

# Unhiding hypertrophic cardiomyopathy, a call for action

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**Clinical Lead for Cardiomyopathy Services**

**The Royal Brompton and Harefield Hospitals**

**Chairman of the ESC WG on Myocardial and Pericardial Diseases**

**Innovative therapeutic approaches in hypertrophic cardiomyopathy**



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# Declarations

Advisory role for BMS

Advisory role for Tenaya

# What is hypertrophic cardiomyopathy?

Hypertrophied myocytes

Fibrosis

A thick heart (hypertrophied)

Loss of myocyte architecture – disarray

Microvascular ischaemia

## Phenocopies

Fabry  
deposition of lipids

*Enzyme replacement*

Amyloidosis  
deposition of amyloid

*Disease stabilizers*

Hypertension ??

## Genetic aetiology

autosomal dominant pattern

MYH7 , MYBPC3

Frequent private mutations

Pathogenic

incomplete

“Predisposing”

penetrance

co-segregation and linkage analysis

VUS

Family history of HCM increases the possibility of positive gene test

# Pathogenesis

Multiple starting points

Reduced levels of proteins or proteins with structural changes

The interaction between actin – myosin and the  $\text{Ca}^{++}$  metabolism are affected

Gene specific pathway

Non-specific hypertrophy pathway

Epigenetic factors

Post translational protein modifications

Environmental factors

Modifier genes?

# Hypertrophic cardiomyopathy in clinic

## Risk of complications

Low but real and tragic

## Symptoms

?

Non-existing

Mild

Moderate

Severe, debilitating

Adaptation

## Family

Gene testing

Family screening



## Function / Haemodynamics

Left Ventricular Ejection Fraction

Diastolic function

Left Ventricular Outflow Tract Obstruction

# Left Ventricular Outflow Tract Obstruction

Hypertrophied interventricular septum

Abnormal mitral valve

Position of the papillary muscles

Angulation of the septum

Contractility

Loading conditions

pathology of the whole LV

# Non-Obstructive Hypertrophic Cardiomyopathy

## Diagnosis

History

Imaging

Provocation

Invasive

## Management

Specific

Non-specific

Heart failure

## Action points

Early and accurate diagnosis

Risk stratification

Patient-centered evaluation of the symptoms

Efficient management of the patients

Modification/prevention of the disease