



## What is sudden **CARDIAC DEATH?**

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- ◆ Sudden, unexpected death. A loss of heart function.
- ◆ Body's organs can't receive any oxygen due to an irregular heart rhythm.
- ◆ The person becomes unconscious.
- ◆ Without immediate treatment, sudden cardiac arrest can lead to death
- ◆ Responsible for half of all heart disease deaths.

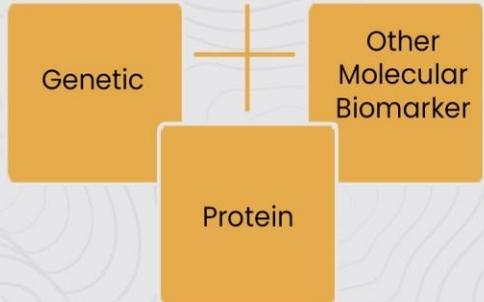
## Criteria for SCD CLINICAL Risks



### Risk factor & Trigger

- ♦ Heart Failure
- ♦ Hypertrophic cardiomyopathy
- ♦ Hipertension
- ♦ Hypercholesterolemia
- ♦ Smoking
- ♦ Diabetes Mellitus
- ♦ Unhealthy Diet
- ♦ Physiological Stress
- ♦ Physical Activity

### Molecular Biomarkers

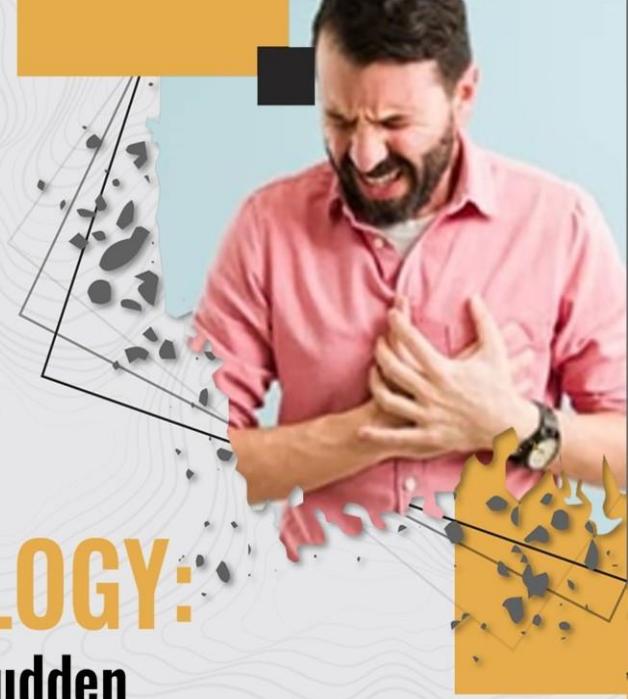


### Symtoms

- ♦ Fainting (losing consciousness).
- ♦ Racing heartbeat.
- ♦ Chest pain.
- ♦ Dizziness.
- ♦ Shortness of breath.
- ♦ Feeling sick to your stomach and throwing up.

### Clinical Laboratory Test

- ♦ ECG (electrocardiogram)
- ♦ Blood tests
- ♦ Echocardiogram
- ♦ Ejection fraction
- ♦ Chest X-ray
- ♦ Nuclear scan
- ♦ Cardiac catheterization



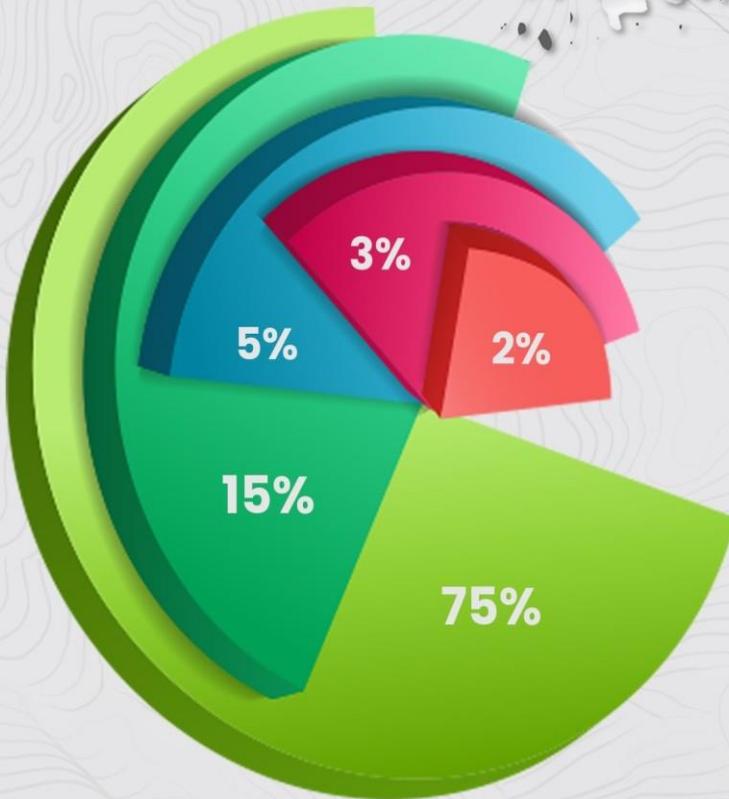
# EPIDEMIOLOGY:

