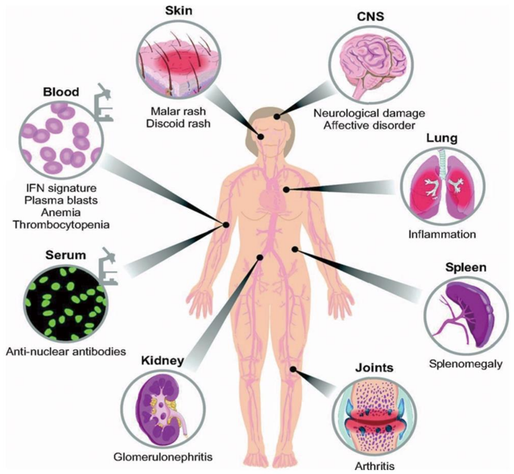
Systemic Lupus Erythematosus(SLE) Gene Methylation Detection Kit (MS-HRM Method)

Disease profile

Systemic Lupus Erythematosus (SLE) is a chronic autoimmune disease with heterogeneous clinical manifestations ranging from mild cutaneous disease to catastrophic organ failure and obstetrical complications. Young women are disproportionately affected by SLE, with a greater prevalence and incidence of this disease in certain ethnic populations such as Black, Asian and Hispanic populations. Reports from the past 5 years on the incidence and prevalence of SLE have shown considerable variation across global regions and even within subpopulations . These differences are probably attributable to true variation, but also to differences in study design and case definition. Unfortunately, SLE is one of the leading causes of death in young women . In a meta-analysis of >26,000 female patients with SLE in the USA, the all-cause mortality was 2.6-fold higher than that of the general population, with a standardized mortality ratio (SMR) of more than 2 for cardiovascular disease and SMRs of almost 5 for infection and renal disease .

Hazards of SLE disease

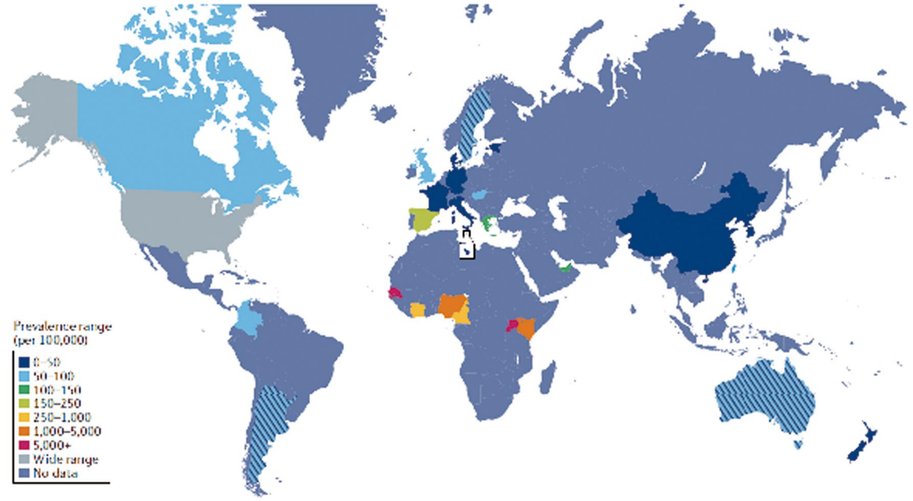
Lupus erythematosus : A Serious Hazard of Human Health



- Multi-organ involvement
- Easy to Mis- or misseddiagnosis
- Difficult-to-cure
- High Death Risk

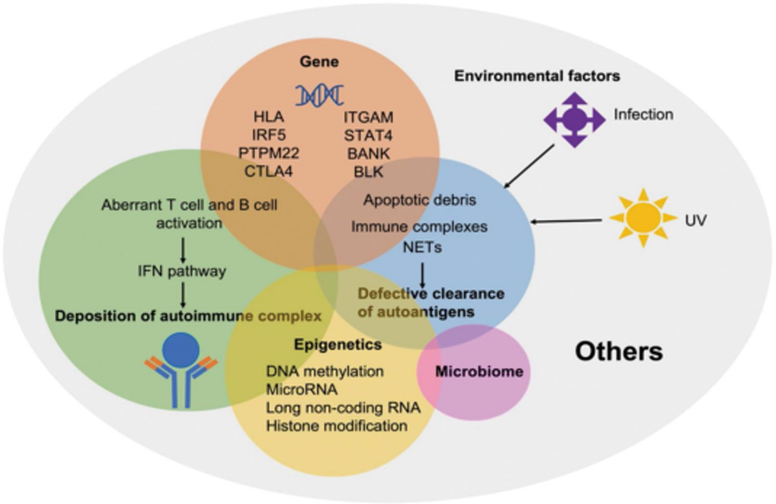
Overview of the Pathogenesis of Systemic Lupus Erythematosus. Genetic, environmental, hormonal, epigenetic, and immunoregulatory factors act either sequentially or simultaneously on the immune system. The action of pathogenic factors results in the generation of autoantibodies, immune complexes, autoreactive or inflammatory T cells, and inflammatory cytokines that may initiate and amplify inflammation and damage to various organs. The target organ affected may be further damaged by local factors.

