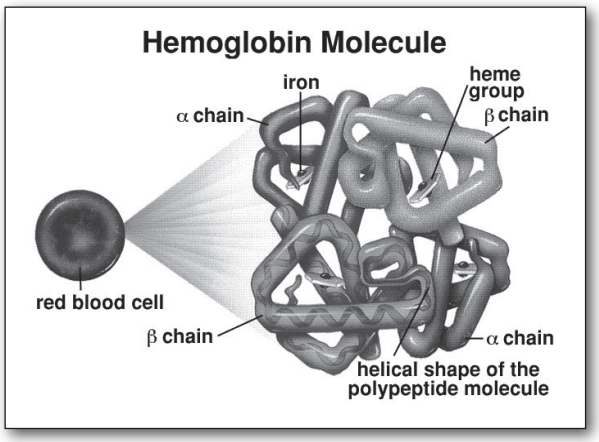
ธาลัสซีเมียอินเตอร์มีเดีย : ปัจจัยทางพันธุศาสตร์ต่อเบต้าธาลัสซีเมีย

Thalassemia Intermedia: Genetics modifier of -thalassemia)

(red blood cell) (He-
moglobin Hb)
(polype-
ptide) (heme group)
2 (alpha chain F chain)
(beta chain -chain) 1
( )
(balance F and -globin chain)



1
( )

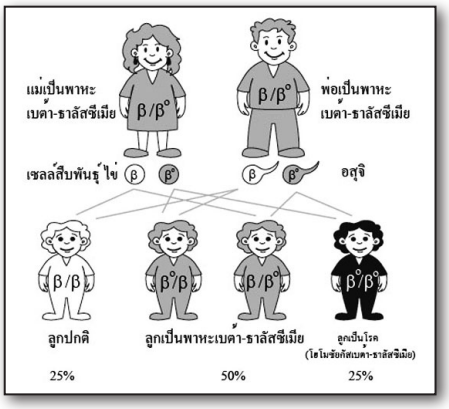
(thalassemia)
thal
2

(F -thalassemia)
(F 0)
1(F -thal1)
(F +) 2
(-thal 2)
(-thalassemia)
(-thal)
(-
thal)

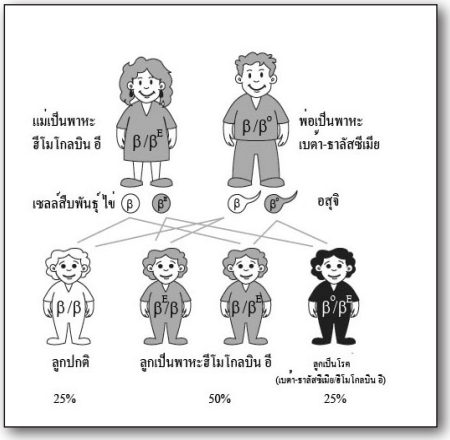
(hemoglobin E Hb E)
(unstable
Hb)

(-thal) (-thal)
(E Hb E)

(autosomal recessive) 2
(alleles)
-
(2)
-
/ (3)



2



3 /

(carrier thalassemia trait) 270 80
(gene mutations)
200
(malarial region)

(transfusion-dependent) (thalassemia trait) (severe
( )
(thalassemia major) (homozygous beta-thalassemia) / (/) (2) 6
20

2
8-10 /
( )
1. /
(/ / /)
2. /
(/ /)

( ) CBC
MCV (mean cell volume)
63.1± 3.4 (femtoliter, fL)
MCV ( ) 69.3 ± 5.6 fL
Hb type
( ) Hb A_2 3.5 - 5.5%
(Hb A_2 < 3.5%)
( )

(genetic counseling)
( )
Codons 41/42 (-TTCT), 17 (A-T), 71/72 (+A), 35 (C-A)
( ) -28 ATA (A-G) Codon 19 (A-G)

(co-inheritance of F-thalassemia)
(Hereditary presistense of hemoglobin F, HPFH)

1

1

- ( )	< 2	> 2
- (/ . .)	6-7	8-10
- -		-
- (%)	>50	10-50
- Hb A_2		
-		
-		
- (HPFH)		
- -		
- XMN1 Polymorphism*		

* ( )

/ (/ / /)
/ (/ /)

1. Thein SL. Genetic modifiers of beta-thalassemia. Haematologica. 2005 May;90(5):649-60.
2. Cappellini MD, Cohen A. Eleftherion A, Piga A, Porter A and Taher A. Guidelines for the Clinical Management of thalassemia. 2nd edition;Nicosia-Cypus:TeamupCreationsLtd, 2007 P 121-31.