INTRODUCTION

The Cytogenetics Laboratory offers a comprehensive array of chromosome investigations for cancers, constitutional abnormalities, and prenatal and postnatal diagnosis. Analyses are performed with the aid of dedicated computerised karyotyping workstations. All reports include a description of the cytogenetic findings in accordance to the International System for Human Cytogenetic Nomenclature (2005), and an interpretation of the results. A hardcopy of the karyotype may be obtained on request. The Laboratory also offers a wide range of Fluorescence In Situ Hybridization (FISH) tests including rapid aneuploidy screening, microdeletion syndromes, and a host of specific malignancy tests for gene fusion / breakapart such as BCR/ABL1, PML/RARA and MLL. The FISH section also offers HER2 gene (ERBB2) amplification detection assay on breast cancer tissue sections. As many of these FISH tests are specific, it is important to check in advance with the laboratory on probe availability.

SPECIAL INSTRUCTIONS ON SAMPLE COLLECTION AND HANDLING

1. Please contact the Cytogenetics Laboratory for an appointment before despatching the specimen over for processing (Tel: 63214678/4650).
2. All samples should be collected under sterile conditions. For requests requiring tissue culturing, send only fresh samples to the Laboratory, and as soon as possible.
3. Do not freeze the specimen. A cool pack may be used to ensure that the samples are not exposed to temperatures in excess of 30°C.
4. Please ensure that all samples arrive at the laboratory at least two (2) hours before closing time.
   - Monday – Friday 8.00 am – 5.30 pm
   - Saturday 8.00 am – 1.00 pm
5. Provide all necessary information on the Request For Cytogenetics Investigation form, including:
   - Patient’s name
   - NRIC number
   - Sex
   - Age
   - Date of birth
   - Presumptive diagnosis
   - Relevant clinical information
   - Specimen type
   - Name of referring doctor
   - Clinic
   - Telephone/fax number
   - Date and time of specimen collection

   For prenatal specimens, also include the following:
   - Gestational age
   - LMP
   - EDD

   For haematological malignancies, also include the following:
   - Total white count

6. Follow standard precaution guidelines. Treat all samples as potentially hazardous.
TEST LISTING

CHROMOSOME ANALYSIS FOR PREGNATAL DIAGNOSIS

Common indications:
- Advanced maternal age (≥ 35 years at EDD)
- Abnormal ultrasound findings
- Positive maternal serum screening
- Previous history of chromosome abnormality
- Family history of Down syndrome
- Parental anxiety

Special instructions:
- If a concurrent FISH test is required, please indicate clearly on the request form. Do not despatch a specimen near a weekend or a public holiday so as to avoid unnecessary delays.

AMNIOTIC FLUID

Sample required:
- Amniotic fluid (15 – 25 mL) from between the 15th and 16th week of gestation. Discard the first 2 mL of the aspirate. Use a sterile 25 or 30 mL syringe. Draw and transfer the specimen into two sterile screw-capped 15 mL V-shaped tubes. Bloody specimens are undesirable. If growth is inadequate, you will be notified within 10 days of receipt.

Method:
- In situ coverslip technique

Test results:
- Normal or Abnormal. If Abnormal, the cytogenetic lesions are listed in accordance to the International System for Human Cytogenetic Nomenclature (ISCN, 2005).

Turnaround time:
- 8 days (mean)

Day(s) test set up:
- Monday – Saturday (office hours)

CHORIONIC VILLI

Specimen required:
- Chorionic villi (10 – 30 mg) by transabdominal or transcervical sampling in a sterile tube containing transport medium supplied by the Cytogenetics Laboratory

Method:
- In situ coverslip technique

Test results:
- Normal or Abnormal. If Abnormal, the cytogenetic lesions are listed in accordance to the International System for Human Cytogenetic Nomenclature (ISCN, 2005).

Turnaround time:
- 9 days (mean)

Day(s) test set up:
- Monday – Saturday (office hours)
FETAL CORD BLOOD/NEONATE BLOOD

**Specimen required**: Fetal cord blood/neonate blood (2 – 3 mL) in sodium/lithium. Invert gently several times to mix the blood and vial contents (clotted blood will not work). Do not use EDTA or plain tubes.