Global incidence estimates for SLE



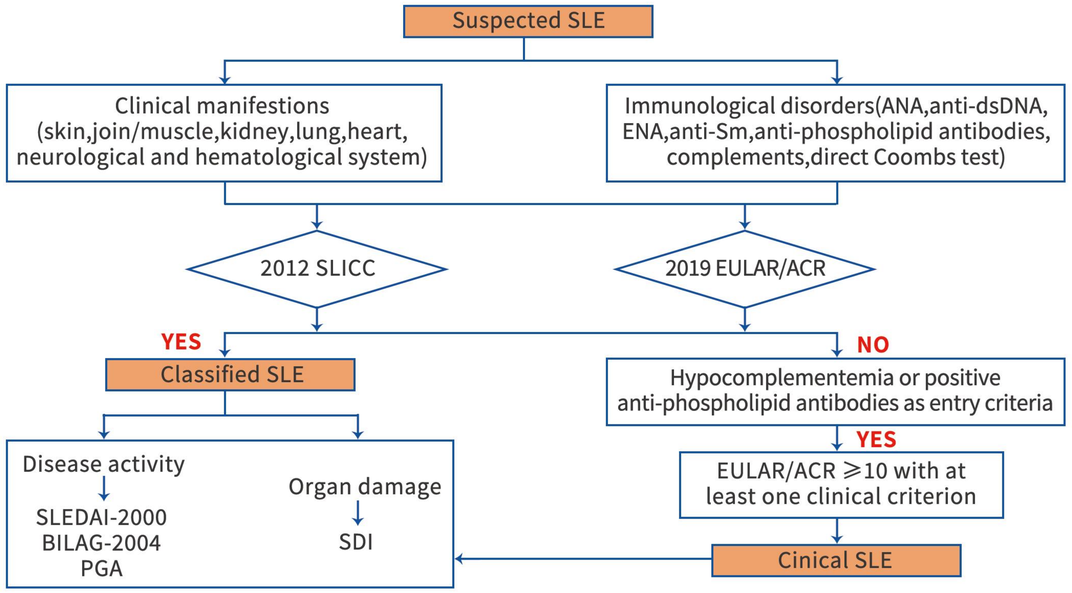
Overview of the Pathogenesis of SLE. Genetic, environmental, hormonal, epigenetic, and immunoregulatory factors act either sequentially or simultaneously on the immune system. The action of pathogenic factors results in the generation of autoantibodies, immune complexes, autoreactive or inflammatory T cells, and inflammatory cytokines that may initiate and amplify inflammation and damage to various organs. The target organ affected may be further damaged by local factors.

Mechanisms of SLE pathogenesis



The pathogenesis of SLE involves a complex interaction of factors including genetic predisposition, epigenetic dysregulation, defective clearance of autoantigens,deposition of autoimmune complex, and dysbiosis of microbiota. Environmental risks such as ultraviolet light (UV) and infections accelerate the production of apoptotic debris and subsequently activate the autoinflammatory cascade in multiple ways.

