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CHROMOSOME ANALYSIS OF CONGENITAL DISORDERS BLOOD

Synonym(s): KARYOTYPE CONGENITAL DISORDERS BLOOD

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Lab Section Category	**Cytogenetics – Postnatal** Click here to find out more about the **write-up (/clinical-departments-centers/pathology/pathology-handbook/lab-discipline-special-instructions/pages/cytogenetics.aspx)**.
Indications	- Previous family history of chromosome abnormality - Children with multiple congenital abnormalities - Recurrent abortions or infertility - Unexplained mental retardation and/dysmorphology
Specimen Required	Peripheral blood (3 - 5ml) in sodium/lithium heparin. Invert gently several times to mix blood and vial contents to prevent clotting (clotted blood will not work). Do not use EDTA or plain tubes.
Method	72-hour synchronised suspension cultures.
Test Result	Normal or Abnormal. If Abnormal, the karyotypic abnormality is described in accordance to the International System for Human Cytogenomic Nomenclature (ISCN, 2016).
Turnaround Time	10 ~ 28 days
Day(s) Test Set up	Monday - Saturday (office hours)

Remarks	Breakage studies for Fragile X and Fanconi Anaemia are not performed by this lab. Indicate clearly the clinical diagnosis on the cytogenetics request form or CPOE. Do not despatch a specimen near a weekend or a public holiday to avoid unnecessary delays.

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Change History Notes

- **08 Dec 2015 08:21 AM**
  Updated Turnaround Time: 10 ~ 28 days

- **06 Jul 2017 10:40 AM**
  - Updated the test result for ISCN, 2013 to ISCN, 2016