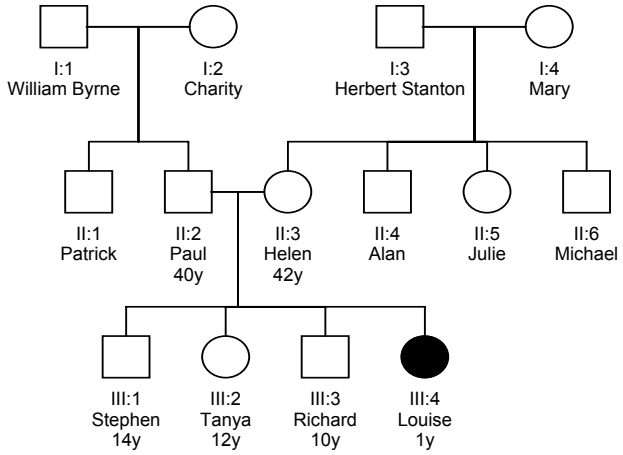
5. What is the cause of Down syndrome in Louise?

Louise (III:4) has Down syndrome.



Q1 What is the most likely chromosome cause of the Down syndrome in Louise?

Studies have shown that in Down syndrome
95% cases caused by *trisomy 21*
4% have the extra copy of chromosome 21 because of a *Robertsonian translocation*
1% have *mosaicism* with normal and trisomy 21 cell lines (and usually have much milder features because of the presence of the normal cells). Mosaicism occurs because of mitotic nondisjunction during growth and development following fusion of gametes which both have normal chromosome complements.

We need to look for clues as to whether the Down syndrome is likely to be due to trisomy 21 or an inherited Robertsonian translocation.

Note that only 1 person is affected with Down syndrome, there are no pregnancy losses (which might suggest a Robertsonian translocation) and Louise's mother, Helen was 41 when Louise was born.

These make it more likely that Louise has regular **trisomy 21** as the cause of her Down syndrome.

Q2 Write down the karyotype you would expect Louise to have.

47,XX,+21

This is the karyotype nomenclature for a female with 3 copies of chromosome number 21 – regular trisomy 21. The total chromosome complement is 47 (the normal 46 plus the additional 21); XX signifies a normal female sex chromosome complement, and +21 shows that the additional chromosome is a whole chromosome number 21.

Q3 What is the piece of information from the pedigree which led you to this conclusion?

Louise's mother, Helen was 41 when Louise was born.
Maternal meiotic **non disjunction** leading to trisomy is associated with increasing maternal age.

Q4 Louise's mother wonders about having another baby so that Louise has a brother or sister nearer her age. How likely is that another baby would also have Down syndrome?

1 in 100 – this figure is obtained from empiric studies: the observed recurrence rate in families with one child with Down syndrome.