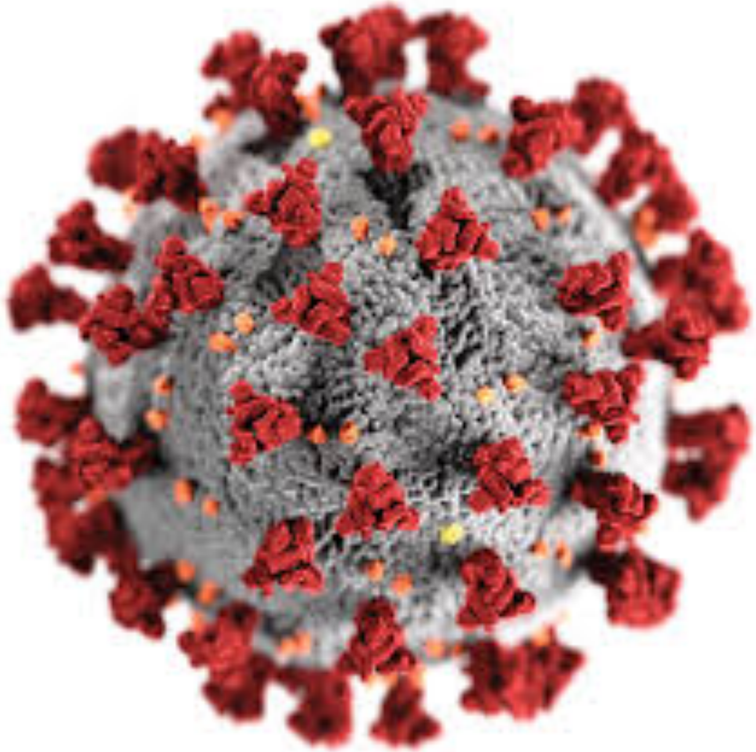
QUIZ SESSION 4
Pedigrees

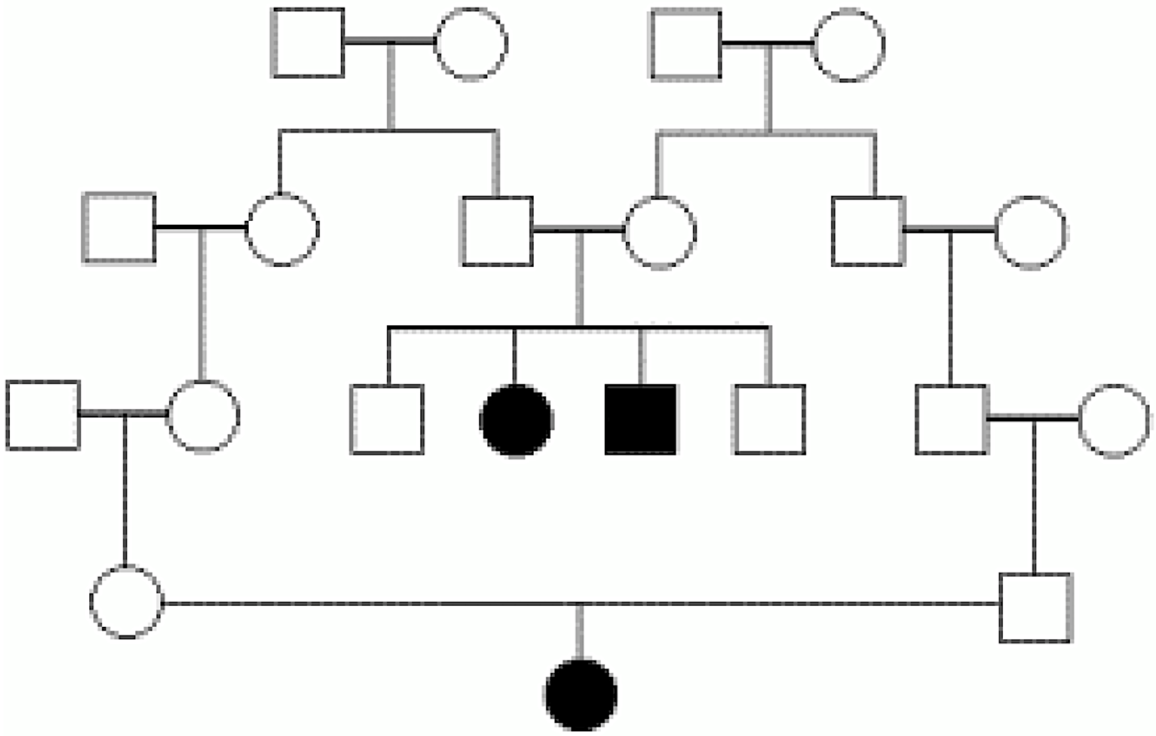


Quiz #1 – Identifying pedigrees (Lesson 16)

Q1. Identify this pedigree and explain

A1. **Autosomal recessive**

1) Both males and females affected (autosomal)
2) Affected children from unaffected parents (recessive)
3) Not many affected offspring (recessive)

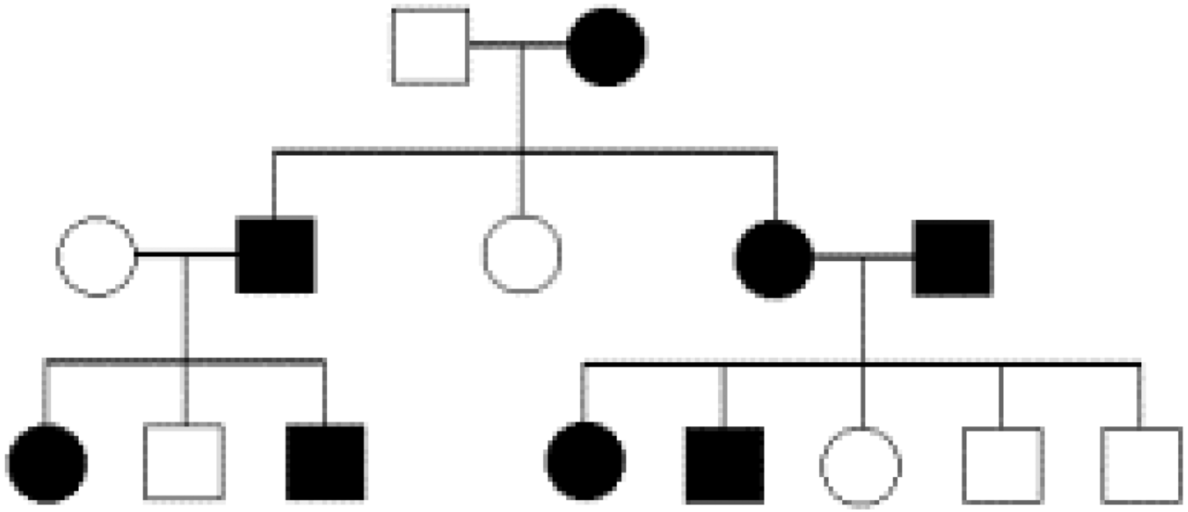


Quiz #2 – Identifying pedigrees (Lesson 16)

Q2. Identify this pedigree and explain

A2. **Autosomal dominant**

1) Both males and females affected (autosomal)
2) Every affected child has (at least) one affected parent (dominant)

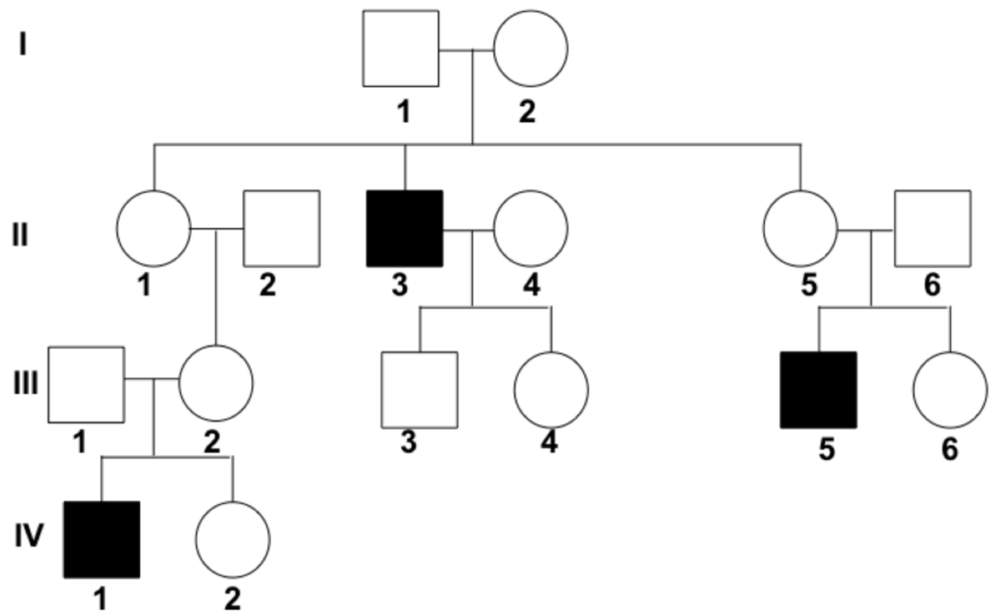


Quiz #3 – Identifying pedigrees (Lesson 16)

Q3. Identify this pedigree and explain

A3. **X-linked recessive**

1) Mainly males affected (X-linked)
2) Affected children from unaffected parents (recessive)
3) Not many affected offspring (recessive)

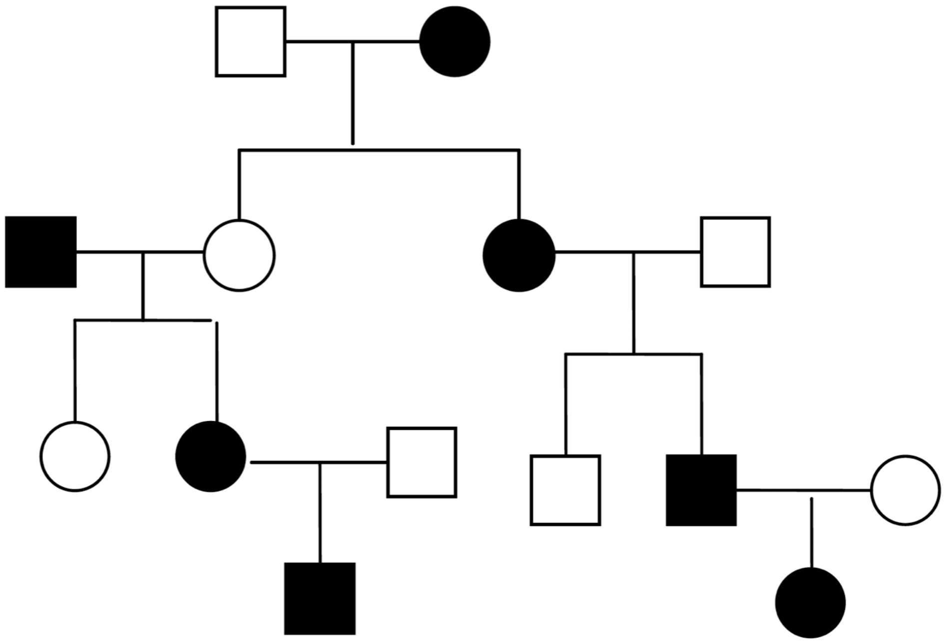


Quiz #4 – Pedigrees & genotypes (Lesson 16)

