Special Investigations At
Sri Nathella Sampathu Chetty Clinical Laboratory
A Unit of Medical Research Foundation
Sankara Nethralaya
41, College Road, Chennai
Email: snsclab@snmail.org

By
Dr. N. Angayarkanni Ph.D,
Prof, Biochemistry

Clinical Biochemistry
Clinical Hematology
Clinical Microbiology
Since 2007

- Clinical Biochemistry : 19
- Clinical Pathology : 19
- & Hematology : 20
- Microbiology & Serology : 27
- Histopathology & Cytopathology : 07

Total : 96 Tests

Routine and special

+ Genetics and Molecular Biology
(tests not under scope of NABL)

(Validity until 13.8.2017)
Special Investigations in Biochemistry

Dr. N. Angayarkanni
HPLC analysis

Agilent HPLC: with UV and Fluorescent detector.
Shimadzu HPLC: UV, Fluorescent, Diode array Electrochemical

- Plasma and urine Amino acids*
- Vitamin A, E analysis
Atomic Absorption Spectrophotometer: A700 – Perkin Elmer
With Flame and Graphite Furnace

Trace Elements and Toxic Heavy metals Analysis

- Copper
- Zinc
- Iron
- Chromium
- Selenium
- Arsenic
- Mercury
- Cadmium
- Lead

Since Jun 2009
### Other special investigations

| Screening for inborn errors of metabolism:  
| Urine screening: Maple syrup syndrome, cystinuria, phenylketonuria, tyrosinosis, Homogentisic aciduria, Mucopolysaccharidosis, homocysteine Galactosemia |
| Angiotensin Converting Enzyme |
| Homocysteine |
| Ornithine |
| Serum Amylase |
| Lactate / pyruvate |
| Eales' special biochemistry Work up (Glutathione, Glutathione peroxidase (GPx), Superoxide Dismutase (SOD), TBARS (oxidative stress marker), Vitamin A, E and C) |
| Ceruloplasmin |
| Electrophoresis: protein (CSF/plasma) |
Histopathology

Special Stains

H&E stain  PAS stain  GMS stain for fungus

Gomori’s trichrome stain  Congo red stain for amyloid  Gram stain for bacteria

Dr. S. Krishna Kumar  Dr. J. Biswas
Detection of Chromosome 3 aberrations for melanoma
Chromogenic *In situ* Hybridisation (CISH).

**LIVER METASTASIS**

**NONINVASIVE**

**NORMAL RETINA**

CISH for chromosome 3:

Hybridized with chromosome 3

[A] Invasive melanoma

[B] Non-metastasising melanoma

[C] Normal retina
Microbiology & Serology

DNA CHIPS FOR FOUR DIFFERENT OCULAR CONDITIONS

INFECTIONOUS ENDOPHTHALMITIS

EXTERNAL OCULAR INFECTIONS

VIRAL RETINITIS

UVEITIS & OTHER CLINICALLY SUSPECTED MYCOBACTERIAL INFECTIONS

Dr. H.N. Madhavan  Dr. K. Lily Therese
Indirect immunofluorescence test for ANA or Fluorescent anti nuclear antibody test (FANA)

Substrate:
- Primate liver tissues
- Rodent tissue sections
- HEp-2 cells as substrate (In house)

ELISA for ANA

Antigen composition
Screening tests:
- whole HEp-2 nuclei
- extract of HEp-2 nuclei
- mixture of defined nuclear antigens,
Diagnostic tests:
- antibody profile (4-8 different antibodies
- single defined antigen
Indirect immunofluorescence test
- Perinuclear anti neutrophil cytoplasmic antibody (pANCA)
- Cytoplasmic anti neutrophil cytoplasmic antibody (cANCA)

Substrate:
Ethanol fixed granulocytes

ELISA
- pANCA - Various antigens - MPO, Lactoferrin, Elastase, Cathepsin G
- cANCA – Proteinase 3 (PR3)
Detection of AQP4 antibodies

Neuro Myelitis Optica Vs Multiple Sclerosis

The detection of anti-AQP4 antibodies facilitates an early stratification of NMO and MS, which is highly important due to the different treatment recommendations.

**Treatment modalities: MS and NMO**

**MS:** Immunomodulatory therapies
- Interferon beta (IFN-β) and
glatiramer acetate (GA)

**NMO:** Corticosteroids and immunosuppressive agents.
- Non responders: Plasma exchange or
treatment with rituximab

NMO-IgG antibody which is highly specific (>99%) , sensitivity 48-72% depending on the assay used.

