

กรมวิทยาศาสตร์การแพทย์ กระทรวงสาธารณสุข

T-Phases of Translational Health Research

- T0: identifies opportunities and approaches to health problems
- T1: move basic discoveries to health applications
- T2: test new therapies (assays) in controlled settings
- T3: implement the guidelines (verify assays) in real-world settings



Diagnostic assays:

- Specificity
- Sensitivity
- Comprehensive
- Cost-effective
- Time-consuming
- Labor requirement
- Equipment/Infrastructure

Infectious Disease:

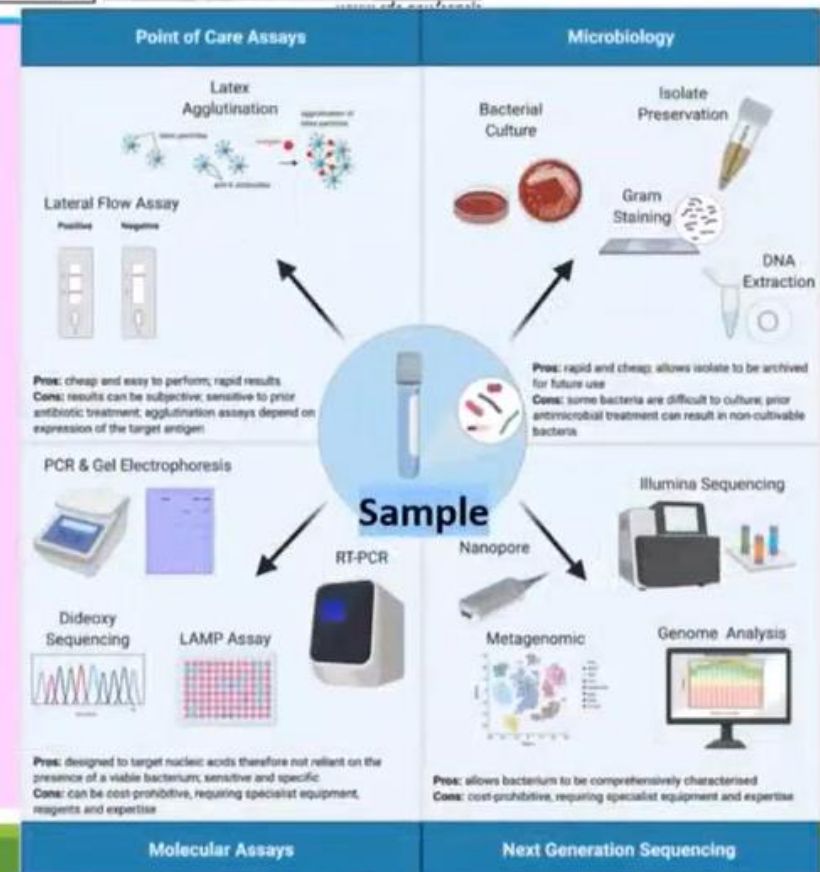
- accurate pathogen identification
- antimicrobial resistance info.

sepsis

a major public health concern with particular high medical cost
a life-threatening organ dysfunction caused by a dysregulated host response to infection

| | | |
|--------------------------|---|---|
| THAI SEPSIS STATS | Each year, at least | Nearly |
| | 175,000 adults in Thailand develop sepsis. | 45,000 Thai die as a result of sepsis each year. |

- Sepsis has specific signs and symptoms.
- Rapid and accurate diagnosis is needed for appropriate treatment.
- Most sepsis is caused by bacterial infections, but it can also be caused by viral infections.