**Method**: 48 & 72 hour synchronised suspension cultures

**Test results**: Normal or Abnormal. If Abnormal, the cytogenetic lesions are listed in accordance to the International System for Human Cytogenetic Nomenclature (ISCN, 2005).

**Turnaround time**: Preliminary Result – 3 days (mean)

**Day(s) test set up**: Monday – Saturday (office hours)

CHROMOSOME ANALYSIS FOR CONGENITAL DISORDERS / SUBFERTILITY

**Common indications**: Previous family history of chromosome abnormality
Children with multiple congenital abnormalities
Recurrent abortions or infertility
Mental retardation of undetermined origin
Breakage studies for Fragile X and Fanconi Anaemia are not performed by this lab.

**Specimen required**: Peripheral blood (3 – 5 mL). Neonate blood (2 – 3 mL) in sodium/lithium heparin. Invert gently several times to mix blood and vial contents (clotted blood will not work). Do not use EDTA or plain tubes.

**Method**: 72 hour synchronised suspension cultures

**Test results**: Normal or Abnormal. If Abnormal, the cytogenetic lesions are listed in accordance to the International System for Human Cytogenetic Nomenclature (ISCN, 2005).

**Turnaround time**: 7 days (mean)

**Day(s) test set up**: Monday – Saturday (office hours)

PRODUCTS OF CONCEPTION

**Special instructions**: Pre-rinse all the specimens in sterile transport medium supplied by the Cytogenetics Laboratory or in sterile 0.9% saline solution.

**Specimen required**: Early gestation (20 – 50 mg) placental tissue in a sterile tube containing transport medium supplied by the Cytogenetics Laboratory

**Late gestation – Fetal gonad**

**Method**: In situ coverslip technique

**Test results**: Normal or Abnormal. If Abnormal, the cytogenetic lesions are listed in accordance to the International System for Human Cytogenetic Nomenclature (ISCN, 2005).

**Turnaround time**: 9 days (mean)

**Day(s) test set up**: Monday – Saturday (office hours)
### CHROMOSOME ANALYSIS FOR HAEMATOLOGICAL MALIGNANCES

**Common indications:**
- Acute Leukaemia
- Lymphoma
- Myelodysplastic Syndrome
- Chronic Myeloid Leukaemia
- Other Myeloproliferative Disorders

**Special instructions:**
Provide all necessary information on the Request For Cytogenetics Investigation form, including diagnosis and white cell count. This information is essential when initiating the cultures for cytogenetic analysis.

**Specimen required:**
Bone marrow (1.5 mL) in sodium/lithium heparin. A blood specimen (5 – 10 mL) in sodium/lithium heparin will yield results if blasts are present in large numbers (preferably >25%). Blood can also be used for investigation of Lymphoproliferative Disorders (LPD) such as Chronic Lymphocytic Leukaemia (CLL).

Do not use EDTA. The specimen should be taken from the first or second aspiration and it should not be haemodiluted. If the first aspirate of marrow is inadequate, it may be helpful to rotate the needle 180° to minimise blood contamination before re-taking the marrow sample.

**Method:**
Direct and 24-hour or 48-hour or 72-hour cultures. A stimulated culture is set up for LPD and multiple myeloma (MM)

**Test results:**
Normal or Abnormal. If Abnormal, the cytogenetic lesions are listed in accordance to the International System for Human Cytogenetic Nomenclature (ISCN, 2005)

**Turnaround time:**
7 days (mean)

**Day(s) test set up:**
Monday – Saturday (office hours)

### CHROMOSOME ANALYSIS OF LYMPH NODES/FINE NEEDLE ASPIRATES

**Sample required:**
Fresh, unfixed lymph nodes (minimum 0.5 mm³) or Fine Needle Aspirates (FNA) in a sterile container with transport medium supplied by the laboratory or Fine Needle Aspirates in sodium/lithium heparin

**Method:**
24-hour culture

**Test results:**
Normal or Abnormal. If Abnormal, the cytogenetic lesions are listed in accordance to the International System for Human Cytogenetic Nomenclature (ISCN, 2005).