## How common is sudden cardiac death in young people?

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- ◆ Most frequently in adults in their mid-30s to mid-40s.
- ◆ Occurs while playing team sports.
- ◆ It happens more often while running or jogging.
- ◆ leading cause of death in young athletes.
- ◆ About 1 in 50,000 to 1 in 80,000 young athletes die each year.

## SCD ETIOLOGY



■ Coronary Heart Disease

■ Cardiomyopathies (DCM, HCM, ARVC)

■ Inherited arrhythmia syndromes (LQT, BrS, CPVT, ERS)

■ Valvular heart disease

■ Others

# Diseases can cause SCD

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- ◆ Congenital Heart Disease (CHD)
- ◆ Cardiac Channelopathies
- ◆ Long QT Syndrome (LQTS)
- ◆ Short QT Syndrome (SQTS)
- ◆ Brugada Syndrome (BS)
- ◆ Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT)
- ◆ Cardiomyopathy
  - ◆ Hypertrophic Cardiomyopathy (HCM)
  - ◆ Dilated Cardiomyopathy (DCM)
  - ◆ Restrictive Cardiomyopathy (RCM)
  - ◆ Arrhythmogenic Right Ventricular Dysplasia (ARVD)
  - ◆ Left Ventricular Non-Compaction (LVNC)
- ◆ Familial Hypercholesterolemia (FH)



# Who should be screened for Sudden **CARDIAC DEATH?**

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- ◆ Family history of SCD at a young age (under 50 years old)?
- ◆ Already been diagnosed with genetic heart disease and are worried about the implications of this for your children?
- ◆ If your family been diagnosed with myocardiorpathy or channelopathy?



## Why do you need genetic testing for Sudden **CARDIAC DEATH?**

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- ◆ Around 20% of Cardiac cases are diagnosed based on genetics.
- ◆ Improve patient treatment, management, and target surveillance for cardiovascular complications
- ◆ If an individual carries a disease-causing variant associated with a heritable risk to develop cardiomyopathy.
- ◆ Implement an effective treatment plan and offer you support and guidance.



A Genomics lab by Nucleome



# Here is the solution, With **DrSeq SCD®**

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- ◆ Drseq SCD® covers 151 genes and regions associated with cardiovascular conditions
- ◆ Capture a target size of 3 Mb.
- ◆ You will know, through your genetics, whether you are at risk of suffering from genetic sudden cardiac death.
- ◆ Family heart disease affects one in every 400 individuals in the general population and a genetic cause can be found in 50% of cases.



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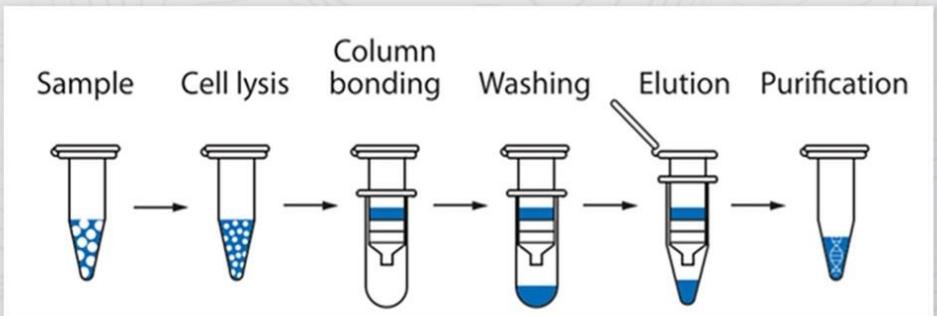
## Genes to be covered in **DRSEQ SCD® PNL**