Q4. Identify this pedigree and explain

A4. **Autosomal dominant**

1) Both males and females affected (autosomal)
2) Every affected child has (at least) one affected parent (dominant)

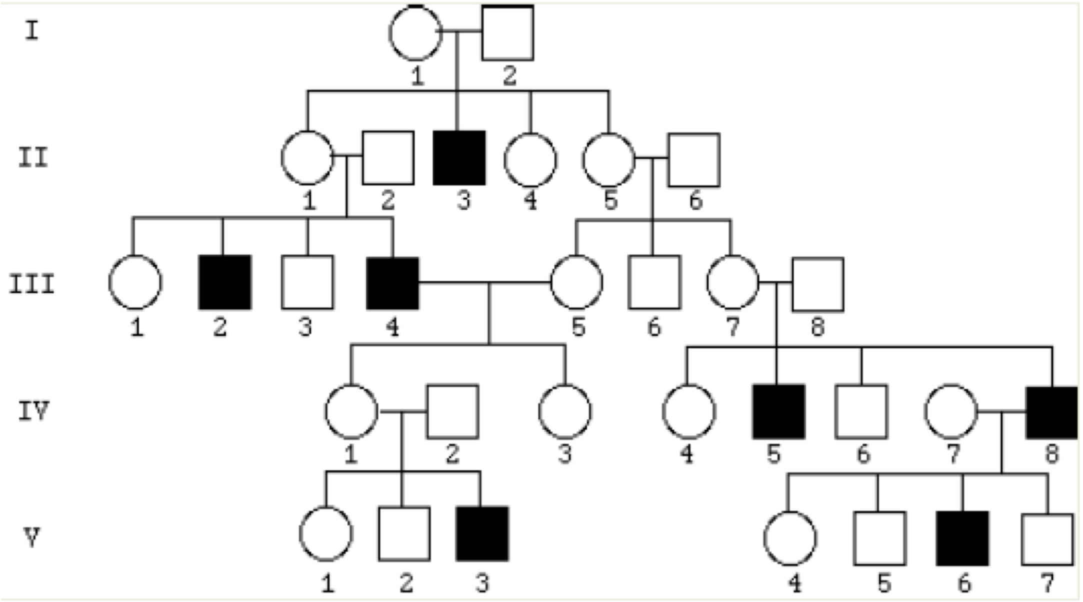


Quiz #5 – Identifying pedigrees (Lesson 16)

Q5. Identify this pedigree and explain

A5. **X-linked recessive**

1) Mostly males affected (X-linked)
2) Affected children from unaffected parents (recessive)
3) Not many affected offspring (recessive)

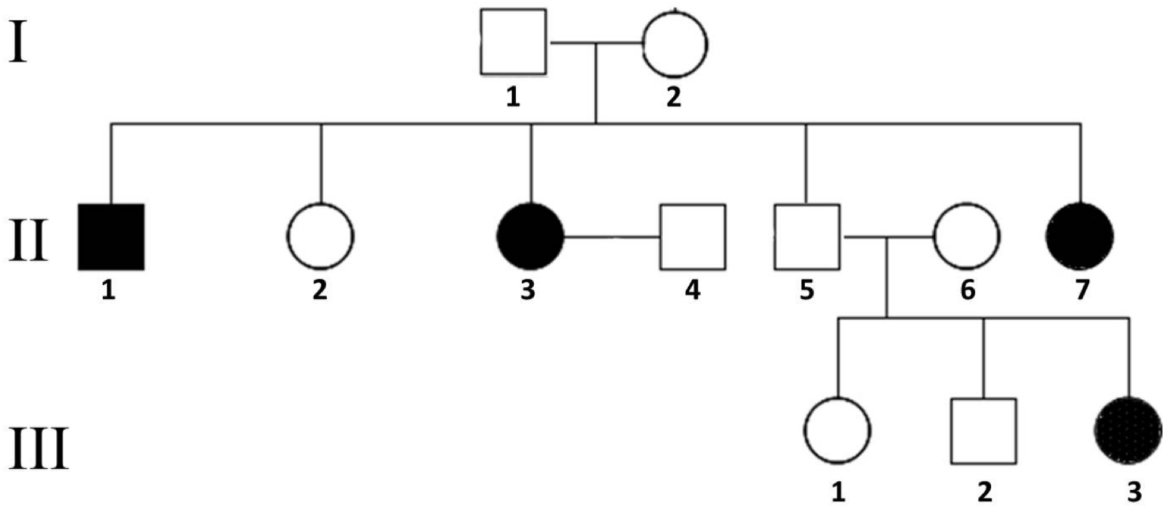


Quiz #6 – Identifying pedigrees (Lesson 16)

Q6. Identify this pedigree and explain

A6. **Autosomal recessive**

1) Both males and females affected (autosomal)
2) Affected children from unaffected parents (recessive)
3) Not many affected offspring (recessive)

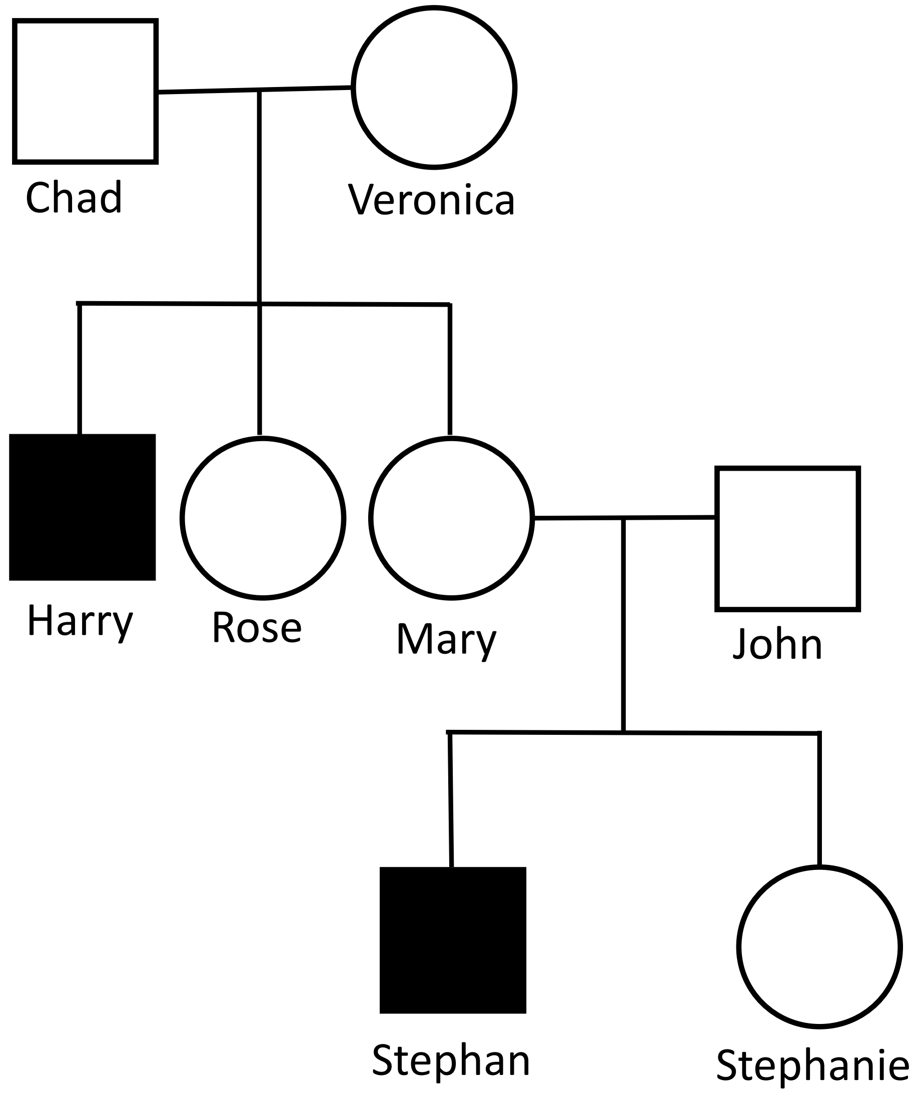


Quiz #7 – Drawing pedigrees (Lesson 16)

Chad and Veronica got married and had Rose, Mary and Harry. It was discovered that Harry had **Duchenne muscular dystrophy (DMD)**. Mary married John and had Stephan and Stephanie. Stephan also had DMD. John's brother Jim also had muscular dystrophy but neither of their parents had the disorder.

Q7a. Draw the pedigree based on the info above

A7a. See graph on right



Quiz #7 – Drawing pedigrees (Lesson 16)

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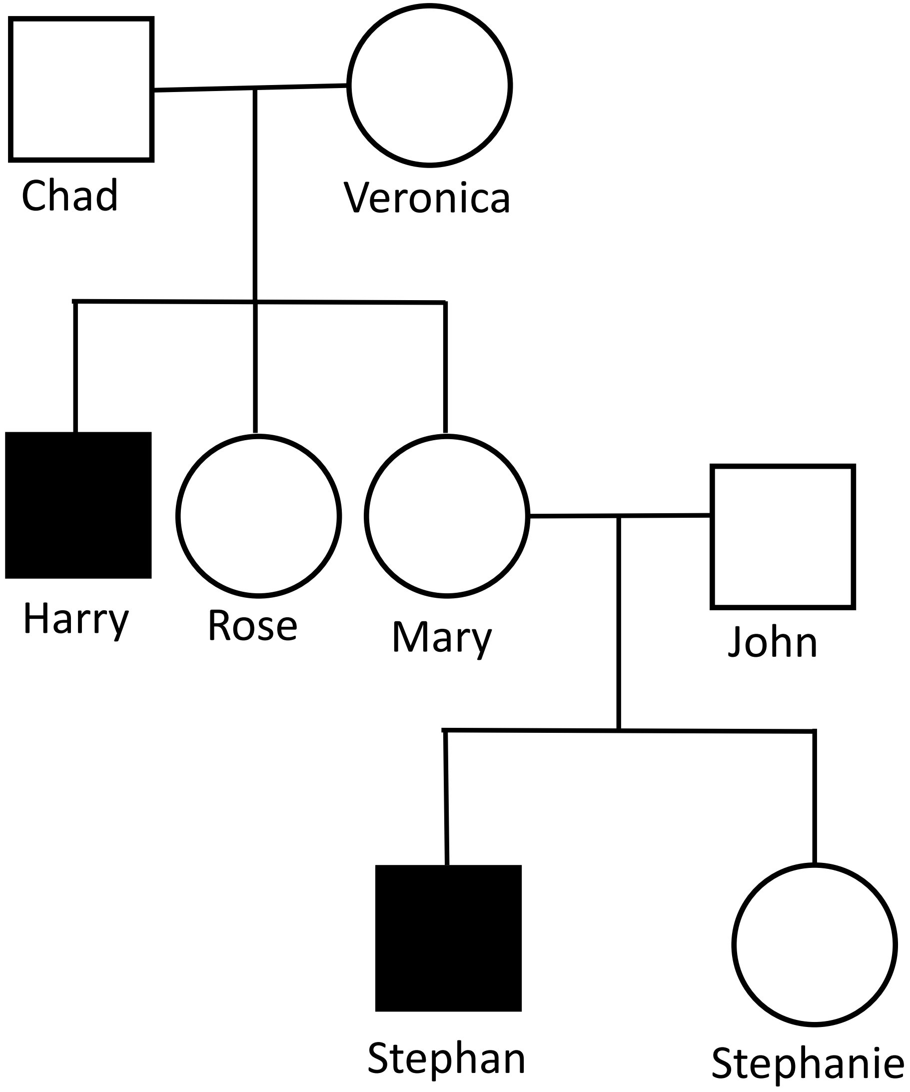
Q7a. Draw the pedigree based on the info above

