

# **USE OF POLYMERASE CHAIN REACTION IN LABORATORY TESTING**

## **INTRODUCTION**

Polymerase Chain Reaction (PCR) is a in vitro amplification of specific DNA sequences by simultaneous primer extension of complementary strands of DNA where numerous copies of small fragments of genetic material can be produced to facilitate the analysis of genetic material.

## **OBJECTIVE**

To assess the efficacy, efficiency and cost implications of PCR technology in medical care.

## **RESULTS, CONCLUSIONS & RECOMMENDATIONS**

PCR is a rapid, safe and cost-effective method.

### **I INFECTIOUS DISEASE**

#### **Tuberculosis**

PCR can assist in early diagnosis of TB, identify mycobacterium in clinical specimens even with smaller number of bacteria, evaluate response to therapy, detect cases of relapse and differentiate mixed infections. PCR is recommended for specific indications like disseminated TB or TB meningitis.

#### **HIV**

PCR can be used to predict HIV progression and survival, as well as the risk of vertical transmission. It can also assist in detecting the viral load including pregnancy and in staging of HIV. PCR should be used to assess HIV risk and therapy.

#### **Hepatitis B Infection**

PCR is able to detect low level of HBV viraemia especially in asymptomatic HbsAg positive subjects. It can also be used in evaluating the degree of response to therapy, identifying HBs Ag-negative patients with liver disease, in identification of HBV infection in liver transplantation, and also to identify mutant strains of HBV. It is recommended that PCR be used in evaluation of anti-viral therapies and for detection of HBV in high-risk patients.

#### **Hepatitis C Infection**

PCR can be used for qualitative and quantitative measurement of viraemia in diagnosis of acute HCV infection, diagnosis of anti-HCV-negative chronic hepatitis C carriers, evaluation of HCV viraemia in asymptomatic blood donors with normal liver enzymes, assessing virological response to treatment, predicting treatment response to alpha-interferon and assessing severity of disease. It is recommended that PCR be used in assessing treatment efficacy.

### **II INVESTIGATION OF HAEMATOLOGICAL DISEASES.**

### **HLA Typing**

Typing by PCR is simpler, accurate and reliable and is recommended as the method of choice for a transplant programme, especially in unrelated bone marrow and cord blood transplants.

### **HPA Typing**

PCR testing is recommended in the identification of post-transfusion purpura, zygosity testing, antenatal diagnosis and thrombocytopenic patients, and to distinguish between homozygous and heterozygous individuals.

### **Thalassaemias (Haemoglobinopathies)**

PCR is recommended for testing patients with haemoglobinopathies especially paediatric patients as well as in prenatal diagnosis, for carrier detection and for detection of alpha and beta thalassaemias

## **III CONGENITAL DISORDERS**

### **Duchenne Muscular Dystrophy**

PCR is recommended as an alternative method for confirmatory diagnosis of Duchenne Muscular Dystrophy.

### **Fragile X Syndrome**

PCR is recommended for the confirmatory diagnosis of Fragile X

## **IV MALIGNANT TUMOURS**

PCR is recommended for screening high-risk populations for genetic predisposition for cancer and for the monitoring of residual tumour cells in peripheral blood.

## **V PATERNITY TESTING**

PCR is recommended for paternity testing in situations where there are only minute quantities of tissue or where the tissue has been damaged.

## **VI FORENSIC IDENTIFICATION**

PCR can be used in mass disaster identification of unknown skeletal remains, sexual assaults against women, establishment of a forensic database, and to establish patient identity in cases of mix-up PCR is a valuable tool in forensic testing.

## **COST IMPLICATIONS**

The cost per test ranges from RM 40.00 to RM150.00