ABCC9, ACTA2, ACTC1, ACTN2, ADAMTS2, AKAP9, ALPK3, ANK2, ANKRD1, ANO5, ASPH, B4GALT7, BAG3, BGN, CACNA1C, CACNA1D, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CALR3, CASQ2, CAV3, CBS, CCM2, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, CRYAB, CSRP3, CTF1, CTNNA3, DES, DMD, DSC2, DSE, DSG2, DSP, DTNA, EFEMP2, ELN, EMD, EYA4, FBLN5, FBNI, FBN2, FHL1, FHL2, FKBP14, FKTN, FLNA, FLNC, FOXE3, GATA4, GATAD1, GJA5, GLA, GPD1L, HCN4, HTRA1, ILK, JPH2, JUP, KCNA5, KCND3, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, KRIT1, LAMA4, LAMP2, LDB3, LMNA, LMNB2, LOX, MAT2A, MED12, MFAP5, MIB1, MYBPC3, MYH6, MYH7, MYH11, MYL2, MYL3, MYLK, MYLK2, MYOT, MYOZ1, MYOZ2, MYPN, NEBL, NEXN, NOTCH1, NPPA, NSD1, PDLIM3, PKP2, PLN, PLOD1, PRDM16, PRKAG2, PRKG1, RANGRF, RBM20, RYR2, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SCN10A, SGCD, SKI, SLC2A10, SLC8A1, SLMAP, SMAD3, SMAD4, SMAD6, SNTA1, TAZ, TBX20, TCAP, TGFB2, TGFB3, TGFBR1, TGFBR2, TMEM43, TMPO, TNNC1, TNNI3, TNNT2, TNXB, TPM1, TRDN, TRIM63, TRPM4, TTN, TTR, TXNRD2, VCL.