A7a. See graph on right

Q7b. What type of disease is DMD? Explain

A7b. **DMD is an X-linked recessive disease**

a. Only male affected
b. Affected offspring from healthy parents (also Jim!)
c. Not too many affected offspring

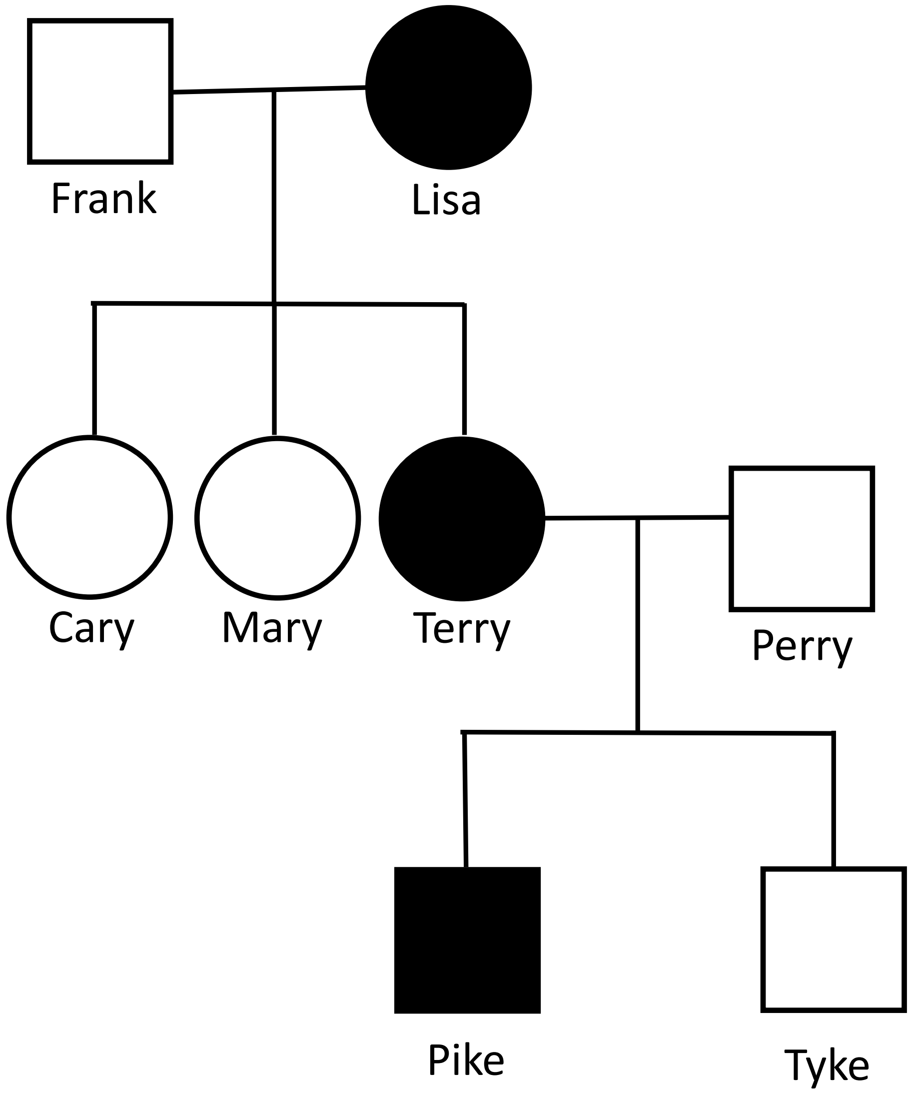


Quiz #8 – Drawing pedigrees (Lesson 16)

Lisa and Frank got married and had three girls: Cary, Mary and Terry. Lisa had **hypertrophic cardiomyopathy (HCM)**. Terry married Perry and had two boys: Pike and Tyke. It was discovered that Terry and Pike also had HCM.

Q8a. Draw the pedigree based on the info above

A8a. See graph on right

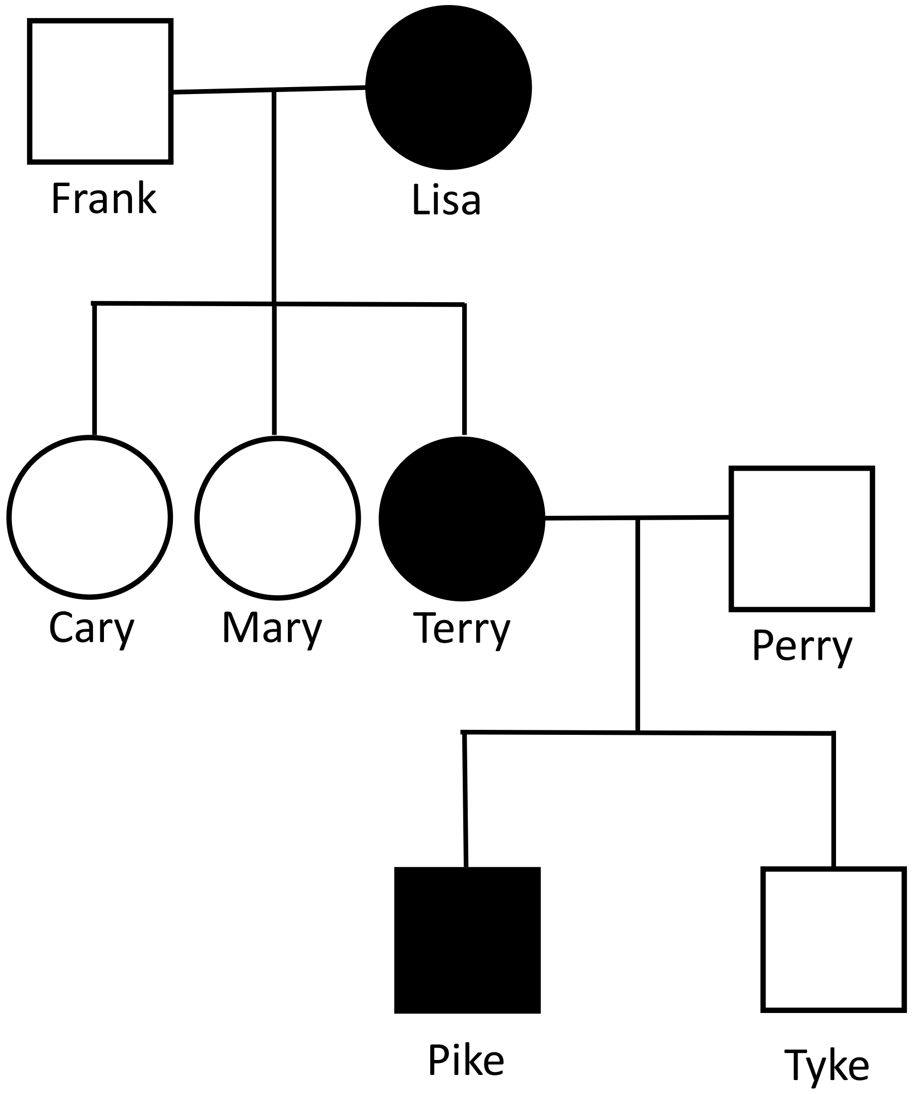


Quiz #8 – Drawing pedigrees (Lesson 16)

Lisa and Frank got married and had three girls: Cary, Mary and Terry. Lisa had **hypertrophic cardiomyopathy (HCM)**. Terry married Perry and had two boys: Pike and Tyke. It was discovered that Terry and Pike also had HCM.

Q8a. Draw the pedigree based on the info above

A8a. See graph on right



Q8b. What type of disease is HCM? Explain

A8b. **HCM is an autosomal dominant disease**

a. Both males and females affected

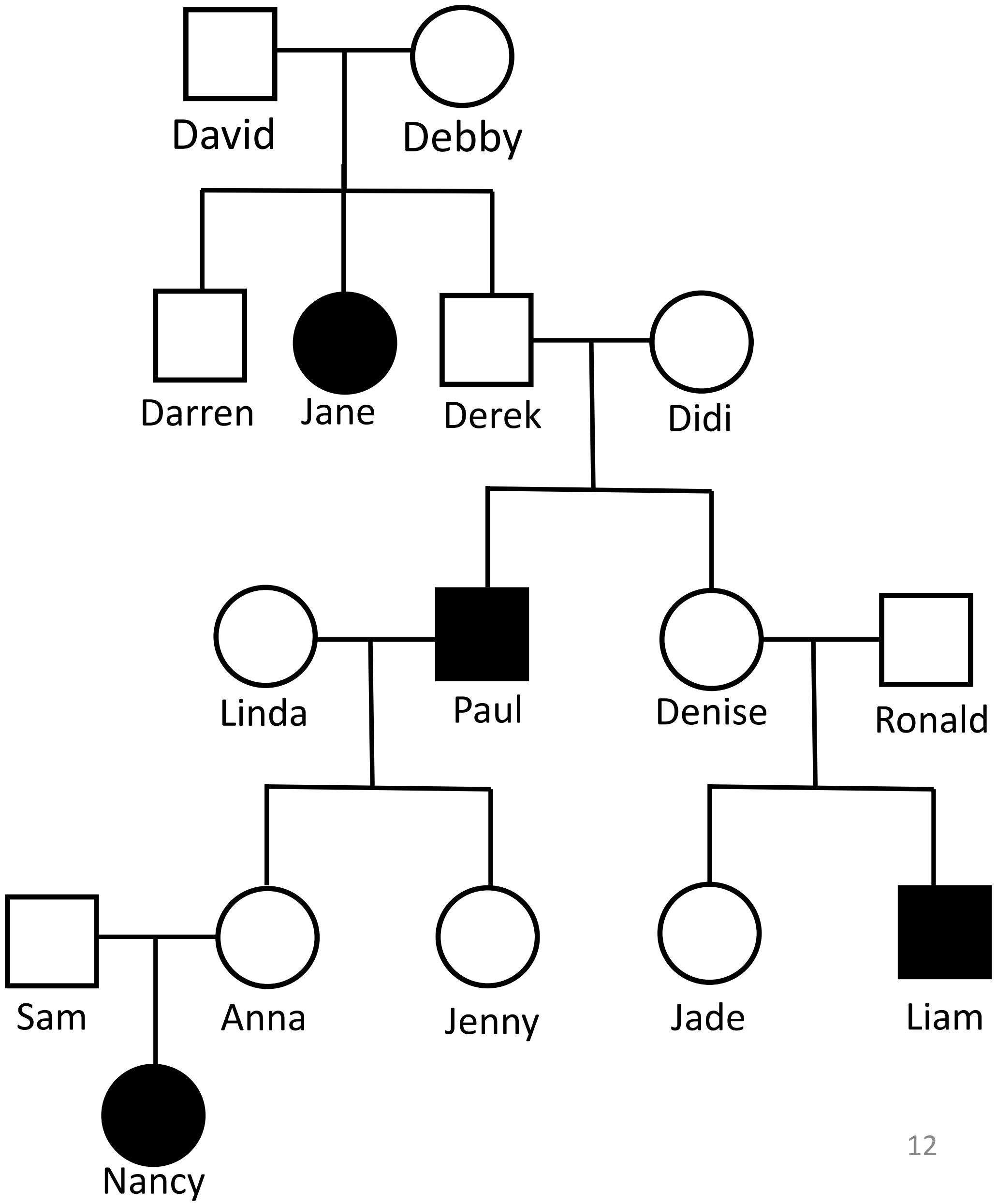
b. Every affected child has (at least) one affected parent

