

# HCM MAP

Interactive Knowledge Resource About Molecular  
Mechanisms of Hypertrophic Cardiomyopathy

**Mila Glavaški, Ph.D. student**  
Faculty of Medicine,  
University of Novi Sad, Serbia

**Prof. Lazar Velicki**  
Faculty of Medicine,  
University of Novi Sad, Serbia

Institute for Cardiovascular Diseases of  
Vojvodina, Serbia

# DISCLOSURE

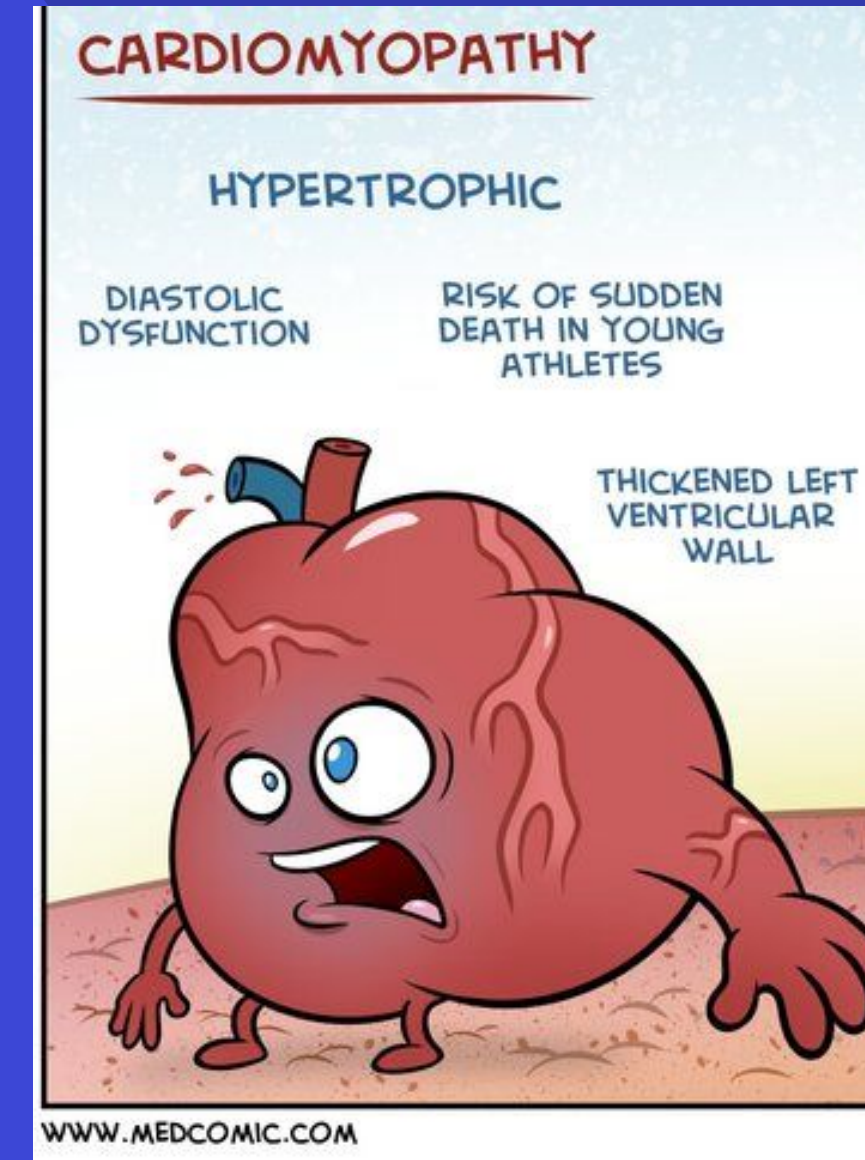
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# HYPERTROPHIC CARDIOMYOPATHY (HCM)

- The most common genetic cardiac disease (1:500)
- Left ventricular hypertrophy unexplained by other causes
- Variable expression: ranging from asymptomatic to heart failure or sudden cardiac death
- Main cause of sudden cardiac death in athletes and young people



# HCM: GENETIC BACKGROUND

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- **Autosomal dominant**  
First degree relatives have 50% chances of carrying the same mutation
- **Very complex and not completely understood**
- **Main cause: mutations in 8 sarcomeric genes**  
More than 40 genes associated with HCM
- **For 40-60% of cases, genetic cause is not known**



# **HCM: GENOTYPE-PHENOTYPE ASSOCIATIONS**

- **HCM genotype-phenotype associations are (also) incompletely understood**
- **Phenotype may differ even within the same family**
- **High variability in disease progression**

# KNOWLEDGE GRAPHS

Many studies have tried to decipher molecular mechanisms underlying HCM.

