UNIVERSITI SAINS MALAYSIA
GERAN PENYELIDIKAN UNIVERSITI
PENYELIDIKAN
LAPORAN AKHIR

DETECTION ON COMMON DELETIONAL ALPHA-THALLASAEMIA IN PREGNANT LADY BY POLYMERASE CHAIN REACTION

PENYELIDIK

PROF. MADYA DR. ROSLINE HASSAN

PENYELIDIK BERSAMA

PROF. MADYA DR. WAN ZAIDAH ABDULLAH
DR. NOR ALIZA ABDUL GHAFFA
PROF. MADYA DR. RAPIAAH MUSTAFFA
DR. ROSNAH BAHAR
DR. TARIQ ROSHAN

2012

UNIVERSITI SAINS MALAYSIA

UNIVERSITY RESEAR
FINAL REPOR
Geran Penyelidikan
Laporan Akh

A.	**PARTICULARS OF RESEARCH** / *MAKLUMAT PENYELIDIKAN:*
(i)	**Title of Research:** *Tajuk Penyelidikan: Detection On Common Deletional Alpha-Thallasaemia I* *Polymerase Chain Reaction*
(ii)	**Account Number:** *Nombor Akaun: 1001/PPSP/812003*
B.	**PERSONAL PARTICULARS OF RESEARCHER** / *MAKLUMAT PENYELIDIK:*
(i)	**Name of Research Leader:** *Nama Ketua Penyelidik:* ***PM DR ROSLINE HASSAN***
	Name of Co-Researcher *Nama Penyelidik Bersama:* *PM Dr Wan Zaidah Abdullah, Dr Nor Aliza Abdul Ghaffar, PM Dr Rapiaah Mustaffa, DR Rosnah Bahar, Dr Tariq Roshan*
(ii)	**School/Institute/Centre/Unit :** *Pusat Pengajian /Institut/Pusat/Unit : School of Medical Sciences*
C.	**Research Platform (Please tick (/) the appropriate box):** *Pelantar Penyelidikan (Sila tanda (/) kotak berkenaan):*

- [ ] **A. Life Sciences** *Sains Hayat*
- [ ] **B. Fundamental** *Fundamental*
- [ ] **C. Engineering & Technology** *Kejuruteraan & Teknologi*
- [ ] **D. Social Transformation** *Transformasi Sosial*
- [ ] **E. Information & Communications Technology (ICT)** *Teknologi Maklumat & Komunikasi*
- [/] **F. Clinical Sciences** *Sains Klinikal*
- [ ] **G. Biomedical & Health Sciences** *Bioperubatan Sains Kesihatan*

D.	**Duration of this research :** *Tempoh masa penyelidikan ini :* ***Duration :** . ..... 3 years..... *Tempoh :* **From** : .....31 Oktober.....2007 **To** : ...29 September...2010 *Dari:* *Ke :*

E. **ABSTRACT OF RESEARCH**

(An abstract of between 100 and 200 words must be prepared in **Bahasa Malaysia and in English**. This abstract will be included in the Annual Report of the Research and Innovation Section at a later date as a means of presenting the project findings of the researcher/s to the University and the community at large)

Abstrak Penyelidikan

(Perlu disediakan di antara 100 - 200 perkataan di dalam **Bahasa Malaysia dan juga Bahasa Inggeris**.
Abstrak ini akan dimuatkan dalam Laporan Tahunan Bahagian Penyelidikan & Inovasi sebagai satu cara untuk menyampaikan dapatan projek tuan/puan kepada pihak Universiti & masyarakat luar).

ABSTRACT

In order to screen the carrier state of the common deletional types of alpha thalassaemia, we have evaluated a screening protocol and examined the prevalence and the molecular basis of the alpha thalassemia in pregnant women at Hospital Universiti Sains Malaysia (HUSM). Blood samples from two hundred (200) pregnant women were screened for α-thalassaemia. Of these, 16 were later excluded because they had been diagnosed as having HbE trait or the β-thalassemia trait. Results were evaluated with the standard haematological analyses including erythrocyte count, haemoglobin quantitation and polymerase chain reaction (PCR) analysis of α-globin gene. Altogether 17 of 184 subjects was detected as α-thalassemia ($-\alpha^{3.7}/\alpha\alpha$ and $--^{SEA}/\ \alpha\alpha$ genotype) carrier. Analysis of hematologic data showed significantly difference in MCVs and MCH (p=0.000), and the best cut-off point for predicting the presence of the α-thalassemia carrier in pregnant women was 86.3fL giving 77% sensitivity and 71% of specificity. $-\alpha^{3.7}$ kb single gene deletion (8.1%) was the commonest α-thalassemia found in this study population followed by double gene South East Asia ($--^{SEA}$) deletion (1.1%). Thus earlier screening is recommended to be performed to pregnant women and followed by paternal testing as those with $--^{SEA}$ deletion; carry higher risk of having hydrop fetalis baby if the father is also $-^{SEA}$ type carrier.

ABSTRAK

Untuk mengesan pembawa kepada pemotongan umum gen thalassemia alfa, kami telah menilai protokol saringan, pemeriksaan dan asas molekular terhadap tersebar luasnya thalassemia alfa di kalangan perempuan mengandung di Hospital Universiti Sains Malaysia (HUSM). Sampel darah dari 200 orang perempuan mengandung diambil dan melalui ujian saringan untuk mengesan thalassemia alfa. 16 daripada 200 subjek kemudiannya disingkirkan kerana telah didiagnos sebagai HbE dan thalassemia beta. Keputusan dinilai berdasarkan analisis standard hematologikal termasuk pengiraan eritrosit, kuantiti hemoglobin dan analisis tindakbalas berantai polimerase (PCR)bagi penentuan gene globin alfa. 17 subjek daripada 184 perempuan mengandung dikesan sebagai pembawa kepada thalassemia alfa ($-\alpha^{3.7}/\alpha\alpha$ and $--^{SEA}/\ \alpha\alpha$ genotaip). Analisis bagi data hematologi menunjukkan bahawa terdapat perbezaan penting bagi nilai MCV dan MCH (p<0.005) dan titik potong yang terbaik untuk mengesan pembawa kepada gen thalassemia alfa dikalangan perempuan mengandung adalah 86.3fL dengan 77% kepekaan dan 71% kekhususan. Pemotongan gen tunggal $-\alpha^{3.7}$ kb (8.1%) adalah merupakan gen thalassemia alfa yang umum ditemui di dalam populasi ini diikuti dengan pemotongan gen berganda South East Asia ($--^{SEA}$) (1.1%). Oleh sebab itu, ujian saringan awal adalah dicadangkan kepada perempuan mengandung dan diikuti dengan ujian ke atas bapa kerana jika ibubapa merupakan pembawa kepada pemotongan $-^{SEA}$ risiko untuk mendapat anak hydrop fetalis adalah tinggi.