**Turnaround time:**
7 days (mean)

**Day(s) test set up:**
Monday – Saturday (office hours)
CHROMOSOME ANALYSIS OF SOLID TUMOURS

Special instructions: Contact the Cytogenetics Laboratory before sending a specimen.

Sample required: Fresh, unfixed tumours (soft tissue, minimum 5 mm³) in a sterile container with transport medium supplied by the laboratory.

Method: Long-term culture

Test results: Normal or Abnormal. If Abnormal, the cytogenetic lesions are listed in accordance to the International System for Human Cytogenetic Nomenclature (ISCN, 2005).

Turnaround time: 18 days (mean)

Day(s) test set up: Monday – Saturday (office hours)

CHROMOSOME ANALYSIS WITH FLUORESCENCE IN SITU HYBRIDIZATION (FISH) TEST FOR MICRODELETION SYNDROMES

Common indications: This test is useful for patients suspected of Prader-Willi (PW) / Angelman (AS), DiGeorge (DGS) or Williams (WS) syndromes. Other microdeletion probes available include the SRY gene probe.

Special instructions: This FISH test is always used in conjunction with conventional cytogenetic studies. Contact the Cytogenetics Laboratory before sending a specimen.

Sample required: Peripheral blood

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 results: Deleted or Not deleted. Nomenclature given is in accordance to the International System for Human Cytogenetic Nomenclature (ISCN, 2005).

Reference values: 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: 6 days (mean)

Day(s) test set up: Monday – Saturday (office hours)
INTERPHASE FLUORESCENCE IN SITU HYBRIDIZATION (FISH) TEST WITH DUAL FUSION TRANSLOCATION PROBES – BCR/ABL

Common indications: This is a rapid test to identify patients with CML (chronic myelogenous leukaemia).

Special instructions: This FISH test may be requested in conjunction with conventional cytogenetic studies or as a standalone test depending on the disease status. The FISH assay should preferably be done on cases prior to treatment so as to determine the baseline pattern.

Sample required: Blood or bone marrow in sodium/lithium heparin

Method: Fluorescence In Situ Hybridization using direct-labelled FISH DNA probes and analysed under fluorescence microscopy

Test results: Normal or with fusion signals. Findings are reported in accordance to the International System for Human Cytogenetic Nomenclature (ISCN, 2005).

Reference values: Equal or greater than 0.8% of interphase cells with a double fusion signal (2F1R1G) or a variant pattern is outside the normal range

Turnaround time: 3 days (mean)

Day(s) test set up: Monday – Saturday (office hours)

INTERPHASE FLUORESCENCE IN SITU HYBRIDIZATION (FISH) TEST WITH DUAL FUSION TRANSLOCATION PROBES – ETO/AML

Specimen required: Blood or bone marrow

Method: Fluorescence In Situ Hybridization using direct-labelled FISH DNA probes and analysed under fluorescence microscopy

Test results: Normal or abnormal with fusion signals. Findings are reported in accordance to the International System for Human Cytogenetic Nomenclature (ISCN, 2005).

Reference values: Equal or greater than 0.8% of interphase cells with a double fusion signal (2F1R1G) or a variant pattern is outside the normal range

Turnaround time: 3 days (mean)

Day(s) test set up: Monday – Saturday (office hours)
**INTERPHASE FLUORESCENCE IN SITU HYBRIDIZATION (FISH) TESTS WITH DUAL-COLOUR BREAKAPART PROBES – CBFB (INV 16), MLL (11Q23), RARA (17Q21) IN ACUTE MYELOID LEUKAEMIA**

**Common indications**: Breakapart DNA probes detect disruptions to gene sequences that are known to be involved in translocations with another partner or in which the gene is disrupted due to other rearrangements such as an inversion.

**Special instructions**: This FISH test may be requested in conjunction with conventional cytogenetic studies or as a standalone test depending on the disease status. The FISH assay should preferably be done on cases prior to treatment so as to determine the baseline pattern.