Quiz #9 – Drawing pedigrees (Lesson 16)

Debbie married David and had three children: Darren, Jane and Derek. Jane was found to have **cystic fibrosis (CF)**. Derek married Didi and had two children, Paul and Denise. Paul – who also had CF - married Linda, and had two daughters, Anna and Jenny. Anna married Sam and had Nancy, who had CF. Denise married Ronald, and had two kids, Jade and Liam. Liam was found to have CF.

Q9a. Draw the pedigree based on the info above

A9a. See graph on right

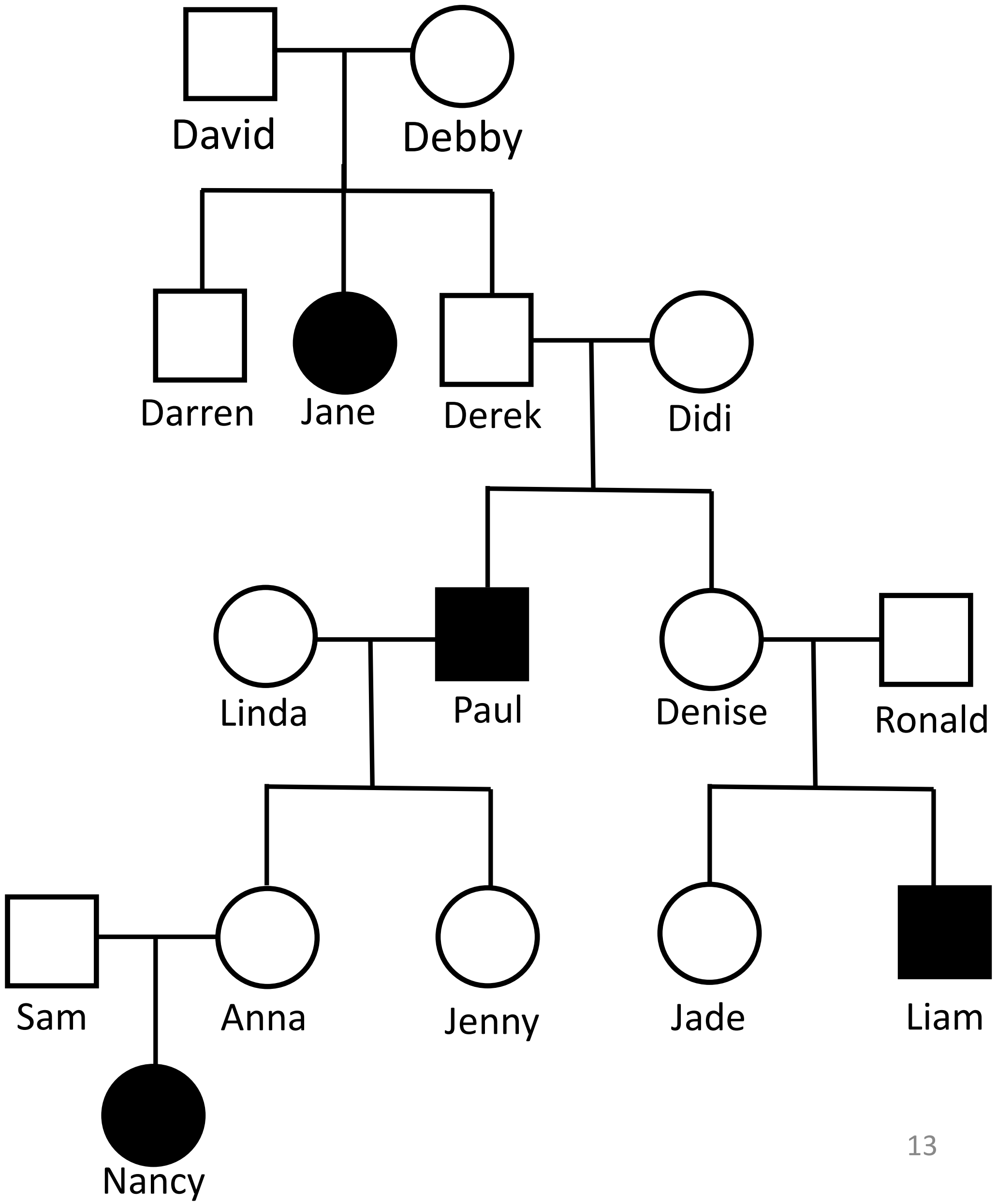


Quiz #9 – Drawing pedigrees (Lesson 16)

Debbie married David and had three children: Darren, Jane and Derek. Jane was found to have cystic fibrosis (CF). Derek married Didi and had two children, Paul and Denise. Paul – who also had CF - married Linda, and had two daughters, Anna and Jenny. Anna married Sam and had Nancy, who had CF. Denise married Ronald, and had two kids, Jade and Liam. Liam was found to have CF.

Q9a. Draw the pedigree based on the info above

A9a. See graph on right



Q9b. What type of disease is CF? Explain

A9b. CF is an autosomal recessive disease

a. Both males and females affected
b. Affected offspring from healthy parents
c. Not too many affected offspring

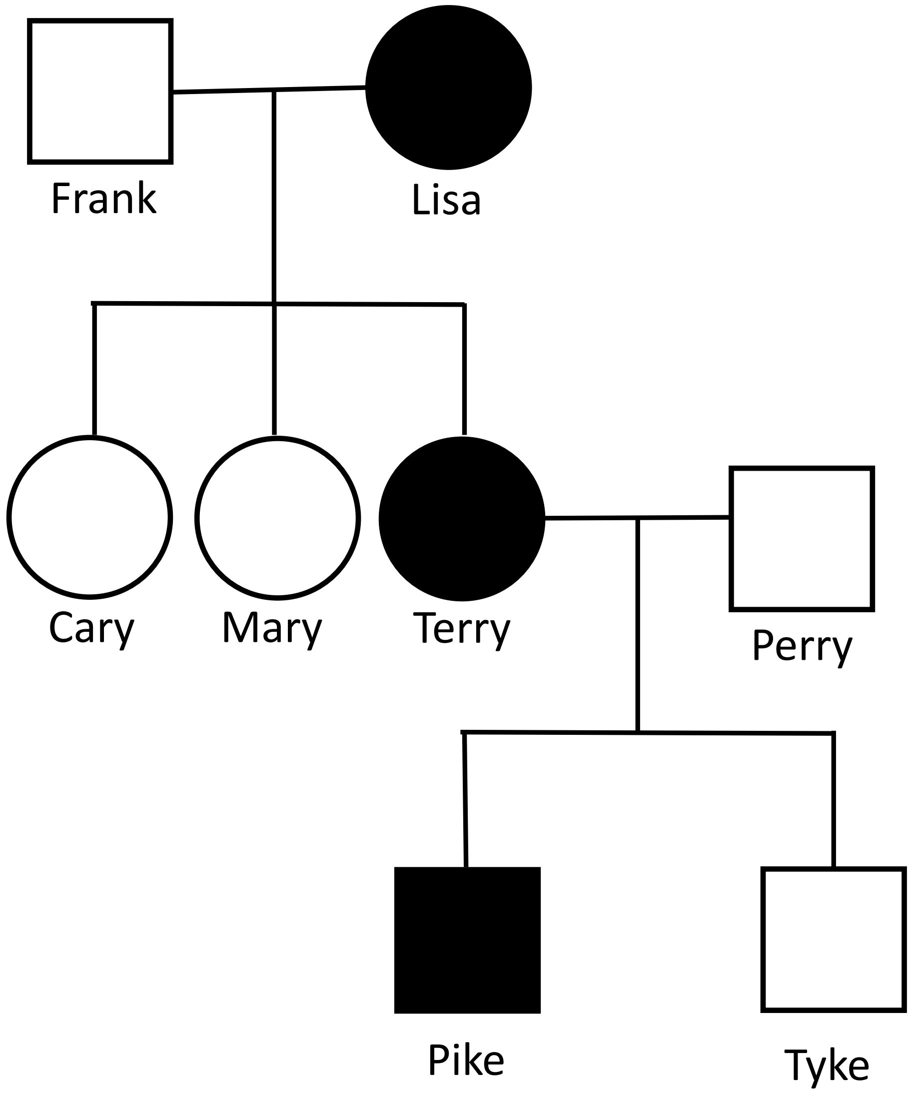
Quiz #10 – Pedigrees and genotypes (Lesson 16)

Let us reconsider Q8 and the pedigree referring to HCM (**autosomal dominant, with alleles H and h**)

Q10. Assign the genotype (G) to all individuals in the pedigree

Note:

- you might have some partially undetermined Gs
- use Punnett squares (Ps) to derive Gs



Quiz #10 – Pedigrees and genotypes (Lesson 16)