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That knowledge is dispersed in scientific papers.

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**HCM MAP: A SUMMARY OF ALL  
MAJOR MOLECULAR PATHWAYS  
INVOLVED IN HCM**

# Disease maps

VISUAL AND INTUITIVE REPRESENTATIONS  
OF DISEASE MECHANISMS

# HCM Map

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## CELLDESIGNER

v. 4.4.2

## SBML

(Systems Biology  
Markup Language)  
format

## SBGN

(Systems Biology  
Graphical Notation)  
diagram

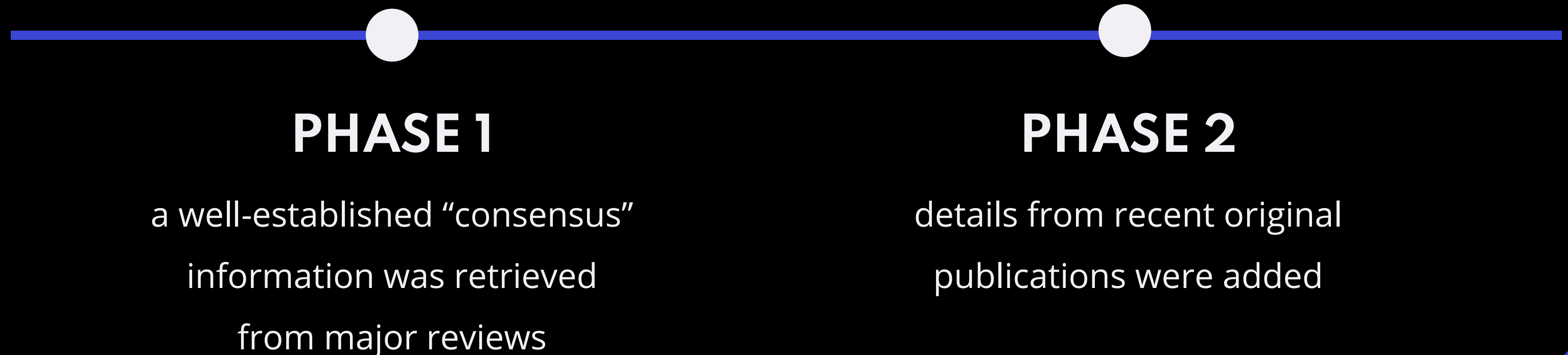
## MINERVA

(Molecular Interaction  
NEtwoRks VisuAlization)  
platform



# HCM MAP

- constructed manually
- molecular interactions were retrieved from PubMed database articles about human HCM



