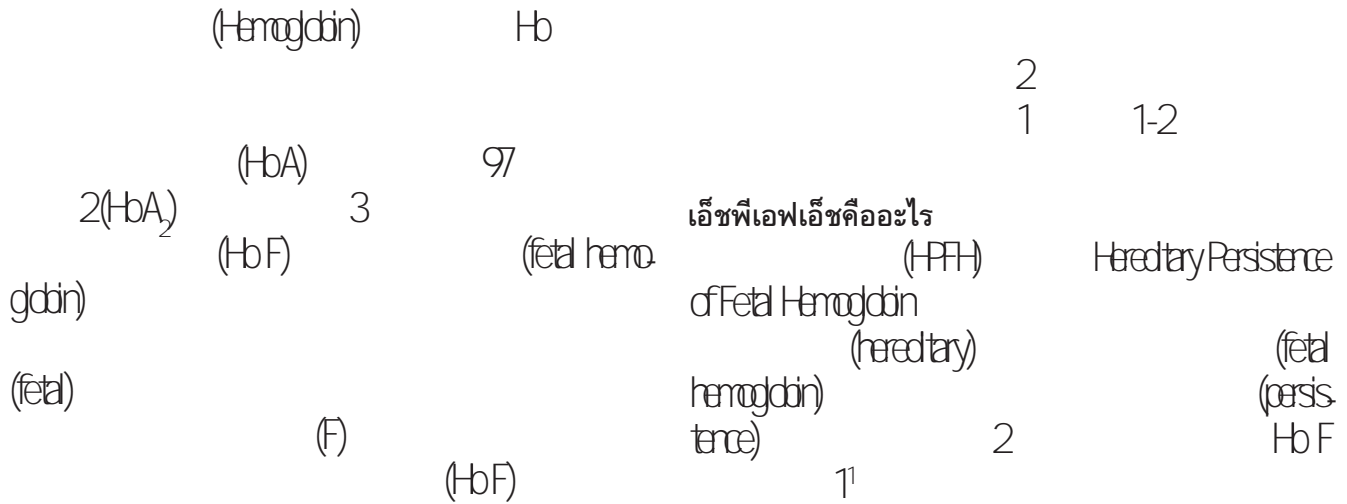


# เอชพีเอฟเอช (HPFH)

(Hereditary Persistence of Fetal Hemoglobin) ศศ. นพ.กิตติ ต่อจรัส

วิวัฒนาการของฮีโมโกลบิน



## ภาวะปกติ (Physiologic)

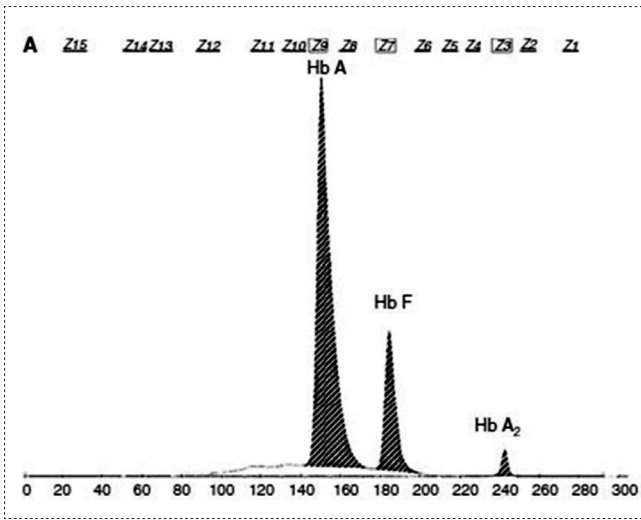
- (neonates)
- (pregnancy)

## ภาวะหรือโรคถ่ายทอดทางพันธุกรรม (Hereditary)

- (Hereditary persistence of fetal hemoglobin (HPFH))
- ( $\beta$ -thalassemia)
- ( $\beta$ -thalassemia major)
- ( $\beta$ -thalassemia intermedia)

## ภาวะหรือโรคที่ไม่ได้ถ่ายทอดทางพันธุกรรม (Acquired)

- (aplastic anemia)
- (leukemia)
- (hepatoma)
-



รูปที่ 1 A.

Hb type  
HPFH

CE

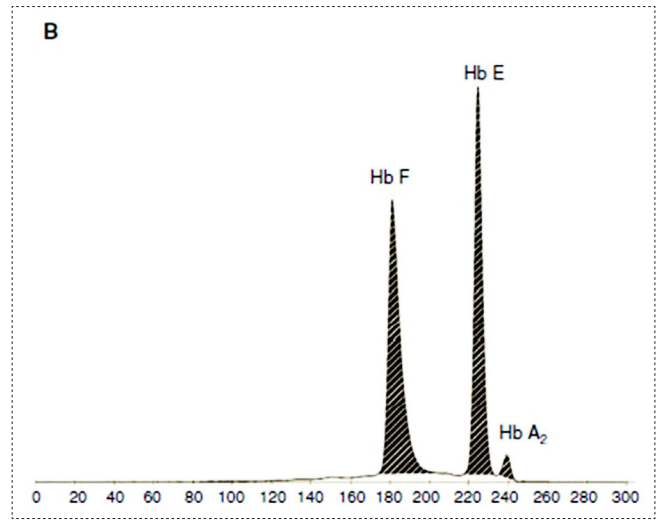
HPFH (deletion) ( $\beta$ -globin gene cluster)  
HPFH-6 (point mutation promoter)<sup>45</sup>  
(nondeletional  $\beta$ -globin gene)

HPFH CBC  
MCV, MCH blood smear normochromic, normocytic red cell  
Hb type Hb A<sub>2</sub>F Hb F 24% (2%)

HPFH Hb E HPFH/hb E (thalassaemia intermedia)

Hb type Capillary zone Electrophoresis (CE) Hb A<sub>2</sub> E F HPFH/hb E  $\beta$ -thalassaemia/hb E (pedigree) Hb type

HPFH (Hb type A E) Hb E (Hb type HPFH/hb E HPFH/hb E DNA study)<sup>6</sup>



รูปที่ 1 B.

