MEDGENOME CAP ACCREDITED COLLEGE of AMERICAN PATHOLOGISTS

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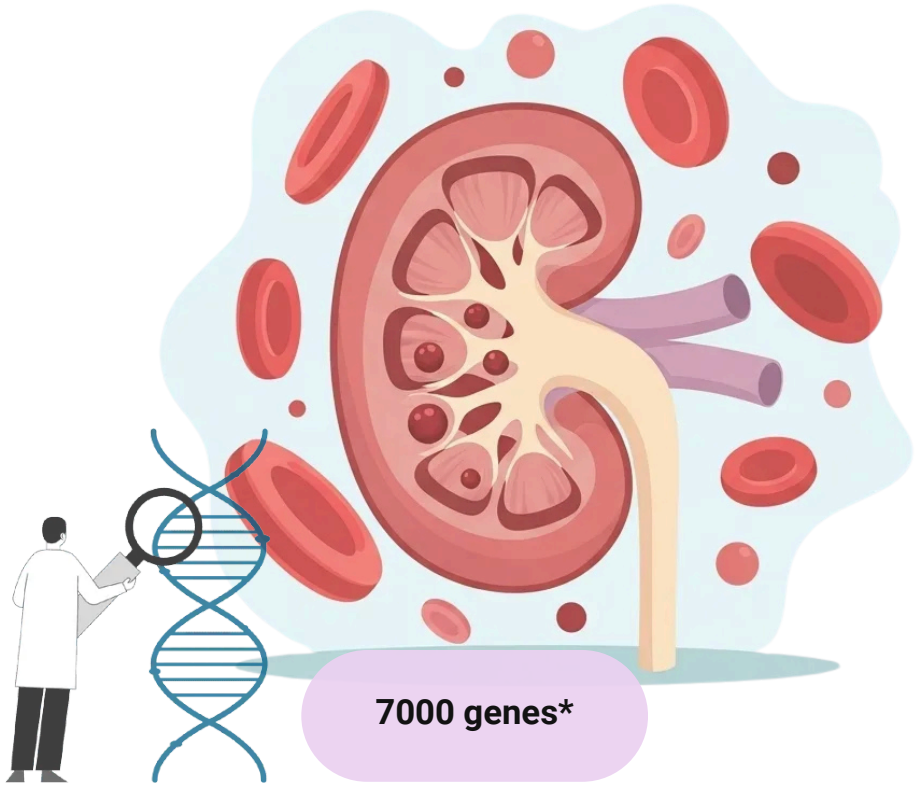
Clinical Exome Sequencing

Hemolytic Uremic Syndrome

Next Generation Sequencing based test to detect genomic biomarker



What is Clinical Exome Sequencing (CES)?

- Analyses **7000 clinically relevant genes**
- Targets **protein-coding regions (exons, ~1% of DNA)**
- Detects **variants** linked to **rare & inherited diseases**
- Can include **targeted analysis** (e.g., **aHUS genes**)

Key Features of CES

Covers ~85% of disease-causing variants

Assesses ~95% of clinically relevant genes

High-depth sequencing → accurate detection

Replace multiple single-gene tests

About Atypical Hemolytic Uremic Syndrome (aHUS)

Rare disease due to compement pathway dysregulation

Causes thrombotic microangiopathy (TMA)

Primarily affcets the kidneys

Genetics in aHUS

- Often linked to **mutations** in
  - **CFH, CFI, CD46, C3, CFB**
- Causes **complement overactivation** → **endothelial damage** → **microthrombi**
- **CES** detects **pathogenic variants** (**diagnostic yield ~40-60%**)
- Helps **confirm diagnosis** and **guide management**

Why Use CES in aHUS?

Genetically complex with multiple genes involved

High heterogeneity → single-gene tests insufficient

Symptoms overlap with other diseases → difficult diagnosis

CES enables fast, comprehensive genetic analysis

How CES Works

1. Sequences coding regions of genes
2. Identifies many variants → filters clinically relevant ones
3. Interpretation based on
   - Clinical presentation
   - Family history
   - Known disease genes

How CES Helps in aHUS

Detects disease-causing variants

Confrims genetic cause of aHUS

Differentiates from other thrombotic microangiopathies

When to Consider testing

Suspected aHUS

Unclear TMA diagnosis

Family history of kidney disease or aHUS

Testing strategy

Clinical Exome Sequencing

Covers 7000 genes

Targeted filtering for aHUS genes

Improves diagnostic efficiency

Clinical value

- Enables **accurate diagnosis** (↑ **yield ~50%**)
- Guides **treatment** (e.g., **complement-targted therapy**)
- Supports **clinical decision-making** & **reanalysis**
- Assists **family risk assessment** & **genetic counselling**

Test Details

Test Code	**MGM1589**
Test Name	**[COMBO] Clinical Exome Sequencing & Hemolytic Uremic Syndrome (CFH, CFHR1, CFHR2, CFHR3 & CFHR5) Deletion/Duplication Analysis (NGS, MLPA)**
TAT	19 days
Sample Type	Peripheral Blood in EDTA (Purple Top)/20-25°C, Direct DNA/1 ug/20-25°C
Storage Condition	CAP, NABL

Exome Sequencing Options

Parameter	Clinical Exome	Whole Exome
Genes	7,000+	20,000+
Design	29.2 Mb	43.2 Mb
On-target	20 Mb	36.5 Mb
Mitochondrial Genes	Optional	Optional
Variant Detection (SNVs, InDels & CNVs*)	Yes	Yes
Reported Pathogenic Variants (+Non-coding)	HGMD* / ClinVar#	HGMD / ClinVar
Reported Pathogenic Variants in SAS-ATLAS$	Yes	NA
Additional Probes for CNV	Yes	No
Sensitivity (NA12878)	~99% (SNVs) >97% (InDels)	~99% (SNVs) >97% (InDels)
Validation Concordance	100%	100%

Our Customers: Hospitals & Specialist Clinics

Hemolytic uremic syndrome (CFH, CFHR1, CFHR2, CFHR3 & CFHR5) depletion duplication analysis by MLPA

Dr Caroline Eng

Hospital Tuanku Ja'afar Seremban

Dr Chan Ling

Hospital Duches of Kent, Sandakan Sabah

MedGenome

MedGenome is a global leader in genetic testing, genomics research, and drug discovery, providing advanced genomic solutions for disease diagnosis & health management. It offers over 1,300 genetic tests & aims to make high-quality, affordable testing widely accessible worldwide.

Witty Charman CoTS/ Medical

Witty Charman CoTS focuses on medical technology, especially forensic pathology and forensic science-related products & services. It is also expanding into diabetes care and digital pathology. It was founded with a vision to improve healthcare through affordable, innovative & compassionate solutions for patients of all ages.

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