REVERSE TRANSCRIPTASE PCR TARGETING 85B GENE TO DETECT VIABLE *MYCOBACTERIUM TUBERCULOSIS* FROM CLINICAL SPECIMENS

**ADVANTAGES OF REVERSE TRANSCRIPTASE PCR**

- Rapid Diagnostic method to detect viable *Mycobacterium tuberculosis* directly from any clinical specimen **within 7-8 hours.**

- Early detection of *M.tuberculosis* genome in active tuberculosis is useful for initiation of antituberculous therapy.
SN ONGC Dept. of Genetics and Molecular Biology

..From Disease to Gene..
Patient care

- Genetic testing and chromosomal study
- Genetic counseling
<table>
<thead>
<tr>
<th>Name of the test</th>
<th>Condition for which the test is done</th>
<th>Specimen</th>
</tr>
</thead>
<tbody>
<tr>
<td>Rhodopsin <em>(RHO)</em> Gene Screening</td>
<td>Retinitis pigmentosa &amp; Congenital Stationary Night Blindness</td>
<td></td>
</tr>
<tr>
<td><em>RPE65</em> Gene Screening</td>
<td>Retinitis Pigmentosa &amp; Leber’s Congenital Amaurosis</td>
<td></td>
</tr>
<tr>
<td><em>CYP1B1</em> Gene Screening</td>
<td>Peter’s anomaly, Congenital Glaucoma, Primary and Juvenile Open Angle Glaucoma</td>
<td></td>
</tr>
<tr>
<td><em>MYOC</em> Gene Screening</td>
<td>Primary and Juvenile Open Angle Glaucoma</td>
<td></td>
</tr>
<tr>
<td><em>PAX6</em> Gene Screening</td>
<td>Peter’s anomaly &amp; Aniridia</td>
<td></td>
</tr>
<tr>
<td>Retinoschisis <em>(RS1)</em> Gene Screening</td>
<td>Retinoschisis</td>
<td></td>
</tr>
<tr>
<td>Cytogenetic analysis for Retinoblastoma</td>
<td>Retinoblastoma</td>
<td></td>
</tr>
<tr>
<td>Screening the three primary mitochondrial mutations for Leber’s Hereditary Optic Neuropathy (LHON)</td>
<td>Lebers Hereditary Optic Neuropathy (LHON)</td>
<td></td>
</tr>
<tr>
<td>Hereditary Optic Neuropathy (LHON)</td>
<td></td>
<td></td>
</tr>
<tr>
<td><em>BBS</em> Genes screening for Bardet – Biedl Syndrome</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Chromosomal Study</td>
<td>Chromosomal abnormalities</td>
<td></td>
</tr>
</tbody>
</table>
Patient Information

Genetic Clinic Information Sheet

<table>
<thead>
<tr>
<th>Name:</th>
<th>Education:</th>
<th>MRD No.:</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sex:</td>
<td>Occupation:</td>
<td>Genetic No.:</td>
</tr>
<tr>
<td>Age:</td>
<td>Place:</td>
<td>Ref. Consultant:</td>
</tr>
<tr>
<td>Age of Onset</td>
<td>Contact No.:</td>
<td></td>
</tr>
</tbody>
</table>

PEDIGREE

Address for communication:
(Temporary Address)

E-mail Address:

SNo. | Name | Sex | Age | Relation to proband | MRD No. / C No. | DNA No. |

Updates:

Prepared by: Approved by:

(PTO)
Patient 1

- Diagnosis - Retinoblastoma

Pre-test counseling
- Clinical evaluation, pedigree construction and analysis for risk transmission to the siblings
- Decision on genetic testing

Genetic testing (RB1 gene)
- Screening and identification of the genetic defect in affected member (Father and sib of patient 1)
- Prenatal testing in the sib for carrier detection and risk transmission

Post test counseling
- Genetic defect / risk identified in Patient 1 and explained the clinical relevance
- Clinical monitoring prenatally and also after birth for symptoms
- Clinical intervention and effective management of the disease once detected

Better lifestyle of the patient
Hematological Investigations

Dr. S. B. Vasanthi  Dr. Doreen Gracias
Forth coming!

COAGULATION ANALYSER

1. Prothrombin Time
2. Activated Partial Thromboplastin Time
3. Protein C
4. Protein S
5. Anti Thrombin III
6. APC – R
7. DRVV Screen (Lupus Screening)
8. DRVV Confirm (Lupus Confirmation)
9. Factor VIII
10. Factor IX
11. VWF (Von Willebrand factor)
12. STA FIB2
13. STA Thrombin2
14. D- Dimer
15. FDP- Plasma

STA COMPACT FULLY AUTOMATED
A Sankara Nethralaya publication

Manual of Medical Laboratory Techniques

S Ramakrishnan
KN Sulochana

Jaypee: publisher
Rs. 450/-
Thank you

SNSC clinical laboratory

The Pro-active Dynamic team

21.7.13