Classification systems and evaluation instruments for SLE



The present problems in clinics and diagnosis of SLE

① High clinical heterogeneity. lack of highly sensitive and specific marker for the diagnosis of SLE

② Difficult to make precise diagnosis

③ Easy to result in Mis- or missed diagnosis

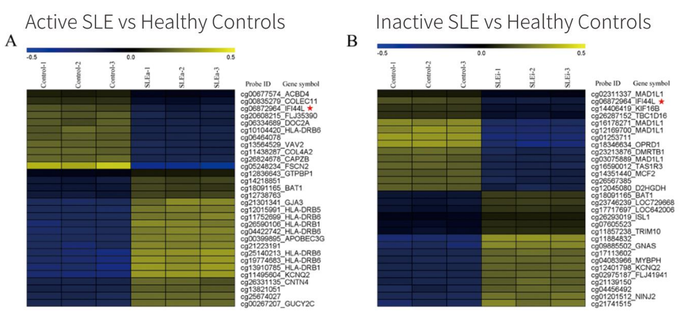
④ Time-consuming : several months or more than one year

⑤ No gene diagnositical method in use

Primary laboratory testing Indicators					
ANA:	specificity ： 65% Sensitivity ： 100%	Anti-sm antibody :	specificity ： 99% Sensitivity ： 25-40%	Anti dsDNA:	specificity ： 94% Sensitivity ： 40-50%

IFI44L gene methylation biomarker-its screen/validation and clincal application for SLE

DNA methylation microarray was used to screen differential DNA methylation sites in peripheral blood cells of SLE



The CG locus of IFI44L gene promoter was found to be significantly hypomethylated in peripheral blood DNA of SLE patients in both active and inactive stages.

Table 1 Baseline characteristics of all subjects*

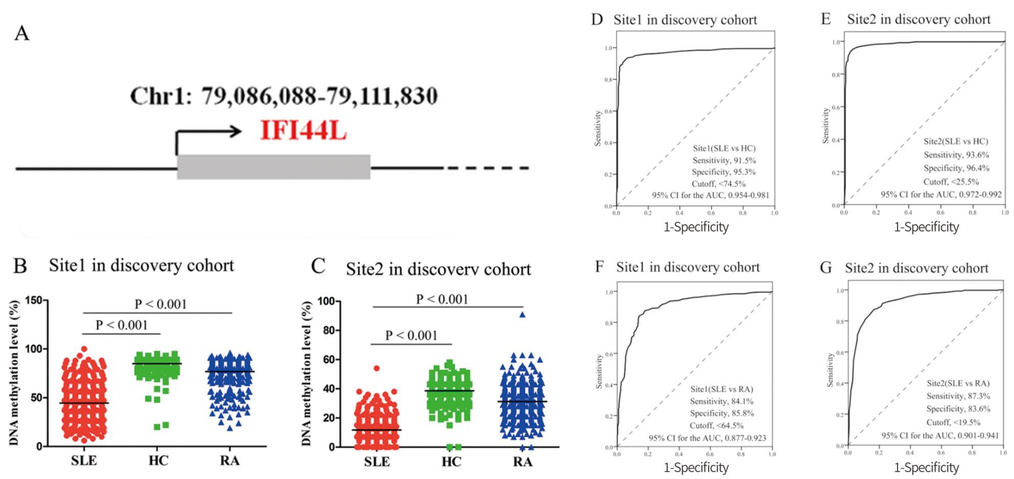
Characteristics	Discovery cohort (China)			Validation cohort 1 (China)				Validation cohort 2 (Europe)	
	Patients with SLE (n=377)	HCs (n=358)	Patients with RA (n=353)	Patients with SLE (n=529)	HCs (n=569)	Patients with RA (n=429)	Patients with pSS (n=199)	Patients with SLE (n=615)	HCs (n=781)
Age (Mean±SD)-years	35.0±12.3	41.1±11.8	48.9±10.9	35.0±12.4	37.7±11.1	48.3±11.5	49.0±13.7	42.7±14.1	50.6±19.4
Sex (%)									
Female	91.7	91.9	91.5	92.1	91.9	92.1	91.0	92.6	57.6

*All subjects included in the Discovery Cohort and Validation Cohort 1 were of Asian descent, and all subjects included in the Validation Cohort 2 were of European descent.
HCs, healthy controls; pSS, primary Sjogren's syndrome; RA, rheumatoid arthritis; SLE, systemic lupus erythematosus.