**Specimen required**: Blood or bone marrow in sodium/lithium heparin

**Method**: Fluorescence In Situ Hybridization using direct-labelled FISH DNA probes and analysed under fluorescence microscopy

**Test results**: Disruption (Positive) or Non-disruption (Negative) of the gene. Findings are reported in accordance to the International System for Human Cytogenetic Nomenclature (ISCN, 2005).

**Reference values**: Equal or greater than 1.8% of interphase cells with a split signal (2F1R1G) or a variant pattern is outside the normal range

**Turnaround time**: 3 days (mean)

**Day(s) test set up**: Monday – Saturday (office hours)

---

**INTERPHASE FLUORESCENCE IN SITU HYBRIDIZATION TEST FOR MINIMAL RESIDUAL DISEASE (FISH – MRD) WITH BCR/ABL1 AND AN X- OR Y- PROBE**

**Common indications**: FISH test using BCR/ABL1 and X- or Y-probe is useful for the identification of a Ph chromosome in recipient cells with chronic myelogenous leukaemia (CML) or acute lymphoblastic leukaemia (ALL) following sex-mismatched post bone marrow transplantation. The test is also useful in determining the level of engraftment, as well as in monitoring minimal residual disease.

**Special instructions**: This FISH test may be used with conventional cytogenetic studies in some cases. When chromosome analysis procedures are performed, they are charged separately. BCR/ABL1 probes may be used in conjunction with other DNA probes.

**Specimen required**: Blood or bone marrow in sodium/lithium heparin

**Method**: Fluorescence In Situ Hybridization using direct-labelled FISH DNA probes and analysed under fluorescence microscopy
Test results: Positive (with fusion signals) or Negative for residual BCR/ABL1 and percentage of donor or recipient cells present. Findings are reported in accordance to the International System for Human Cytogenetic Nomenclature (ISCN, 2005).

Reference values: Equal or greater than 0.8% of interphase cells with a double fusion signal (2F1R1G) or a variant pattern is outside the normal range.

Turnaround time: 3 days (mean)

Day(s) test set up: Monday – Saturday (office hours)

INTERPHASE FLUORESCENCE IN SITU HYBRIDIZATION (FISH) TEST WITH DUAL FUSION TRANSLOCATION PROBES – PML/RARA

Common indications: This is a rapid test to identify patients with AML-M3 (acute promyelocytic leukaemia).

Special instructions: This FISH test may be requested in conjunction with conventional cytogenetic studies or as a standalone test depending on the disease status. The FISH assay should preferably be done on cases prior to treatment so as to determine the baseline pattern.

Specimen required: Blood or bone marrow in sodium/lithium heparin

Method: Fluorescence In Situ Hybridization using direct-labelled FISH DNA probes and analysed under fluorescence microscopy.

Test results: Normal or abnormal with fusion signals. Findings are reported in accordance to the International System for Human Cytogenetic Nomenclature (ISCN, 2005).

Reference values: Equal or greater than 0.8% of interphase cells with a double fusion signal (2F1R1G) or a variant pattern is outside the normal range.

Turnaround time: 3 days (mean)

Day(s) test set up: Monday – Saturday (office hours)

INTERPHASE FLUORESCENCE IN SITU HYBRIDIZATION (FISH) PANEL TEST FOR CHRONIC LYMPHOCYTIC LEUKAEMIA (CLL)

Common indications: FISH test DNA probes LSI ATM, CEP 12, LSI D13S25 and LSI TP53 are used in the detection of multiple chromosome aberrations that are often found in B-cell Chronic Lymphocytic Leukaemia (CLL). Many of these chromosome aberrations are subtle or are found in non-dividing cells that interphase FISH can be used to delineate instead of conventional cytogenetics.

Special instructions: The FISH Panel test is charged as separate test from the accompanying cytogenetic test. Four DNA probes/probe sets are used in this panel test.