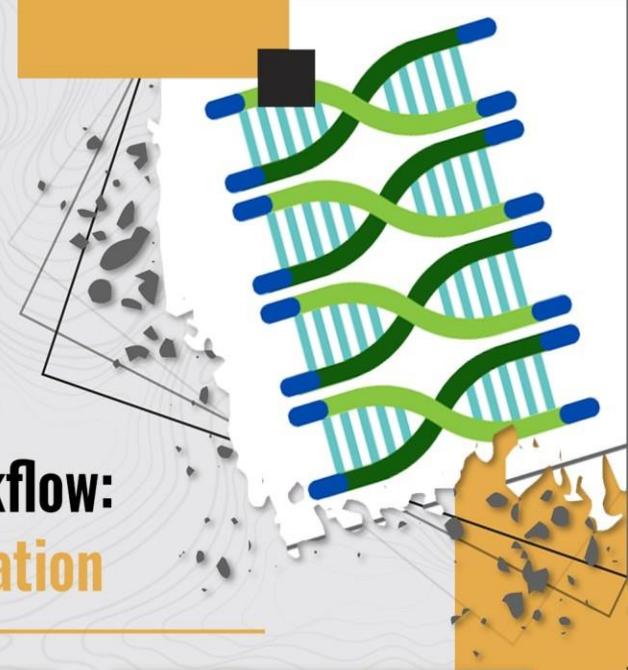
## Work flow of DRSEQ SCD®

### Experimental Workflow: (i) DNA Extraction

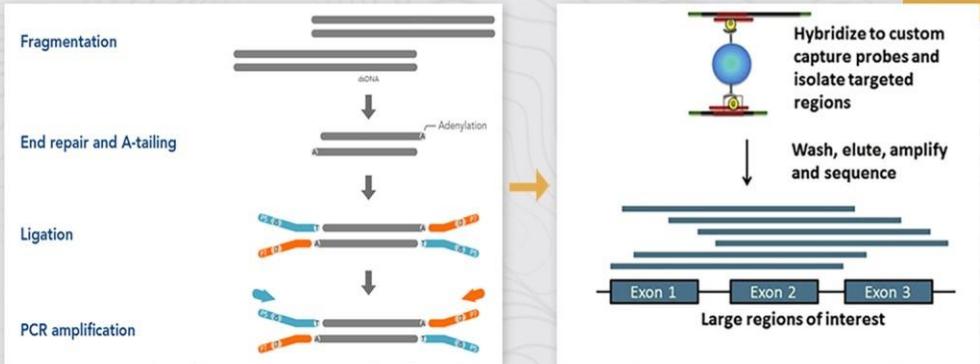


Blood Samples

Genomic DNA from Peripheral Blood

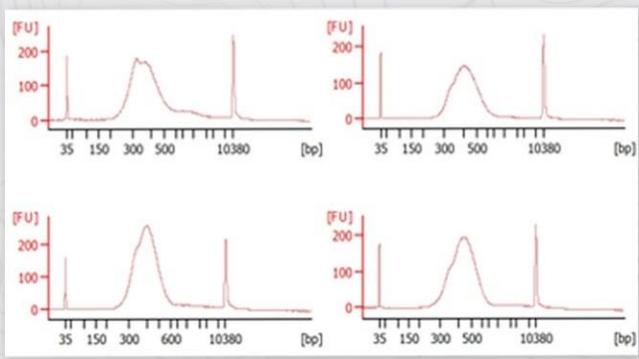


## Experimental Workflow: (ii) Library Preparation



Library Preparation

Hybrid Capture and Amplification

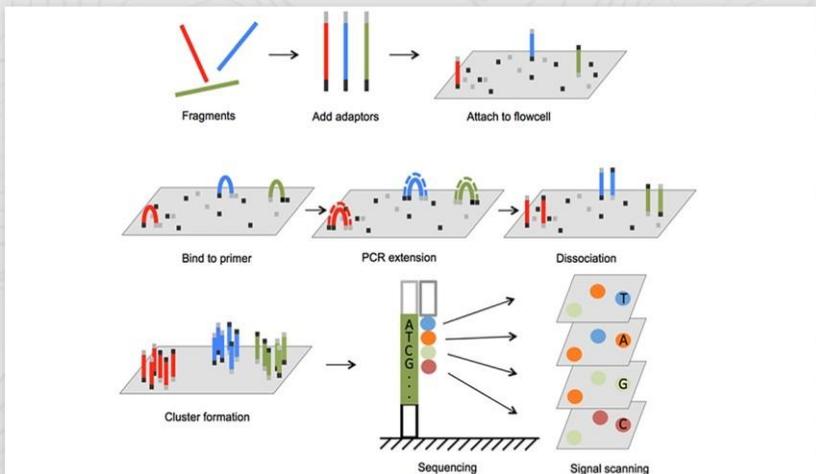


Quantification

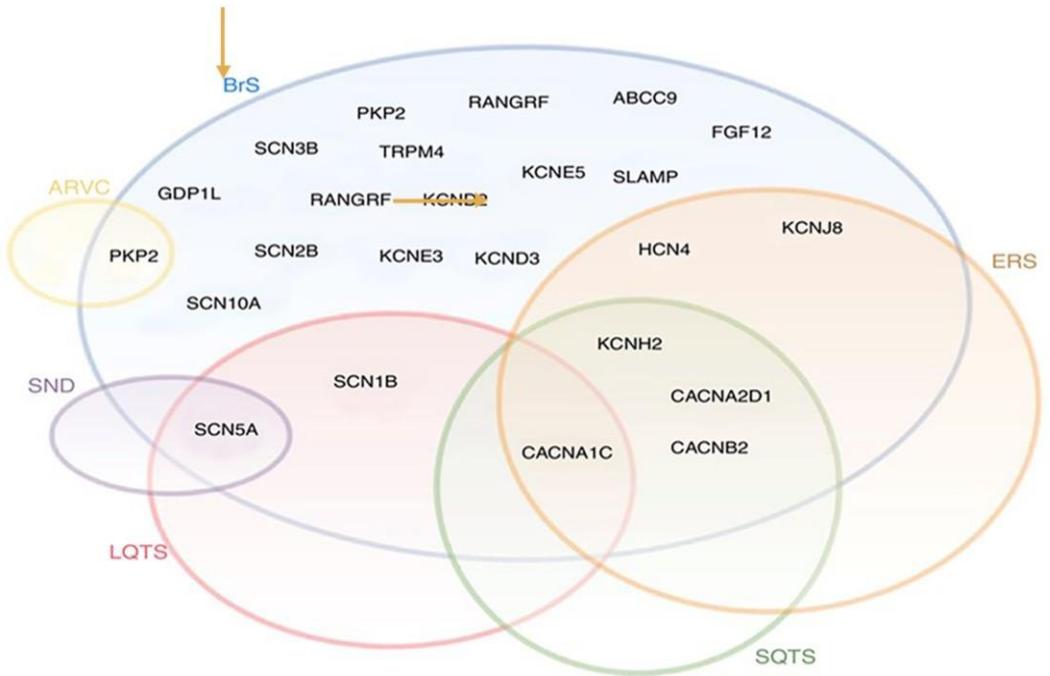
## Experimental Workflow: (iii) Sequencing



Sequenced using Illumina  
NovaSeq 6000



## Genes association with SCD





A Genomics lab by Nucleome

# How to explore **DrSeq SCD® PNL**

**Research  
R&D field**

**Healthcare  
Marketing  
Strategies**

## **Exploration of SCD**

**Awareness  
of the test**

**Hospitals  
and  
clinics**





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