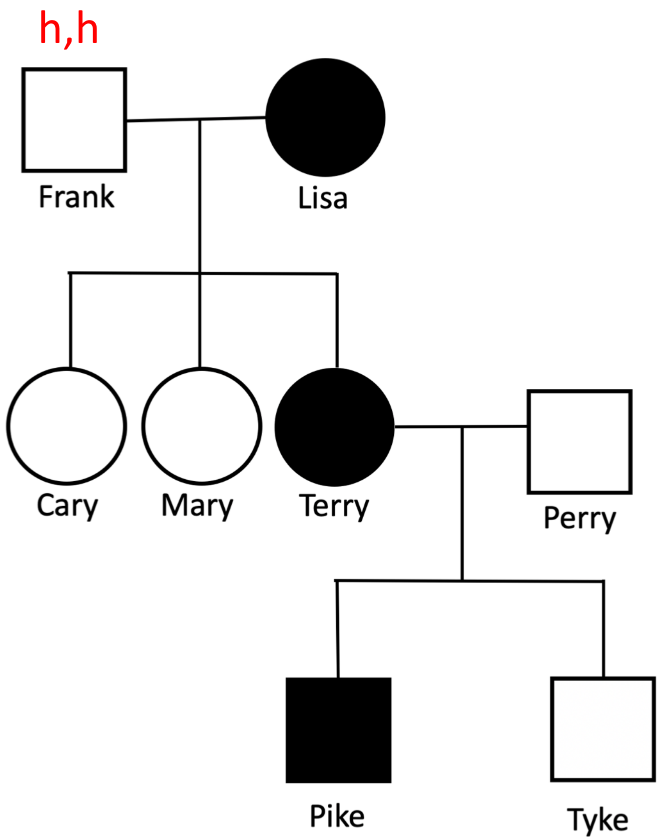
A10. Frank is healthy → **Frank has G(h,h)** (HCM AD)

Lisa has HCM → Lisa might have either G(H,H) or G(H,h) (HCM AD)

If Lisa G(H,H) → Lisa x Frank = (H,H) x (h,h)

→ all their offspring should have 100% G(H,h) and be all with HCM (HCM AD, 1 H allele is enough)

Lisa	H	H
Frank h	Hh	Hh
h	Hh	Hh



Quiz #10 – Pedigrees and genotypes (Lesson 16)

A10. Frank is healthy → Frank has G(h,h) (HCM AD)

Lisa has HCM → Lisa might have either G(H,H) or G(H,h) (HCM AD)

If Lisa G(H,H) → Lisa x Frank = (H,H) x (h,h)

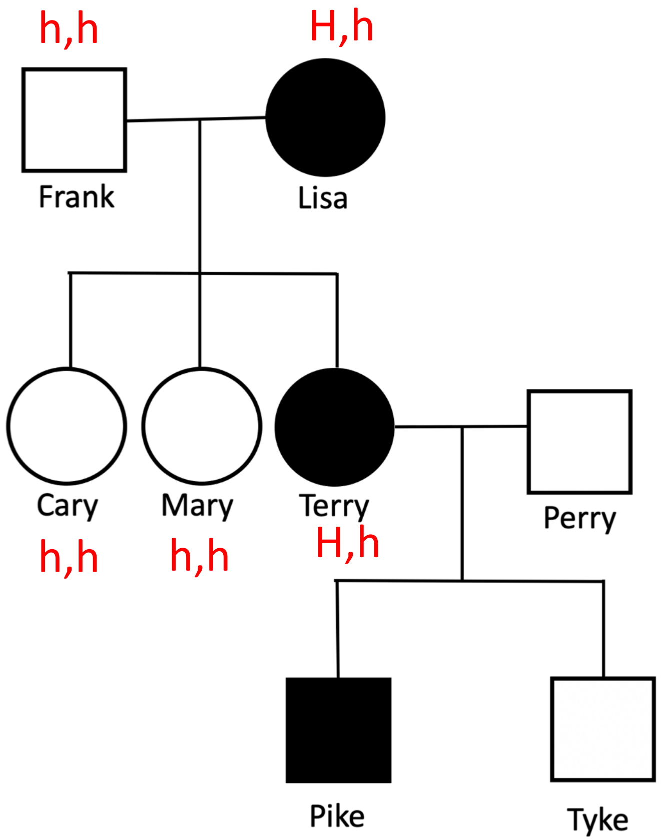
→ all their offspring should have 100% G(H,h) and be all with HCM (HCM AD, 1 H allele is enough)

But **Terry has HCM** while **Cary and Mary are healthy**

→ Lisa G must be G(H,h)

→ Lisa x Frank = (H,h) x (h,h)

→ their offspring have 50% probability to have G(H,h) and HCM and 50% probability to have G(h,h) and be healthy



Lisa	H	h
Frank h	Hh	hh
h	Hh	hh

Terry G must be (H,h) while Cary and Mary both have G(h,h)

Quiz #10 – Pedigrees and genotypes (Lesson 16)

A10. Frank is healthy → Frank has G(h,h) (HCM AD)

Lisa has HCM → Lisa might have either G(H,H) or G(H,h) (HCM AD)

If Lisa G(H,H) → Lisa x Frank = (H,H) x (h,h)

→ all their offspring should have 100% G(H,h) and be all with HCM (HCM AD, 1 H allele is enough)

But Terry has HCM while Cary and Mary are healthy

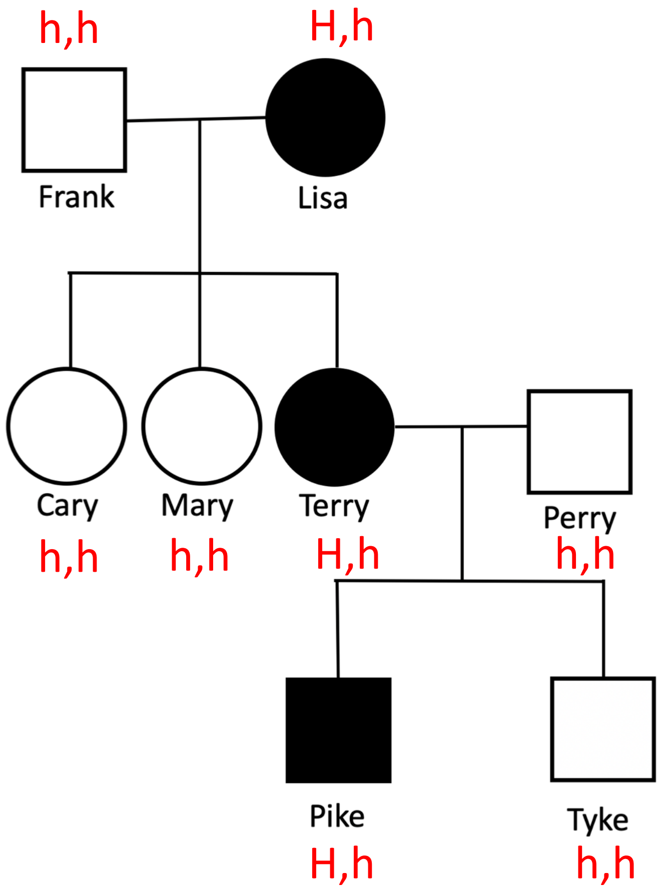
→ Lisa G must be G(H,h)

→ Lisa x Frank = (H,h) x (h,h)

→ their offspring have 50% probability to have G(H,h) and HCM and 50% probability to have G(h,h) and be healthy

Perry is healthy → **Perry has G(h,h)**

The cross Terry x Perry is identical to Frank x Lisa → **Pike has G(H,h) and Tyke has G(h,h)**



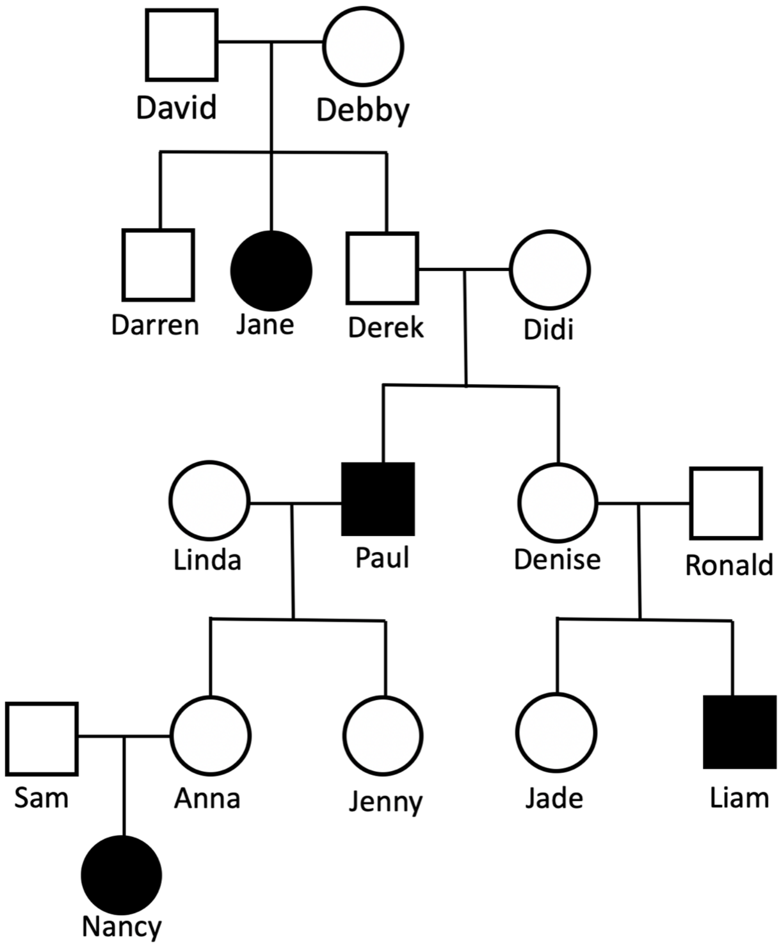
Quiz #11 – Pedigrees and genotypes (Lesson 16)

Let us reconsider Q9 and the pedigree referring to CF (**autosomal recessive, with alleles F and f**)

Q11. Assign the genotype (G) to all individuals in the pedigree

Note:

- you might have some partially undetermined Gs
- use Punnett squares (Ps) to derive Gs



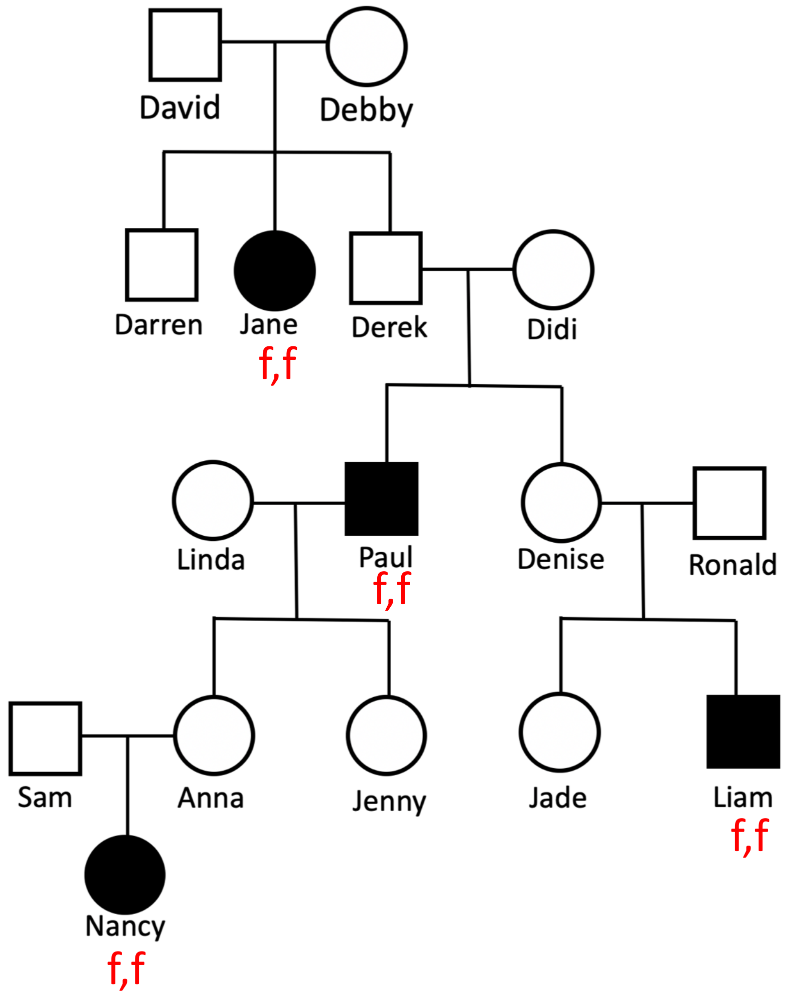
Quiz #11 – Pedigrees and genotypes (Lesson 16)