Specimen required: Blood or bone marrow in sodium/lithium heparin
Method: Fluorescence In Situ Hybridization using direct-labelled FISH DNA probes and analysed under fluorescence microscopy. The probe set includes the LSI ATM probe that hybridizes to the 11q22.3 region of chromosome 11, the CEP 12 probe that hybridizes to the centromeric region of chromosome 12 (12p11.1-q11), the LSI D13S25 probe that hybridizes to the 13q14.3 region of chromosome 13, and the LSI TP53 probe that hybridizes to the 17p13.1 region of chromosome 17.

Test results: Positive or Negative for CLL. Findings are reported in accordance to the International System for Human Cytogenetic Nomenclature (ISCN, 2005).

Reference values: Equal to or greater than 5% of interphase cells with losses or gains

Turnaround time: 5 days (mean)

Day(s) test set up: Monday – Saturday (office hours)

**INTERPHASE FLUORESCENCE IN SITU HYBRIDIZATION (FISH) PANEL TEST FOR MULTIPLE MYELOMA (MM)**

Common indications: FISH test DNA probes LSI FGFR3/IGH@, CCND1/IGH@, LSI RB1, and LSI TP53 are used in the detection of multiple chromosome aberrations often found in multiple myeloma (MM). If hyperdiploidy is suspected, CEP 9, CEP 11, and CEP 15 probes can also be offered. Many of the cytogenetic abnormalities are subtle or are found in non-dividing cells that interphase FISH can be used to enumerate instead of conventional cytogenetics.

Special instructions: The FISH Panel test is charged as separate test from the accompanying cytogenetic test. Four DNA probes/probe sets are used in this panel test.

Specimen required: Blood or bone marrow in sodium/lithium heparin

Method: Fluorescence In Situ Hybridization using direct-labelled FISH DNA probes and analysed under fluorescence microscopy. The probe set includes the FGFR3/IGH@ dual fusion translocation probe that hybridizes to t(4;14) with breakpoints at the FGFR3 region on 4p16 and the IGH@ region on 14q32, the CCND1/IGH@ dual fusion translocation probe that hybridizes to t(11;14) with breakpoints at the major translocation cluster on 11q13 and at the IGH@ region on 14q32, the LSI RB1 probe that hybridizes to the 13q14.3 region of chromosome 13 and the LSI TP53 probe that hybridizes to the 17p13.1 region of chromosome 17.

Test results: Positive or Negative for MM. Findings are reported in accordance to the International System for Human Cytogenetic Nomenclature (ISCN, 2005).

Turnaround time: 5 days (mean)

Day(s) test set up: Monday – Saturday (office hours)
INTERPHASE FLUORESCENCE IN SITU HYBRIDIZATION (FISH) PANEL TEST FOR MYELODYSPLASTIC SYNDROME (MDS)

Common indications: FISH test DNA probes: LSI EGR1, LSI D7S486, LSI D20S108, and CEP 8 are used in the detection of myelodysplastic syndrome (MDS).

Special instructions: The FISH Panel test is charged as a separate test from the accompanying cytogenetic test. Four DNA probes/probe sets are used in this panel test.

Specimen required: Blood or bone marrow in sodium/lithium heparin

Method: Fluorescence in Situ Hybridization using direct-labelled FISH DNA probes and analysed under fluorescence microscopy. The probe set includes the LSI EGR1 probe that hybridizes to 5q31, the LSI D7S486 probe that hybridizes to 7q31, the LSI D20S108 probe that hybridizes to 20q12, and the CEP 8 probe that hybridizes to the centromere of chromosome 8.

Test results: Positive or Negative for MDS. Findings are reported in accordance to the International System for Human Cytogenetic Nomenclature (ISCN, 2005).

Reference values: Equal or greater than 5% of interphase cells with loss or gains

Turnaround time: 5 days (mean)

Day(s) Test set up: Monday – Saturday (office hours)

INTERPHASE FLUORESCENCE IN SITU HYBRIDIZATION TESTS FOR OTHER HAEMATOLOGICAL MALIGNANCIES

- ASS (9q34)
- IGH@ breakapart (14q32)
- p16 (9p21)
- TEL/AML1 dual fusion probe set

INTERPHASE FLUORESCENCE IN SITU HYBRIDIZATION (FISH) TEST FOR ANAPLASTIC LYMPHOMA KINASE (ALK)

Common indications: This FISH test using the LSI ALK dual colour, break apart rearrangement DNA probe is used in the detection of disruption of the ALK gene on 2p23 seen in t(2;5) rearrangements and its variants.