# HCM MAP

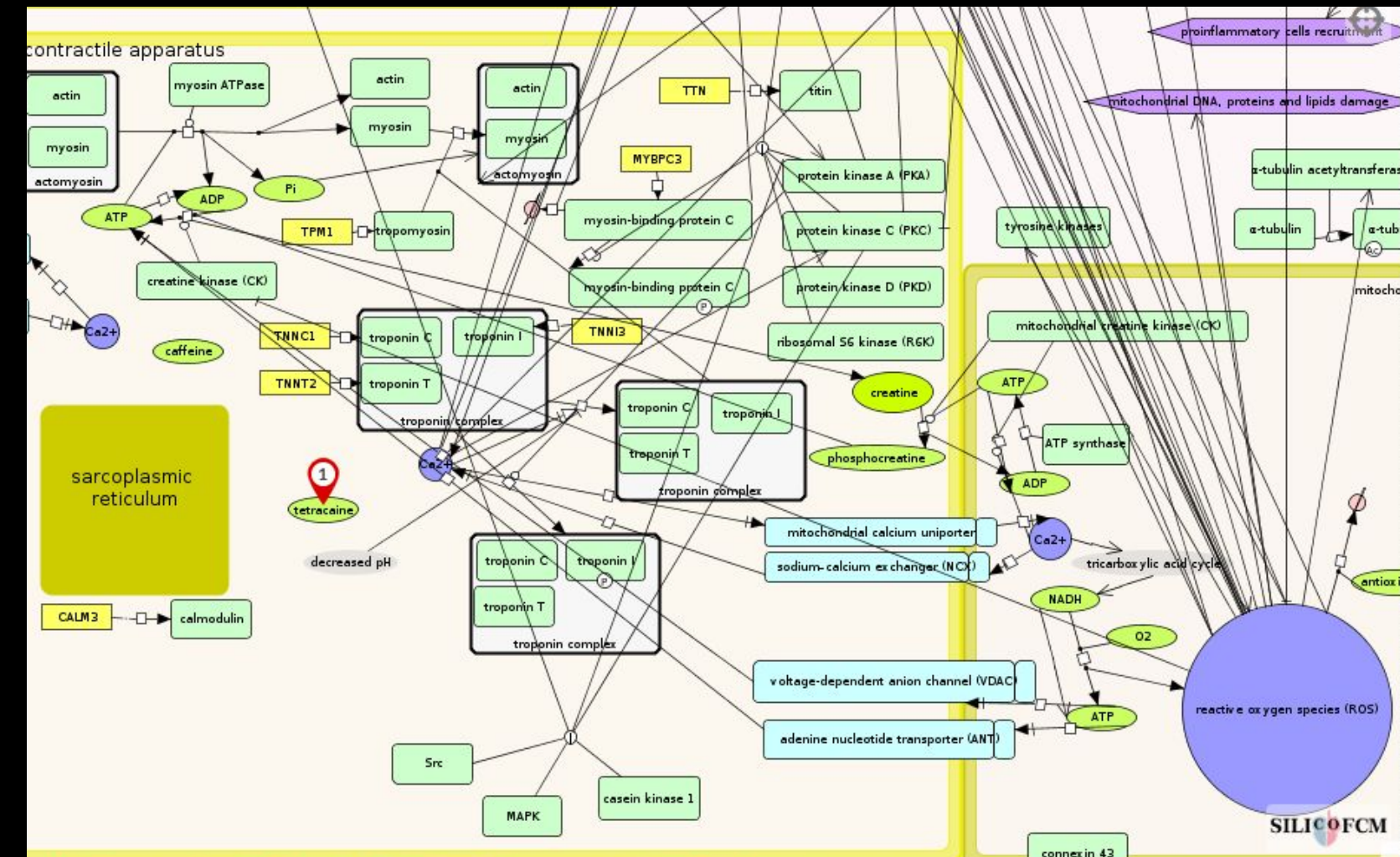
- **INFORMATION ABOUT THE ENTITIES AND LINKS TO DESCRIPTIONS IN BIOMEDICAL DATABASES**

Gene IDs, UniProt accession numbers, PubMed IDs...

- **INTERFACES WITH**  
DrugBank, ChEMBL, CTD, and miRTarBase

- **CAN BE EXPORTED INTO DIFFERENT FORMATS FOR FURTHER ANALYSIS**

PNG, PDF; elements, reaction types, columns...



## **HCM MAP: BODY OF KNOWLEDGE AVAILABLE TODAY**

SOME INTERACTIONS HAVE YET TO BE IDENTIFIED

## **EACH COMPONENT CAN BE COMMENTED**

THE MAP WILL BE CONTINUOUSLY UPDATED AND IMPROVED  
INVOLVING THE COMMUNITY OF HCM SIGNALING EXPERTS

# HCM Map

available at <https://silicofcm.eu/interactive-map/>

- **THE ONLY PUBLICLY AVAILABLE KNOWLEDGE RESOURCE DEDICATED TO HCM**
- **A BASIS FOR *IN SILICO* EXPLORATION**
- **A PLATFORM FOR SHARING AND GATHERING MOLECULAR MECHANISMS OF HCM**

**THANK YOU  
FOR  
YOUR ATTENTION**