A11. CF is an autosomal recessive disease → to have CF your G MUST be (f,f) → **Jane, Paul, Liam and Nancy all have G(f,f)**

David and Debby are healthy → their G could be either (F,F) or (F,f)

If they both had G(F,F) → David x Debby = (F,F) x (F,F) → 100% offspring with G(F,F) and healthy (try PS)

→ but Jane has CF → **at least one of them must have G(F,f)**



Quiz #11 – Pedigrees and genotypes (Lesson 16)

A11. CF is an autosomal recessive disease → to have CF your G MUST be (f,f) → **Jane, Paul, Liam and Nancy all have G(f,f)**

David and Debby are healthy → their G could be either (F,F) or (F,f)

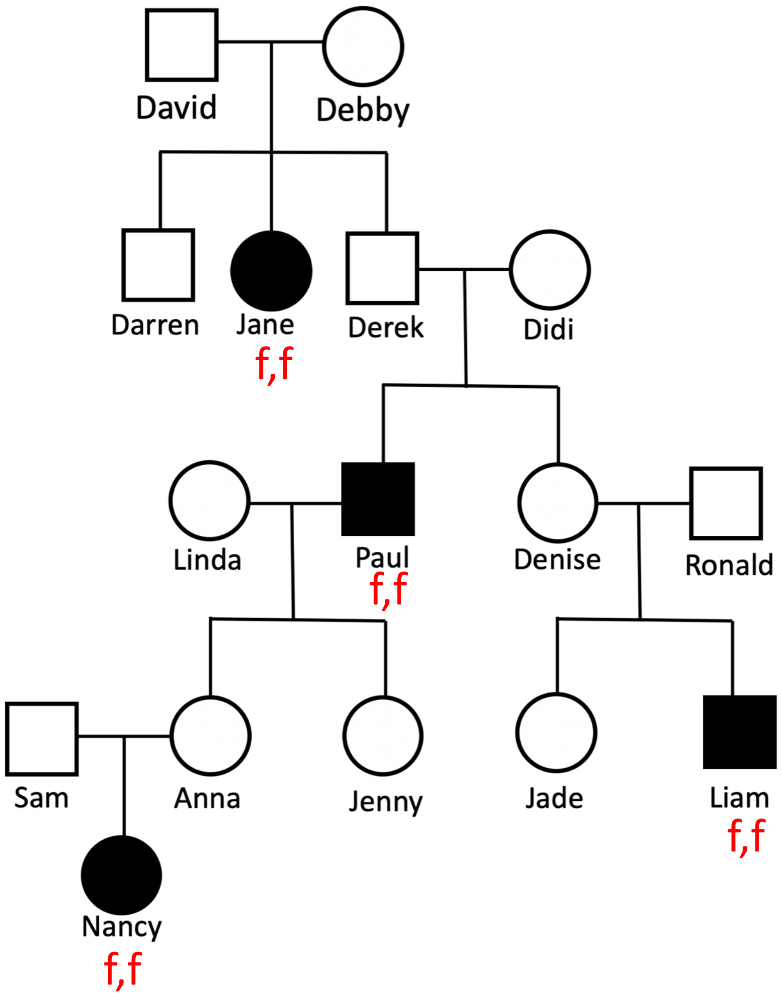
If they both had G(F,F) → David x Debby = (F,F) x (F,F) → 100% offspring with G(F,F) and healthy (try PS) → but Jane has CF → **at least one of them must have G(F,f)**

Let us suppose that David has G(F,F) and Debby had (F,f)

→ David x Debby = (F,F) x (F,f) → their offspring should be 100% healthy with 50% G(F,F) and 50% G(F,f)

But Jane has CF

	Debby	F	f
David F		FF	Ff
F		FF	Ff



Quiz #11 – Pedigrees and genotypes (Lesson 16)

A11. CF is an autosomal recessive disease → to have CF your G MUST be (f,f) → **Jane, Paul, Liam and Nancy all have G(f,f)**

David and Debby are healthy → their G could be either (F,F) or (F,f)

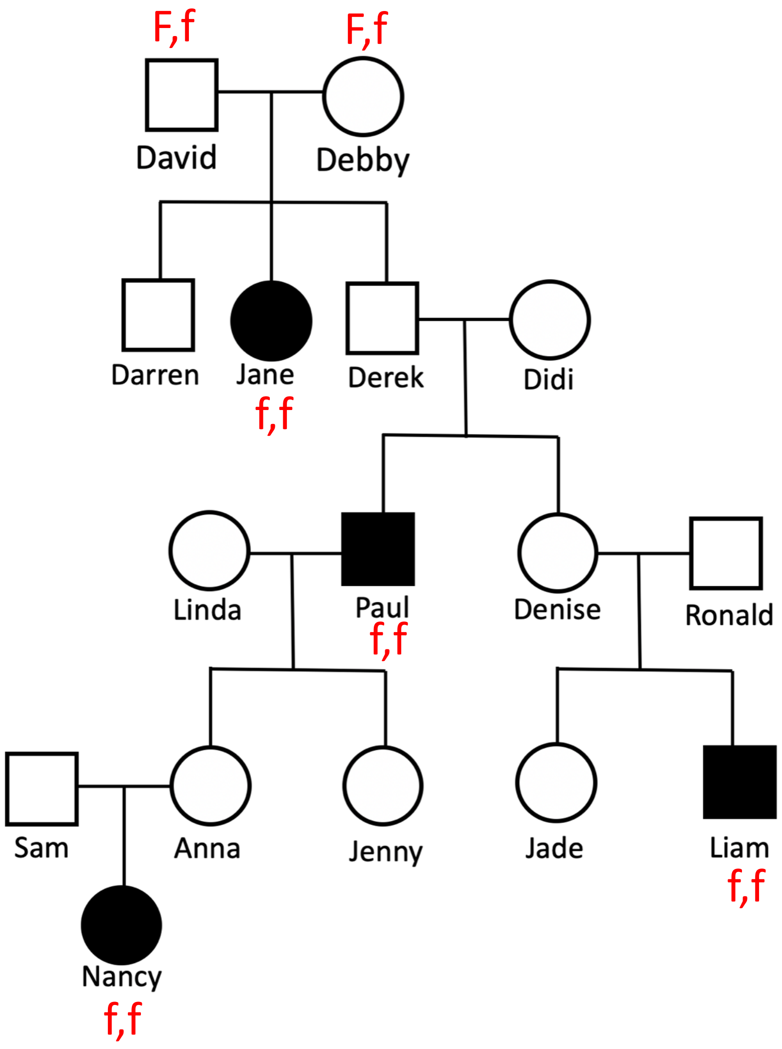
If they both had G(F,F) → David x Debby = (F,F) x (F,F) → 100% offspring with G(F,F) and healthy (try PS) → but Jane has CF → **at least one of them must have G(F,f)**

Let us suppose that David has G(F,F) and Debby had (F,f) → David x Debby = (F,F) x (F,f) → their offspring should be 100% healthy with 50% G(F,F) and 50% G(F,f)

But Jane has CF

→ Debby must have G(F,f) and David must also have G(F,f)

Debby	F	f
David F	FF	Ff
f	FF	ff



It goes the same way if we suppose Debby (F,F) and David (F,f), of course ☺

Quiz #11 – Pedigrees and genotypes (Lesson 16)

A11. CF is an autosomal recessive disease → to have CF your G MUST be (f,f) → Jane, Paul, Liam and Nancy all have G(f,f)

David and Debby are healthy → their G could be either (F,F) or (F,f)

If they both had G(F,F) → David x Debby = (F,F) x (F,F) → 100% offspring with G(F,F) and healthy (try PS) → but Jane has CF → at least one of them must have G(F,f)

Let us suppose that David has G(F,F) and Debby had (F,f) → David x Debby = (F,F) x (F,f) → their offspring should be 100% healthy with 50% G(F,F) and 50% G(F,f) → also David must have G(F,f)

It goes the same way if we suppose Debby (F,F) and David (F,f), of course ☺

Darren is healthy but his G could be either (F,F) or (F,f)

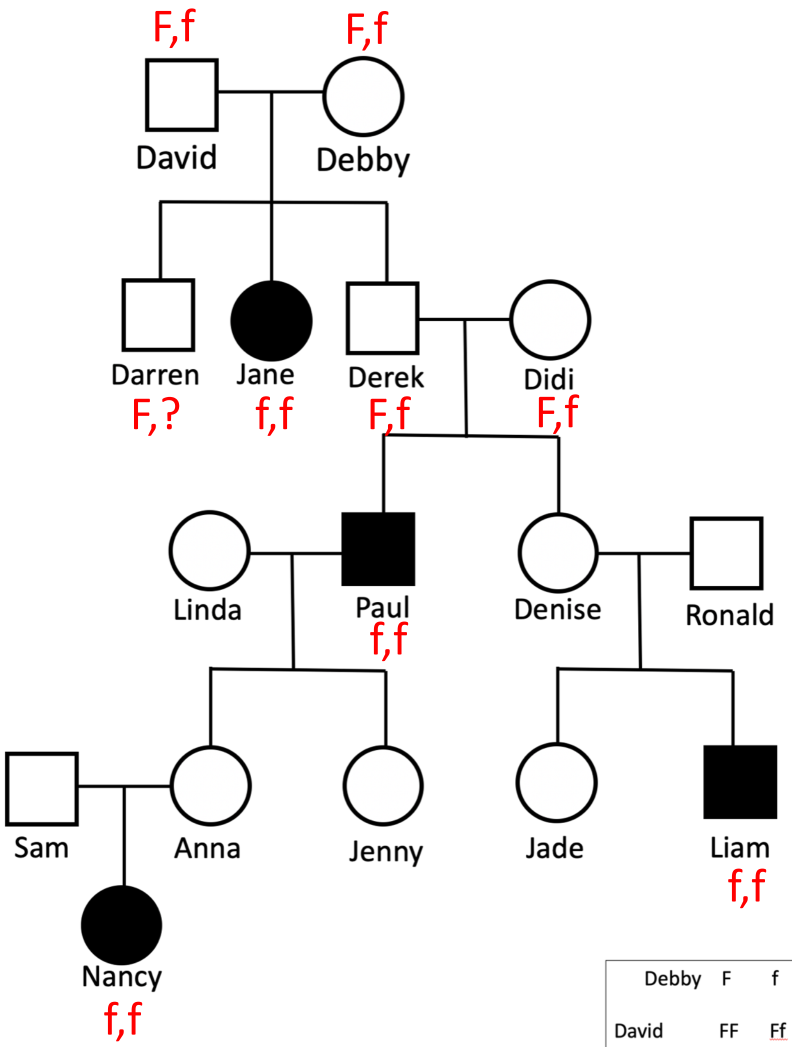
→ **Darren G is (F,?)** (we do not have further info)

For Derek (healthy), we know that his G could be either (F,F) or (F,f). For Didi (healthy), she also can have G(F,F) or G(F,f).