Special instructions: The FISH test is optimal with fresh tissue samples. Tissue sections should preferably be between 4 – 6 µm in thickness.

Specimen required: Freshly-cut tissue sections, tissue imprints or bone marrow specimen

Method: Fluorescence in Situ Hybridization using direct-labelled FISH DNA probe and analysed under fluorescence microscopy
SECTION 4: SAMPLE COLLECTION & HANDLING – SPECIAL INSTRUCTIONS & LAB TESTS

Test results: Disruption (Positive) or Non-disruption (Negative) of ALK. Findings are reported in accordance to the International System for Human Cytogenetic Nomenclature (ISCN, 2005).

Reference values: Equal to or greater than 8% of interphase cells with disruption of the ALK gene

Turnaround time: 5 days (mean)

Day(s) Test set up: Monday – Saturday (office hours)

INTERPHASE FLUORESCENCE IN SITU HYBRIDIZATION TEST FOR BREAST CANCER TISSUES USING HER2 (ERBB2) DNA PROBE

Common indications: FISH test using HER2 (ERBB2) DNA-probe is useful for the detection of amplification of the HER2 (ERBB2) gene that plays a key role in the regulation of cell growth. This gene is amplified in human breast cancer.

Special instructions: The FISH test is optimal with fresh tissue samples. Tissue sections should preferably be between 4 – 6 µm in thickness.

Sample required: Newly-cut fresh breast cancer sections is ideal

Method: Fluorescence In Situ Hybridization using direct labelled LSI HER2 (ERBB2) and CEP 17 DNA probes and analysed under fluorescence microscopy

Test results: Positive, Borderline Positive or Negative for HER2 (ERBB2) amplification. Findings are reported in accordance to the International System for Human Cytogenetic Nomenclature (ISCN, 2005).

Reference values: Normal specimens have a HER2 (ERBB2) to CEP 17 signal ratio of < 2.0. Specimens with amplification have a ratio of ≥ 2.0 but results at or near the cut-off point of 1.8 – 2.2 should be interpreted with caution.

Turnaround time: 5 days (mean)

Day(s) test set up: Monday – Saturday (office hours)

INTERPHASE FLUORESCENCE IN SITU HYBRIDIZATION (FISH) TEST FOR EWING SARCOMA

Common indications: This FISH test using the LSI EWSR1 dual colour, breakapart rearrangement probe is used in the detection of disruption of the EWSR1 gene on 22q12 seen in t(11;22) rearrangements and its variants.

Special instructions: The FISH test is optimal with fresh tissue samples. Tissue sections should preferably be between 4 – 6 µm in thickness.

Specimen required: Freshly-cut tissue sections or tissue imprints
Method: Fluorescence in Situ Hybridization using direct-labelled LSI EWSR1 dual colour DNA probe and analysed under fluorescence microscopy

Test result: Disruption (Positive) or Non-disruption (Negative) of the EWSR1 gene. Findings are reported in accordance to the International System for Human Cytogenetic Nomenclature (ISCN, 2005).

Reference values: Equal to or greater than 8% of interphase cells with disruption of the EWSR1 gene

Turnaround time: 5 days (mean)

Day(s) test set up: Monday – Saturday (office hours)

INTERPHASE FLUORESCENCE IN SITU HYBRIDIZATION (FISH) TESTS FOR LYMPHOMA (PARAFFIN SECTIONS) – OTHER PROBES

- BCL6 (3q27) breakapart probe
- CCND1/IGH@[t(11;14)]
- IGH@/BCL2 [t(14;18)]
- IGH@/MYC/CEP8 dual fusion probe set
- MALT1 breakapart probe
- MYC (8q24) breakapart
- SYT (18q21) breakapart (Synovial Sarcoma)
- EVT6 (for isochromosome 12p in Germ Cell Tumour)
- T0PII/CEP17

New probes are continually being added to our existing array of tests. Please check with the laboratory on probe availability. The requirements are the same as for the above FISH tests on paraffin sections.