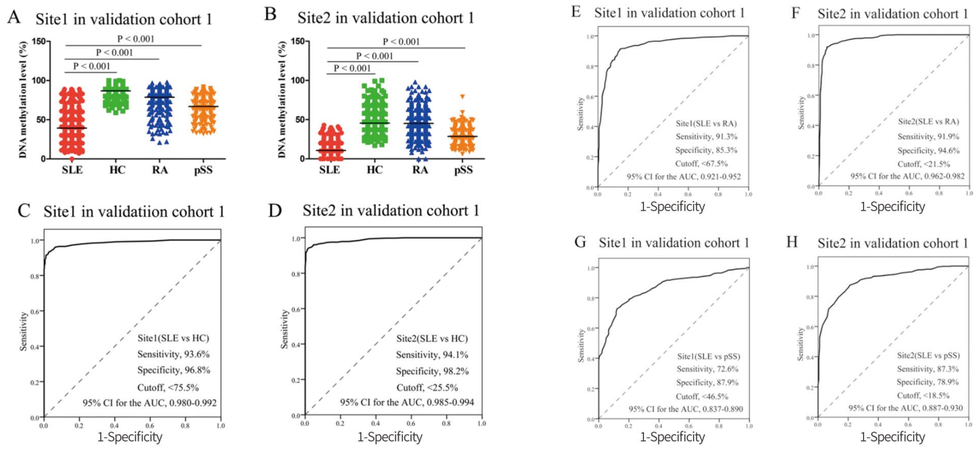
Zhao M, *et al*. *Ann Rheum Dis* 2016;**75**:1998–2006. doi:10.1136/annrheumdis-2015-208410

The differences in the methylation level of CG site in the promoter region of IFI44L gene and the specificity and sensitivity of SLE diagnosis were verified by pyrosequencing

Discovery Cohort 1 （The Chinese population 1）
377 SLE patients, 358 Healthy Controls, 353 RA patients



Validation Cohort 1 （The Chinese population 2）
529 SLE patients, 569 Healthy Controls, 429 RA patients, 199 pSS patients



Systemic Lupus Erythematosus(SLE) IFI44L Gene Methylation Biomarker Detection Kit

(MS-HRM Test)

The SLE gene methylation detection kit utilizes the method of Methylation-Sensitive High Resolution Melting (MS-HRM) technology, to detect the IFI44L biomarker as a way of diagnosis of SLE disease.

Using methylation-specific primers, the target fragments to be detected were amplified by qPCR to obtain high-resolution melting curves (HRM), which were analyzed in comparison with reference curves to determine the methylation level of the promoter region of the IFI44L gene in the PBMC samples, and were used for the genetic diagnosis of SLE and the Clinical assessment of disease.

Features

- Early diagnosis
- Differential diagnosis
- Monitoring prognosis
- Medication guidance
- High sensitivity
- High specificity
- Low cost

Workflow

200ul EDTA anti-coagulation whole blood

Automated Nucleic Acid Extraction

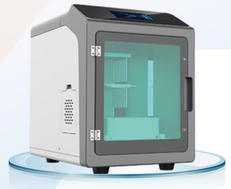
Bisulfite conversion

Real-time quantitative PCR amplification and HRM detection

Result interpretation and analysis

Results report

Instruments



Automatic Nucleic Acid Extractor (48T)

Automatic DNA Extraction and Methylation instrument

Automatic Nucleic Acid Extractor (96T)

Order Information

Cat.No.	Product	Packing Size	Methods	Qualification
MD-01/02/03/04	Blood collection card	1/10/20/40 tests	Fingertip-blood	CE / NMPA
MD-05	Diagnostic Kit For the Detection of IFI44L Gene Methylation of Systemic Lupus Erythematosus(SLE)(MS-HRM Method)	40 tests	MS-HRM Test	CE / NMPA
MD-15	Nucleic Acid Extraction and Purification Kit	64 tests	Magnetic	CE / NMPA
MD-19	Nucleic Acid Extraction and Purification Kit	100 tests	Column	CE / NMPA
MD-53	DNA Methylation Conversion Kit	200 tests	Column	CE / NMPA
MD-50/52	Nucleic Acid Extraction and Methylation Conversion Kit	32/8 tests	Magnetic	CE / NMPA
MD-001B/C	Automatic Nucleic Acid Extraction and Purification Instrument	SET-48/96	Magnetic	CE / NMPA
MD-002A/B	Automatic DNA Extraction and Methylation instrument	SEP-8/32	Magnetic	CE / NMPA

Shenzhen Sciarray Biotech Co.,Ltd.

Add: FL 2402-03, TowerB, Shenye U Center, Zhoushi Rd, Hangcheng Street, 518128, Baoan District, Shenzhen, Guangdong, P.R.China

Tel : +86 755-26625938 +86 755-26623699

Fax: +86 755-26625380

E-mail: services@sciarray.com **Website:** www.sciarray.com/en

Scan QR Code