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CHROMOSOME ANALYSIS WITH FLUORESCENCE IN SITU HYBRIDIZATION TEST FOR MICRODELETION SYNDROMES

Synonym(s): MICRODELETION FISH ADD-ON

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Lab Section Category	**Cytogenetics - PP FISH** Click here to find out more about the **write-up (/clinical-departments-centers/pathology/pathology-handbook/lab-discipline-special-instructions/pages/cytogenetics.aspx)**.
Indications	This test is useful for patients suspected of Prader-Willi (PWS) / Angelman (AS), Di-George (DGS) or William (WS) syndromes.
Specimen Required	Peripheral Blood/Amniotic Fluid/Chorionic Villi
Method	Long-term culture to obtain metaphases. Probes used for metaphase and interphase FISH (PWS/AS, DGS or WS region DNA probes) are labelled with fluorophores and analysed under fluorescence microscopy.
Test Result	Deleted or Not Deleted or Duplicated. Nomenclature given is in accordance to the International System for Human Cytogenomic Nomenclature (ISCN, 2016). Concurrent interphase FISH will detect duplication of that gene of interest.
Reference Interval/Value	Individuals with a microdeletion will show two copies of the internal control signals but only one copy of the locus-specific signal for that region of interest.
Turnaround Time	8 ~ 12 days
Day(s) Test Set up	Monday – Saturday (office hours)
Remarks	This FISH test is always used in conjunction with conventional cytogenetic studies. Contact the Cytogenetics Laboratory before sending a specimen.

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Change History Notes

- **08 Dec 2015 08:28 AM**
  Updated Turnaround Time: 8 ~ 12 days

- **06 Jul 2017 10:47 AM**
  - Updated the test result for ISCN, 2013 to ISCN, 2016