Avoid sending samples on Saturdays and eve of public holidays.

INTERPHASE FLUORESCENCE IN SITU HYBRIDIZATION (FISH) TEST FOR MUCOSA-ASSOCIATED LYMPHOID TISSUE (MALT) TYPE LYMPHOMA

Common indications: This FISH test using the LSI API2/MALT1 dual colour, dual fusion translocation probe is used in the detection of the t(11;18) rearrangements involving the API2 gene on 11q21 and the MALT1 gene on 18q21 seen in mucosa-associated lymphoid tissue (MALT) type lymphoma.

Special instructions: The FISH test is optimal with fresh tissue samples. Tissue sections should preferably be between 4 – 6 µm in thickness.

Specimen required: Freshly-cut tissue sections or tissue imprints.

Method: Fluorescence in Situ Hybridization using direct-labelled FISH DNA probes and analysed under fluorescence microscopy.
INTERPHASE FLUORESCENCE IN SITU HYBRIDIZATION (FISH) TEST FOR NEUROLOGICAL MALIGNANCIES

Common indications: This FISH test uses the LSI 1p36/LSI 1q25 & LSI 19q13/LSI 19p13 dual colour probe sets to detect chromosomal deletions involving the 1p36 and 19q13 regions.

Special instructions: The FISH test is optimal with fresh tissue samples. Tissue sections should preferably be between 4 – 6 µm in thickness.

Specimen required: Freshly-cut tissue sections or tissue imprints

Method: Fluorescence in Situ Hybridization using direct-labelled FISH DNA probes and analysed under fluorescence microscopy

Test results: Deleted or Non-deleted signals. Findings are reported in accordance to the International System for Human Cytogenetic Nomenclature (ISCN, 2005).

Reference values: Losses of 1p36 and/or 19q13 regions equal to or greater than 25% of interphase cells, and/or a ratio of 1p36/1q25 and/or 19q13/19p13 of equal to or less than 0.8

Turnaround time: 5 days (mean)

Day(s) Test set up: Monday – Saturday (office hours)

RAPID PRENATAL ANEUPLOIDY SCREENING, DOWN SYNDROME SCREENING AND SEX CHROMOSOME DETERMINATION WITH FLUORESCENCE IN SITU HYBRIDIZATION TEST

Common indications: Advanced maternal age (≥ 35 years at EDD), Abnormal ultrasound findings, Positive maternal serum screening, Previous history of chromosome abnormality, Family history of Down syndrome, Parental anxiety

Special instructions: This FISH test is usually requested in conjunction with conventional cytogenetic studies.

Sample required: Amniotic fluid (3 mL), chorionic villi (5 mg), or fetal cord blood/neonate blood (0.5 mL)

Method: Chromosome analysis as with Amniotic Fluid/Chorionic Villi or Fetal Cord Blood/Neonate Blood FISH assay using LSI 13 and 21, and CEP X, Y, and 18 DNA probes labelled with fluorophores are analysed under fluorescence microscopy.
<table>
<thead>
<tr>
<th>Test results</th>
<th>Normal or Abnormal, with the abnormalities listed. Findings are reported in accordance to the International System for Human Cytogenetic Nomenclature (ISCN, 2005).</th>
</tr>
</thead>
<tbody>
<tr>
<td>Reference values</td>
<td>Individuals with a trisomy will show three signals of a particular DNA probe. Normal individuals will show two signals of each DNA probe. Females will show two signals of the X probe, males one signal of the X and Y probe each.</td>
</tr>
<tr>
<td>Turnaround time</td>
<td>1 day (mean)</td>
</tr>
<tr>
<td>Day(s) test set up</td>
<td>Monday – Friday (office hours)</td>
</tr>
</tbody>
</table>