

Unique case.....Unique contributions.....

**1. Diagnosis of previously lethal, undiagnosed immune deficiencies**

4 month old baby presented in 2005 with an unexplained family history of 15 deaths in males in 3 generations. Investigations done at the Immunology Department of MRI revealed the diagnosis as X linked severe combined immunodeficiency (SCID). The baby succumbed to the condition. In 2010 the mother presented at 20 weeks of pregnancy. A scan revealed that it was a male baby. His blood was tested at Department of Immunology and the diagnosis was confirmed as SCID. The genetic unit of the Faculty of Medicine too confirmed the diagnosis by gene sequencing. After birth he underwent a stem cell transplant (bone marrow transplant) in India. He celebrated his 1<sup>st</sup> birthday on the 15<sup>th</sup> of July.

Another baby whose brother had died at 3 months was also diagnosed as having the same disease & has been referred to USA for gene therapy.

**2. Bubonic plague – how an epidemic was averted in Sri Lanka**

Plague is a disease which has caused millions of deaths in the past. Eg. In the 14<sup>th</sup> century 1/3<sup>rd</sup> of the world's population succumbed to this disease. Plague is transmitted from infected rats to humans via fleas. There is human to human transmission through respiratory secretions. An outbreak of plague in India occurred in 1994. A sailor from a Russian ship, which had called at Mumbai port, developed symptoms similar to plague. The ship docked at the Colombo Port, and a team from MRI headed by the late Dr. RSB Wickremasinghe, Consultant Microbiologist, visited the ship. Investigations at the MRI confirmed the diagnosis of plague. The rats in the harbor were screened for infected fleas. The measures taken by the MRI averted a major epidemic.

**3. Diagnosis of rare fungal diseases**

The Department of Mycology is the reference laboratory for the diagnosis of fungal infections. A 1 ½ year old child from a remote village off Badulla presented with a lump on the nose and swelling of the eye. A tumour was suspected and biopsy taken. The department of Mycology isolated a fungus, rarely diagnosed, *Saksenia varsiformis*. This fungus has been identified in only 26 patients world wide. The patient was appropriately treated.

**4. Diagnosis of bleeding disorders**

Two sisters from Jaffna aged 16 and 18 years were referred to haematology department of MRI with recurrent episodes of bleeding. Their brother had died due to bleeding in to the brain platelet function test done at MRI revealed Glanzmanns Disease and the diseased sisters will be treated appropriately .

**5. Electron microscopy**

The MRI has 2 electron microscopes, a scanning and a transmission microscope. The transmission microscope is the only one in Sri Lanka. These can magnify specimens X 500,000. The electron microscope can be used on biological specimens from human, animal and plant tissue, to identify the ultrastructure of cells and tissue and identify viruses. They can be used on non biological specimen, such as nano particles, as has been done at the MRI.

**6. Animal centre**

The MRI animal centre has produced antibodies used in the diagnosis of viral and bacterial

pathogens of plants, including those attacking coconut plants, potato, lime, pineapple and anthurium. This work was carried out in collaboration with the Plant virus indexing centre, Department of Agriculture.