The cross Derek (F,F) x Didi (F,F) MUST be ruled out as they have 1 affected kid (Paul) (FFxFF → 100% FF, healthy)

The cross Derek (F,F) x Didi (F,f) (or vice versa) MUST also be ruled out as they have 1 affected kid (Paul) (FFxFf → 50% FF + 50% Ff, all healthy)

The genotype of both Derek and Didi MUST be (F,f) (as for David and Debby)



Debby	F	f
David F	FF	Ff
f	FF	ff

Quiz #11 – Pedigrees and genotypes (Lesson 16)

A11. CF is an autosomal recessive disease → to have CF your G MUST be (f,f) → Jane, Paul, Liam and Nancy all have G(f,f)

David and Debby are healthy → their G could be either (F,F) or (F,f)

If they both had G(F,F) → David x Debby = (F,F) x (F,F) → 100% offspring with G(F,F) and healthy (try PS) → but Jane has CF → at least one of them must have G(F,f)

Let us suppose that David has G(F,F) and Debby had (F,f) → David x Debby = (F,F) x (F,f) → their offspring should be 100% healthy with 50% G(F,F) and 50% G(F,f) → also David must have G(F,f) (draw the relevant Ps to convince yourself)

It goes the same way if we suppose Debby (F,F) and David (F,f), of course ☺

Darren is healthy but his G could be either (F,F) or (F,f) → **Darren G is (F,?)** (we do not have further info)

For Derek (healthy), we know that his G could be either (F,F) or (F,f). For Didi (healthy), she also can have G(F,F) or G(F,f).

The cross Derek (F,F) x Didi (F,F) MUST be ruled out as they have 1 affected kid (Paul) (FFxFF → 100% FF, healthy)

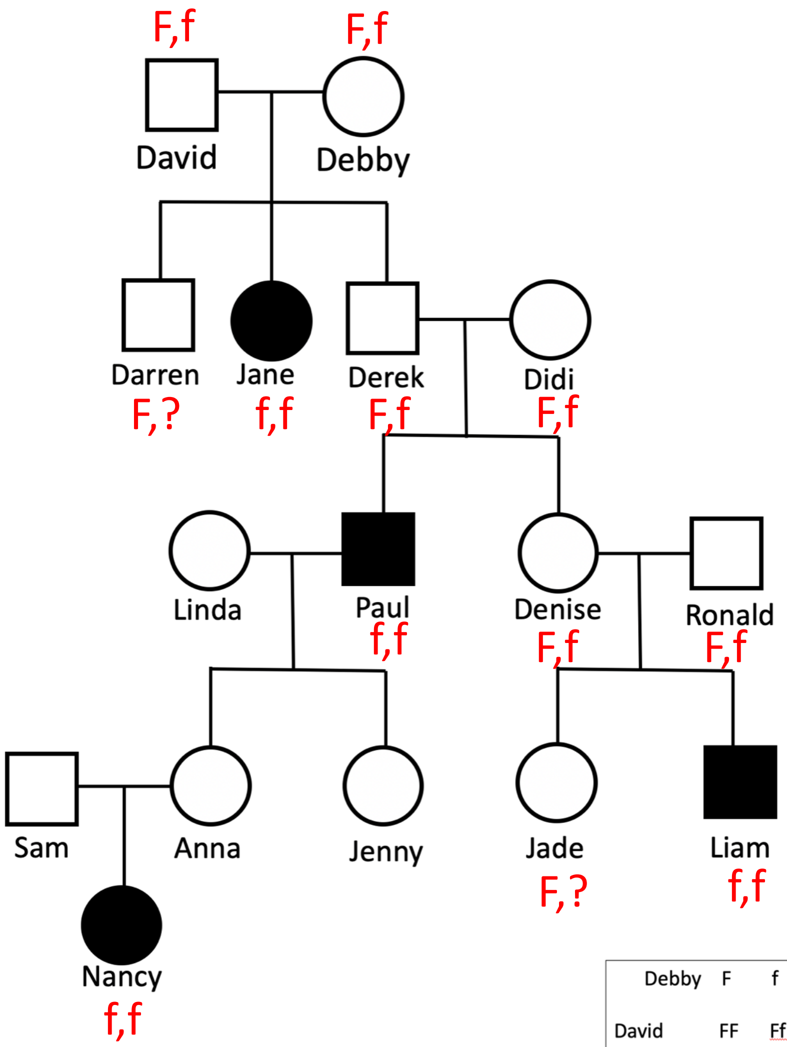
The cross Derek (F,F) x Didi (F,f) (or vice versa) MUST also be ruled out as they have 1 affected kid (Paul) (FFxFf → 50% FF + 50% Ff, all healthy)

The genotype of both Derek and Didi MUST be (F,f) (as for David and Debby)

Denise G could be either (F,F) or (F,f). Ronald is healthy → his G can also be either (F,F) or (F,f) → but they have Liam with CF → same situation as for Derek and Didi (or David and Debby)

→ **both Denise and Ronald G MUST be (F,f)**

Jade (healthy) G could either be (F,F) or (F,f) → Jade G = (F,?) (as for Darren)



	Debby	F	f
David	F	FF	Ff
	f	FF	ff

Quiz #11 – Pedigrees and genotypes (Lesson 16)

A11. CF is an autosomal recessive disease → to have CF your G MUST be (f,f) → Jane, Paul, Liam and Nancy all have G(f,f)

David and Debby are healthy → their G could be either (F,F) or (F,f)

If they both had G(F,F) → David x Debby = (F,F) x (F,F) → 100% offspring with G(F,F) and healthy (try PS) → but Jane has CF → at least one of them must have G(F,f)

Let us suppose that David has G(F,F) and Debby had (F,f) → David x Debby = (F,F) x (F,f) → their offspring should be 100% healthy with 50% G(F,F) and 50% G(F,f) → also David must have G(F,f) (draw the relevant Ps to convince yourself)

It goes the same way if we suppose Debby (F,F) and David (F,f), of course ☺

Darren is healthy but his G could be either (F,F) or (F,f) → **Darren G is (F,?)** (we do not have further info)

For Derek (healthy), we know that his G could be either (F,F) or (F,f). For Didi (healthy), she also can have G(F,F) or G(F,f).

The cross Derek (F,F) x Didi (F,F) MUST be ruled out as they have 1 affected kid (Paul) (FFxFF → 100% FF, healthy)

The cross Derek (F,F) x Didi (F,f) (or vice versa) MUST also be ruled out as they have 1 affected kid (Paul) (FFxFf → 50% FF + 50% Ff, all healthy)

The genotype of both Derek and Didi MUST be (F,f) (as for David and Debby)

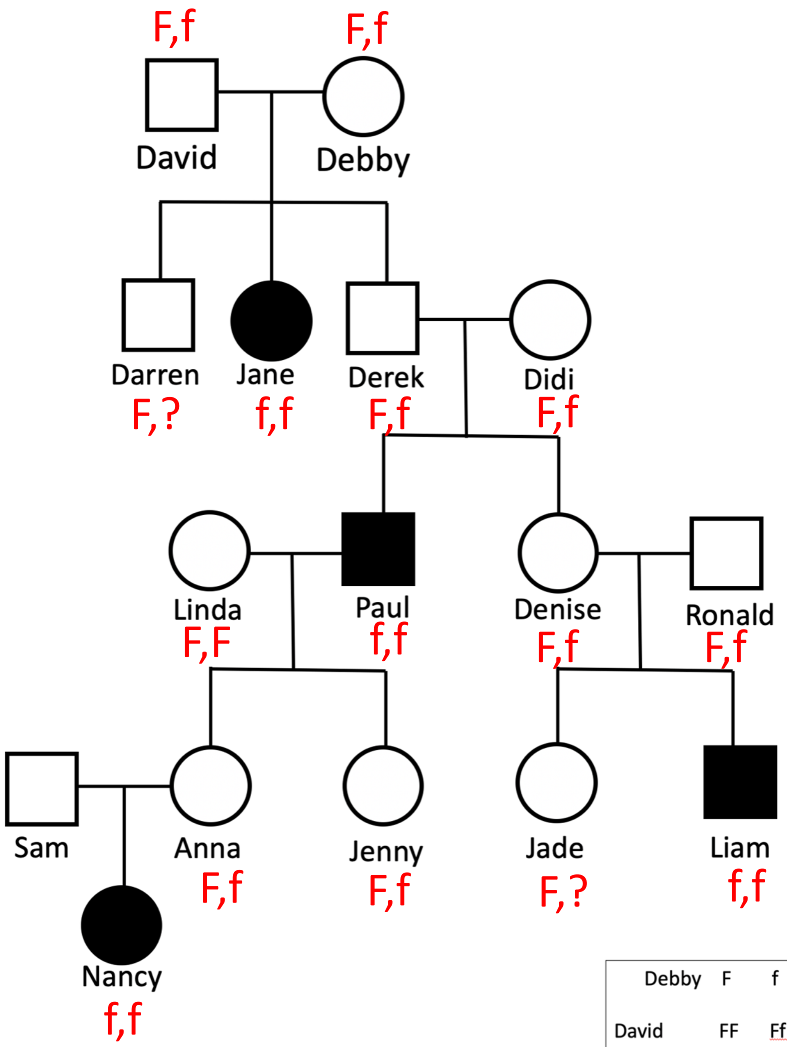
Denise G could be either (F,F) or (F,f). Ronald is healthy → his G can also be either (F,F) or (F,f) → but they have Liam with CF → same situation as for Derek and Didi (or David and Debby) → **both Denise and Ronald G MUST be (F,f)**

Jade (healthy) G could either be (F,F) or (F,f) → Jade G = (F,?) (as for Darren)

Linda genotype must be (F,F) since Paul has CF (G(f,f)) and they have 100% healthy offspring (Anna and Jenny) both with G(F,f)

Linda	F	F
Paul f	Ff	Ff
f	Ff	Ff

Linda	F	f
Paul f	Ff	ff
f	Ff	ff



Debby	F	f
David F	FF	Ff
f	FF	ff

Quiz #11 – Pedigrees and genotypes (Lesson 16)

A11. CF is an autosomal recessive disease → to have CF your G MUST be (f,f) → Jane, Paul, Liam and Nancy all have G(f,f)

David and Debby are healthy → their G could be either (F,F) or (F,f)

If they both had G(F,F) → David x Debby = (F,F) x (F,F) → 100% offspring with G(F,F) and healthy (try PS) → but Jane has CF → at least one of them must have G(F,f)

Let us suppose that David has G(F,F) and Debby had (F,f) → David x Debby = (F,F) x (F,f) → their offspring should be 100% healthy with 50% G(F,F) and 50% G(F,f) → also David must have G(F,f) (draw the relevant Ps to convince yourself)

It goes the same way if we suppose Debby (F,F) and David (F,f), of course ☺

Darren is healthy but his G could be either (F,F) or (F,f) → **Darren G is (F,?)** (we do not have further info)

For Derek (healthy), we know that his G could be either (F,F) or (F,f). For Didi (healthy), she also can have G(F,F) or G(F,f).

