Constitutional marking summary

Analysis / karyotype: max 2 marks

- Correct: 2 marks
- Incorrect: 0 marks
- Partial: 1 or 1.5 marks depending on significance, e.g. one half of unique translocation, or minor clone missed. Incomplete FISH investigation essential for analysis

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Clerical Accuracy: max 2 marks

- ISCN
  - Correct: 1 mark
  - Incorrect: 0 mark
  - Minor error or multiple clerical
  - One clerical or non-significant error: comment only
- Written description of karyotype including cell numbers and normal cells.
  - Correct: 1 mark
  - Incorrect: 0 mark

Interpretation: max 2 marks

½ mark deducted if any of these that are applicable are missing

1. Correlation with referral reason/clinical indication
2. Report states that the karyotype is indicative of/consistent with syndrome, if applicable.
3. Follow up as necessary for complete interpretation of the case, e.g. parental bloods, if applicable.
4. Give limitations of test result, if applicable, e.g. normal result on a PWS.
5. Prognostic indication of risk, e.g. PND future pregnancies, risk of miscarriage if known. If risk calculation is done by clinical geneticist then the laboratory should submit the clinical geneticist's letter with its cytogenetic report. Risk assessment appropriate for the situation (do not have to be specific figures).
6. Onward referral to clinical geneticist as applicable.