Hb type  
 $\delta\beta$ -thalassaemia/Hb E

$\delta\beta$ -thalassaemia จำแนกเป็น 3 กลุ่มดังนี้

1. Heterozygous  $\delta\beta$ -thalassaemia  
CBC mild anaemia, hypochromic, microcytic red cell  
Hb type Hb A<sub>2</sub>F Hb F 22%<sup>46</sup>

2 Homozygous  $\delta\beta$ -thalassaemia  
thalassaemia intermedia  
anaemia, jaundice, mild hepatosplenomegaly, CBC mild anaemia, reduced red cell indices (MCV 78fL, MCH 20pg) hypochromic, microcytic red cell.  
Hb typing Hb F 100%<sup>3</sup>

3  $\delta\beta$ -thalassaemia/Hb E  
thalassaemia intermedia

anaemia, jaundice, mild hepatosplenomegaly, CBC anaemia, reduced red cell indices  
Hb type Hb A<sub>2</sub> E F  
( Capillary zone Electrophoresis (CE)  
Hb F 54.1% 1 B  
Hb F Hb E Hb type  
 $\beta^0$ -thalassaemia/Hb E <sup>3</sup>

ตารางที่ 2 แสดง Hematologic data และ Hb type ของ Heterozygous HPFH-6 และ Heterozygous  $\delta\beta$ -thalassemia

	Heterozygous HPFH-6	Heterozygous $\delta\beta$ -thalassemia
Number	70	60
$\beta$ -genotype	$\delta\beta^H\beta^A$	HPFH-6 $\beta^A$
$\alpha$ -genotype	$\alpha\alpha/\alpha\alpha$	$\alpha\alpha/\alpha\alpha$
Hb (g/dL)	11.8 $\pm$ 1.7	121 $\pm$ 45
Hct (%)	36.3 $\pm$ 4.8	36.3 $\pm$ 4.5
MCV (fL)	74.8 $\pm$ 8.7	79.3 $\pm$ 5.3
MCH (pg)	24.7 $\pm$ 1.6	25.6 $\pm$ 2.3
MCHC (g/dL)	32.5 $\pm$ 1.0	32.7 $\pm$ 1.4
RDW (%)	21.2 $\pm$ 2.9	17.0 $\pm$ 3.2
Hb Type	A <sub>2</sub> AF	A <sub>2</sub> AF
HbA <sub>2</sub>	2.3 $\pm$ 0.4	2.2 $\pm$ 0.4
HbF	2.21 $\pm$ 7.8	2.43 $\pm$ 4.4

ตารางที่ 3 แสดง Hematologic data และ Hb type ของ  $\delta\beta$ -thalassemia/Hb E และ  $\beta^0$ -thalassemia/Hb E

	$\delta\beta$ -thalassemia/HbE	$\beta^0$ -thalassemia/HbE
Number	10	45
$\beta$ -genotype	$\delta\beta^H\beta^E$	$\beta^0/\beta^E$
$\alpha$ -genotype	$\alpha\alpha/\alpha\alpha$	$\alpha\alpha/\alpha\alpha$
Rbc ( $\times 10^9/L$ )	5.6 $\pm$ 0.7	3.8 $\pm$ 0.9
Hb (g/dL)	11.9 $\pm$ 2.3	7.5 $\pm$ 1.5
Hct (%)	34.2 $\pm$ 5.9	24.0 $\pm$ 4.4
MCV (fL)	66.1 $\pm$ 4.2	61.4 $\pm$ 8.5
MCH (pg)	22.3 $\pm$ 2.3	18.8 $\pm$ 2.6
MCHC (g/dL)	34.2 $\pm$ 2.2	31.2 $\pm$ 3.2
RDW (%)	21.1 $\pm$ 3.6	29.1 $\pm$ 4.8
Hb Type	A <sub>2</sub> EF	A <sub>2</sub> EF
HbA <sub>2</sub>	2.2 $\pm$ 0.3	6.1 $\pm$ 1.9
HbE	41.6 $\pm$ 2.7	57.2 $\pm$ 13.0
HbF	5.41 $\pm$ 5.3	3.48 $\pm$ 1.51

**การรักษา**

1 heterozygous HPFH  
heterozygous  $\delta\beta$ -thalassemia  
mild anemia  
folic

2 HPFH/Hb E  $\delta\beta$ -thalas-  
semia/HbE thalassaemia intermedia  
folic

**การให้คำแนะนำปรึกษาทางพันธุศาสตร์**

(pedgree) CBC,  
Hb type DNA analysis ( )

**เอกสารอ้างอิง**

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