

Solutions

2.1

- X-linked (both recessive and dominant).

2.2

- $1/4$ ✓.

2.3

- All daughters affected (100 %), no sons affected ✓.

3.1

- Prior risk $1/2$ (affected parent is Aa).
- If say 40 % of carriers have onset by 28: $P(\text{unaffected at 28} \mid \text{carrier}) = 0.6 \rightarrow \text{posterior} = (1/2 \times 0.6) / (1/2 \times 0.6 + 1/2 \times 1) = 0.375$ – evidence, not proof.
- Predictive testing resolves it to ~ 0 or ~ 1 (with counselling on insurability and psychological burden).

3.2

- ~ 80 % de-novo (FGFR3, paternal age effect).
- Sib recurrence near population risk; the child's own children at $1/2$.
- Germline mosaicism in a parent gives a small recurrence above population (~ 1 %).