The cross Derek (F,F) x Didi (F,F) MUST be ruled out as they have 1 affected kid (Paul) (FFxFF → 100% FF, healthy)

The cross Derek (F,F) x Didi (F,f) (or vice versa) MUST also be ruled out as they have 1 affected kid (Paul) (FFxFf → 50% FF + 50% Ff, all healthy)

The genotype of both Derek and Didi MUST be (F,f) (as for David and Debby)

Denise G could be either (F,F) or (F,f). Ronald is healthy → his G can also be either (F,F) or (F,f) → but they have Liam with CF → same situation as for Derek and Didi (or David and Debby) → **both Denise and Ronald G MUST be (F,f)**

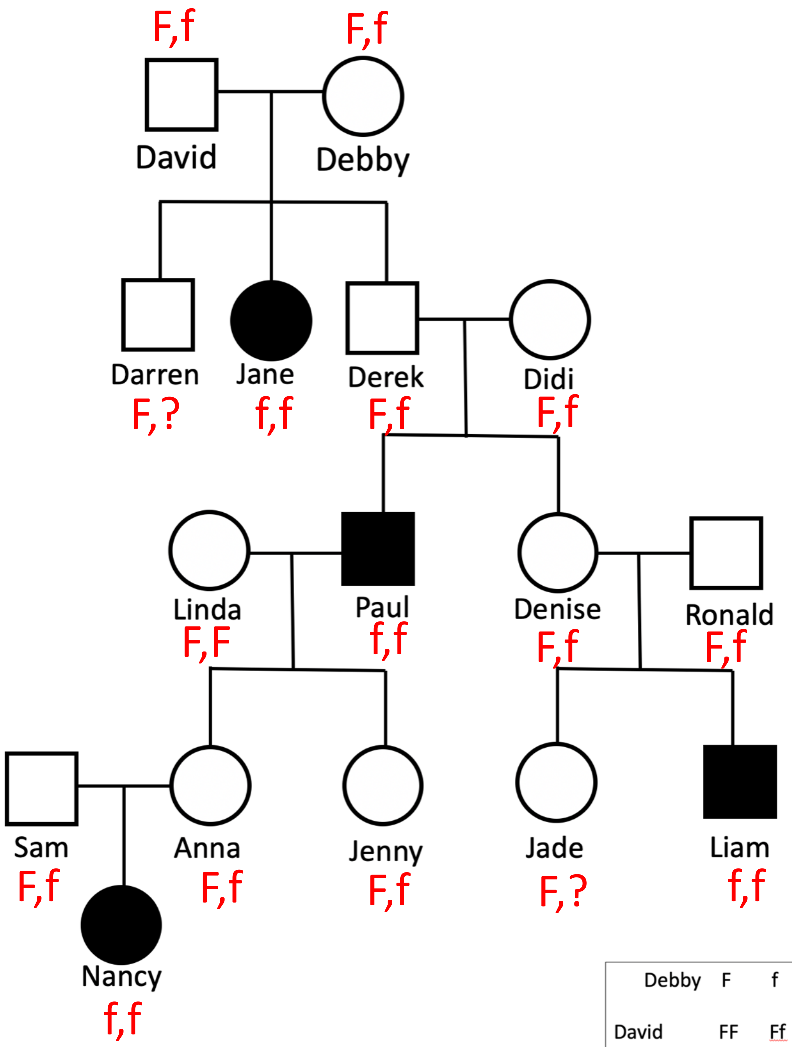
Jade (healthy) G could either be (F,F) or (F,f) → Jade G = (F,?) (as for Darren)

Linda genotype must be (F,F) since Paul has CF (G(f,f)) and they have 100% healthy offspring (Anna and Jenny) both with G(F,f)

Sam is healthy → he can have either G(F,F) or G(F,f)

Sam genotype G(F,F) can be ruled out it has an affected daughter (Nancy, G(f,f)) and Anna G is (F,f) → the cross F,F x F,f will give 100% healthy offspring with 50% G(F,F) and 50% G(F,f)

→ Sam genotype must be (F,f)



	Debby F	f
David F	FF	Ff
f	FF	ff

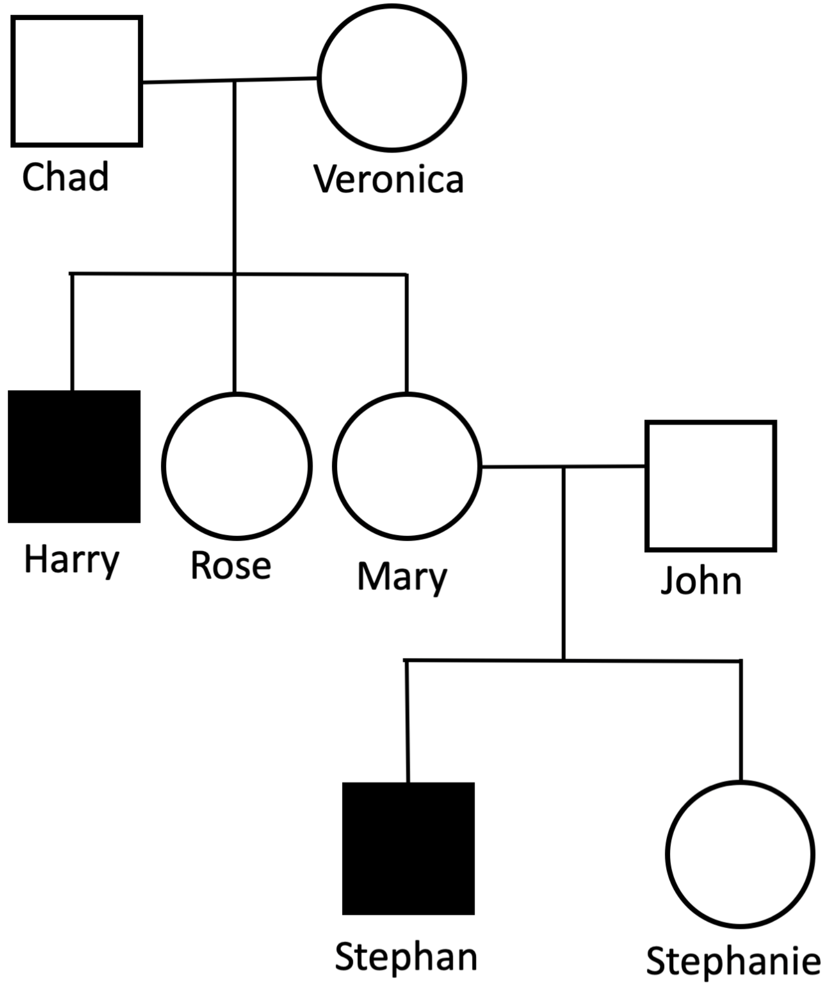
Quiz #12 – Pedigrees and genotypes (Lesson 16)

Let us reconsider Q7 and the pedigree referring to DMD (**X-linked recessive, X^d, X^D**)

Q12. Assign the genotype (G) to all individuals in the pedigree

Note:

- you might have some partially undetermined Gs
- use Punnett squares (Ps) to derive Gs



Quiz #12 – Pedigrees and genotypes (Lesson 16)

A12. Chad is healthy → **Chad's G MUST be (X^D,Y)**

a. X-linked recessive is transmitted from mother

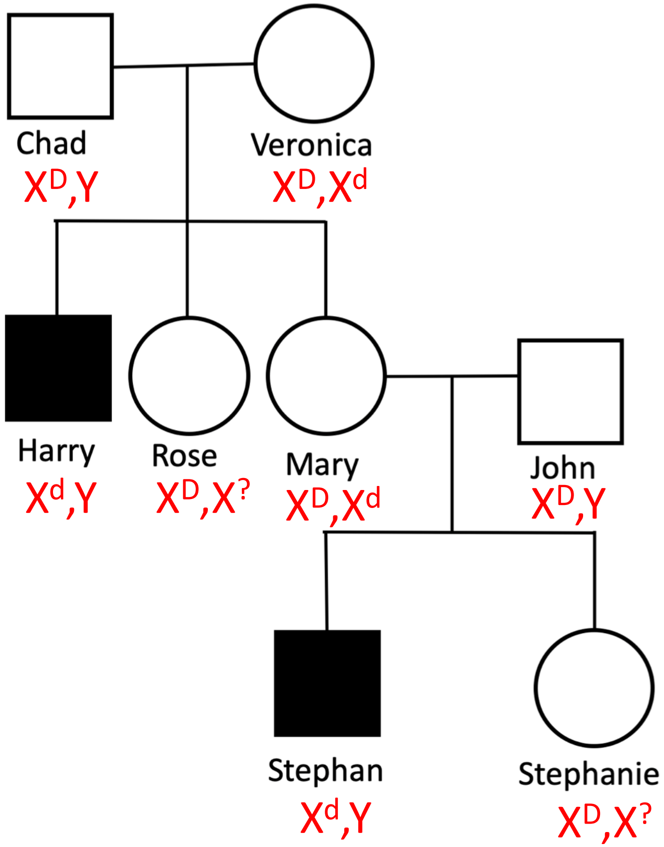
b. affect mostly man

c. Chad and Veronica has one affected son → **Veronica's G must be (X^D,X^d)**

The cross Chad x Veronica is (X^D,Y) x (X^D,X^d) -------->

Thus Harry's G must be **(X^d,Y) since he has DMD**

Chad	X^D	Y
Veronica X^D	X^DX^D	X^DY
X^d	X^DX^d	X^dY



Rose is healthy → Rose's G could either be (X^D,X^D) or (X^D,X^d) → we do not have enough info → **Rose's G is (X^D,$X^?$)**

Mary is healthy → Mary's G could either be (X^D,X^D) or (X^D,X^d)

John is healthy → John's G MUST be (X^D,Y)

Stephan has DMD → Stephan's G must be (X^d,Y)

Mary's G MUST be (X^D,X^d)

Stephanie is healthy → Stephanie's G could either be (X^D,X^D) or (X^D,X^d) → we do not have enough info → **Stephanie's G is (X^D,$X^?$)**