

Solutions

2.1

- Karyotype (microscopy of metaphase spreads).

2.2

- Chromosomal microarray.

2.3

- Population risk for that syndrome.

3.1

- Each sibling and each parent: $1/2$ a priori.
- Cascade testing: relatives tested for THE familial variant → positives enter screening, negatives return to population risk – uncertainty becomes binary.
- Considerations: autonomy (the 19-year-old decides); implications for insurability and psychological burden; timing.

3.2

- Defer the adult-onset result until the child can consent.
- Consent before testing sets the return-of-results menu (opt in/out); after the fact, the choice is already gone.
- If childhood-actionable (treatable/preventable now), return it - the child's health benefit overrides deferral.