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# 香港中文大學醫院 心血管精準醫療門診

## CUHK Medical Centre Cardiovascular Precision Medicine Clinic



香港中文大學醫院  
**CUHK**  
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## 什麼是心血管精準醫學？ What is Cardiovascular Precision Medicine?

心血管精準醫學利用遺傳學或基因組、表觀遺傳學和代謝組學（“omic”）的信息，使您在香港中文大學醫院接受優質的護理及醫療體驗。技術的進步使評估和治療能夠根據個人的基因組和生活經歷而度身設計。我們亦會為您的家人提供適當的篩查和診斷測試，以及循證調查和治療。

Cardiovascular Precision Medicine is a clinical pathway that takes advantage of genetic or genomic, epigenetic and metabolomic (‘omic’) information to optimise your care and healthcare journey at the CUHK Medical Centre. Technological advances have enabled evaluation and treatment that is custom-tailored to one’s genetic make-up and life exposures. We will also take care of your family and provide appropriate screening and diagnostic tests, as well as current evidence-based investigations and treatments.

## 心血管精準醫學團隊由什麼成員組成？

### What members make up the Cardiovascular Precision Medicine team?

團隊由心臟科醫生、臨床遺傳學家、遺傳諮詢師和跨專科團隊組成，會根據您的病情而作出適當的諮詢。如病情需要涉及介入程序，我們將轉介您到適當的專科醫生跟進。

The team consists of cardiologists, clinical geneticists, genetic counsellors, and a multidisciplinary team. They will provide appropriate consultations based on your condition. If your condition requires intervention procedures, we will refer you to the appropriate specialist(s) for further follow-up.

## 我們的服務 Our Services

### 我們提供的醫療程序 Service that we offer

心血管精準醫學門診除了遺傳或基因組測試外，還提供全面的服務，包括詳細的心臟病評估、影像服務（例如超聲心動圖、冠狀動脈電腦斷層造影（CT）、磁力共振造影（MRI）、核掃描）、運動生理學和壓力測試（例如跑步機、腳踏車測功儀），以及入侵性測試（如必要）。

由於遺傳或基因組評估的信息較複雜並且會隨時間而改變，因此您將會在測試前後與臨床遺傳學家進行多次諮詢。遺傳諮詢將幫助患有遺傳疾病的患者或家庭了解測試的必要性、局限性和好處，以及疾病的性質、遺傳模式和遺傳性質、家族內疾病發生的風險和頻率，以便在家庭計劃中作出決策及預防。心臟科醫生亦可能會進行其他檢查以評估病情，及安排檢測以確認診斷結果，為您制定最合適的治療計劃。

Cardiovascular Precision Medicine offers a comprehensive service beyond genetic or genomic testing. Includes a detailed cardiology assessment, imaging (e.g. echocardiography, Computed Tomography (CT) angiography, Magnetic Resonance Imaging (MRI), nuclear scan), exercise physiology and stress testing (e.g. treadmill, cycle ergometry), and invasive tests if needed.

As information from genetic or genomic assessment is complex and changes over time, it is common to often consult with a clinical geneticist both before and after undergoing the test(s). Genetic counselling will help patients or families with a genetic disease to understand the need, limitation and benefits of testing, as well as the nature, mode and nature of inheritance of the disease, risk and frequency of occurrence within the family, and the means of prevention to make informed decisions in family plans. The cardiologist may perform other examinations to evaluate a condition, arrange tests to confirm a diagnosis as to develop the best treatment plan for you.

## 服務對象 Target Group

### 遺傳性心血管疾病 Inherited cardiovascular diseases

#### 心律失常和離子通道疾病 Arrhythmias and channelopathies

- 布魯蓋達氏症候群 Brugada syndrome
- 兒茶酚胺敏感性多形性室性心動過速 Catecholaminergic polymorphic ventricular tachycardia (CPVT)
- 短QT綜合症 Short QT syndrome (SQTS)
- 長QT綜合症 Long QT syndrome (LQTS)

#### 心肌病 Cardiomyopathy (CMP)

- 致心律失常性右心室心肌病 Arrhythmogenic right ventricular cardiomyopathy (ARVC)
- 擴張型心肌病 Dilated cardiomyopathy (DCM)
- 肥厚型心肌病 Hypertrophic cardiomyopathy (HCM)
- 左心室竇竇化性心肌病 Left ventricular non-compaction cardiomyopathy (LVNC)

#### 先天性結構性心臟病 Congenital structural heart disease

#### 結締組織疾病 Connective tissue disorder

- 埃勒斯－當洛二氏綜合症 Ehlers-Danlos syndrome
- Loey-Dietz 綜合症 Loey-Dietz syndrome
- 馬凡綜合症 Marfan syndrome

#### 遺傳性出血性毛細血管擴張症 Hereditary haemorrhagic telangiectasia

#### 心臟異位和鏡像右位心 Heterotaxy and situs inversus

#### 利德爾綜合症 Liddle syndrome

#### 努南綜合症 Noonan syndrome

#### 肺動脈高血壓 Pulmonary artery hypertension (PAH)

### 增加患心臟病風險的遺傳病

#### Inherited conditions that increase the risk of heart disease

#### 家族性高膽固醇血症 Familial hypercholesterolaemia

#### 遺傳性血色素沉着症 Hereditary haemochromatosis

#### 澱粉樣變性 Amyloidosis

### 相關家族病史 Related family medical history

#### 心源性猝死 Sudden cardiac death

## 基因測試 Genetic Testing

基因測試會從患者採集少量血液或組織樣本，測試樣品細胞中包含的DNA（基因），以檢測當中任何有致病風險的變異或突變。視乎進行哪種基因測試，基因測試報告需時數週。

Genetic testing usually collects a small sample of blood or tissue from the client. DNA (genes) contained in the sample cells can be tested to detect variation or mutation that are at risk of developing any genetic condition. Depending on the type of genetic test performed, the availability of genetic result takes weeks.

### 以疾病為中心的基因組 Disease-focused Gene Panels

#### 全外顯子組測序 Whole Exome Sequencing (WES)

#### 全基因組測序 Whole Genome Sequencing (WGS)