Human Genetic Disease: Thalassemia

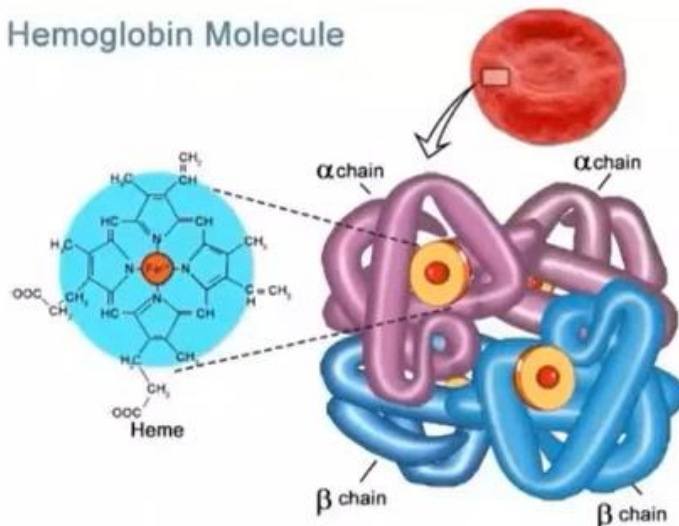
THALASSEMIA IN THAILAND

| | |
|--|-----------|
| α -Thalassemia (α -thal 1 and α -thal 2) | 20 - 30 % |
| Hb Constant Spring (α -thal 2 like effect) | 1 - 8 % |
| β -Thalassemia | 3 - 9 % |
| Hemoglobin E | 10 - 53 % |

Total number of thalassemic patients and the number of births per year (total births = 1 million/yr)

| Disease | Couple at Risk per year | Birth per year | Living patient |
|---------------------------------|-------------------------|----------------|----------------|
| Homozygous β -Thalassemia | 2,500 | 625 | 6,250 |
| β -Thalassemia/Hb E | 13,000 | 3,250 | 97,500 |
| Hb Bart's Hydrops Fetalis | 5,000 | 1,250 | 0 |
| Hb H Disease | 28,000 | 7,000 | 420,000 |
| TOTAL | 48,500 | 12,125 | 523,750 |

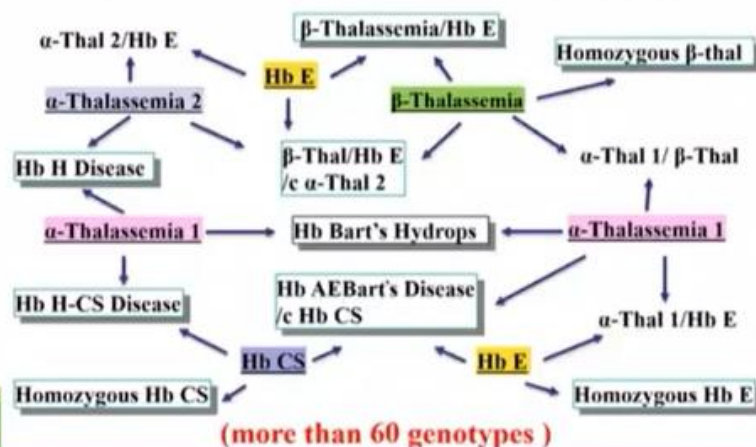
Hemoglobin Molecule



Number of Variants Interactions

- ✓ 1 Variant (Carrier)
- ✓ 2 Variants
- ✓ 3 Variants
- ✓ 4 Variants

Thalassemia and Hb Variants Interactions



A Database of Human Hemoglobin Variants and Thalassemia mutations

Summaries of mutation categories

| Query | Count of results |
|--|------------------|
| Total entries in database | 1851 |
| Total hemoglobin variant entries | 1412 |
| Total thalassemia entries | 536 |
| Total entries in both variant and thalassemia categories | 69 |
| Variants with compound variants | 315 |

Types of Variants

- ✓ Point mutation
 - All nucleotide substitution
 - All position on gene
- ✓ Small insertion/deletion
 - 1, 2, 3, 4, ... nucleotides
 - 1x, 2x, 3x, ... nucleotides
- ✓ Large deletion
 - 1xx, 2xx, 3xx, ... nucleotides
 - 1K, 2K, 3K, ... nucleotides
 - >10K, ... nucleotides

สำนักงานหลักประกันสุขภาพแห่งชาติ (สปสช.)

อัตราค่าใช้จ่ายการตรวจยืนยันทางห้องปฏิบัติการ

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|-----------------------|-----------|
| Hemoglobin typing | 270 บาท |
| Alpha - thalassemia 1 | 800 บาท |
| Beta - thalassemia | 3,000 บาท |