SPGH
Sociedade Portuguesa de Genética Humana

ANOS 25 Virtual ANNUAL MEETING
SOCIEDADE PORTUGUESA DE GENÉTICA HUMANA

18 - 19 NOV 2021

CONTRIBUTION OF *HFE* AND *HPSE* GENES AND METHAEMOGLOBIN REDUCTASE ACTIVITY TO HEART FAILURE

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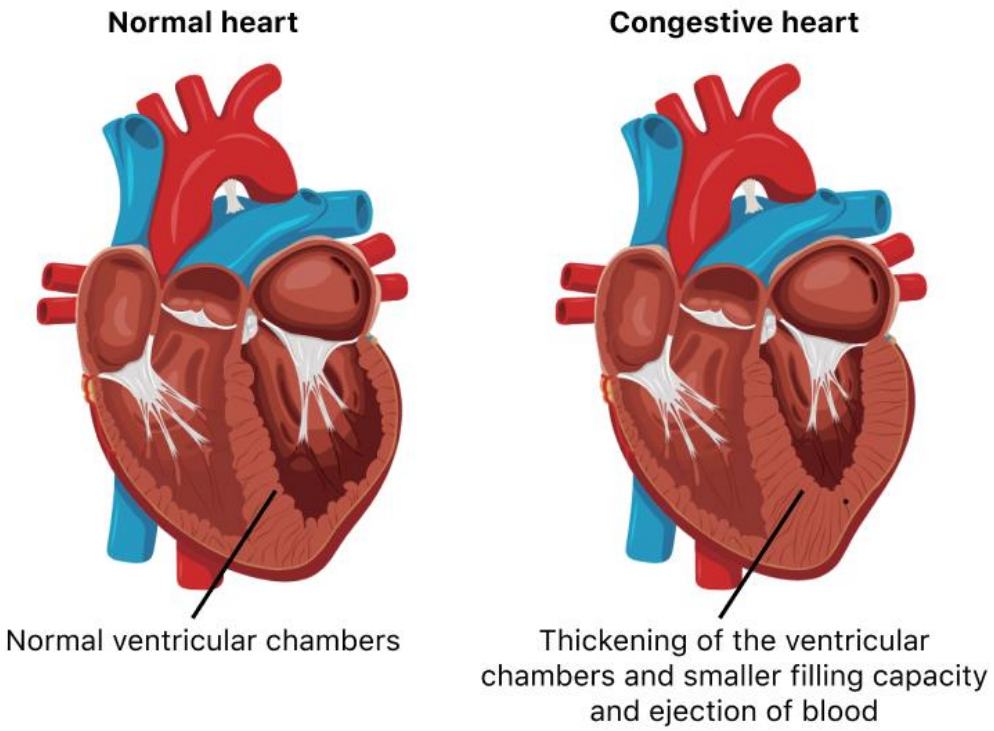
FUNDADO EM 1922

Introduction

Heart Failure (HF):

- Structural anomaly and/or committed cardiac function => inadequate cardiac output unable to meet the metabolic necessities of the organism.



https://www.drugwatch.com

Aim

- Can *HFE* and *HPSE* polymorphisms and methaemoglobin reductase activity influence HF?

Methodology

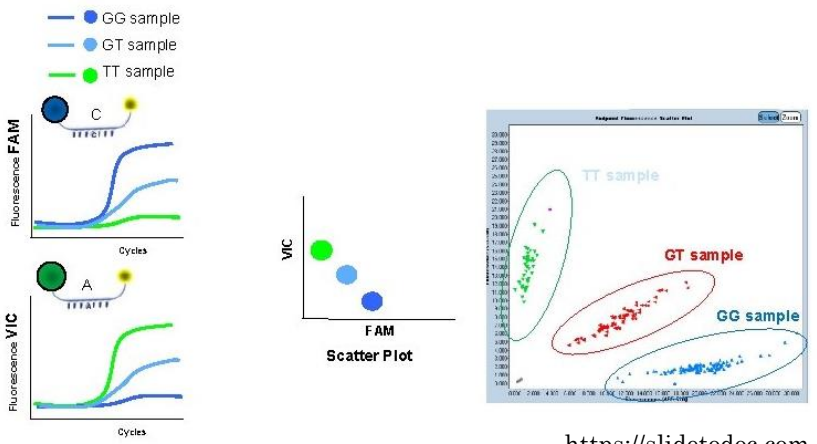
252 DNA samples:
143 (HF patients) +
109 (Controls)

(1) Spectrophotometric analysis

- Methaemoglobin reductase activity

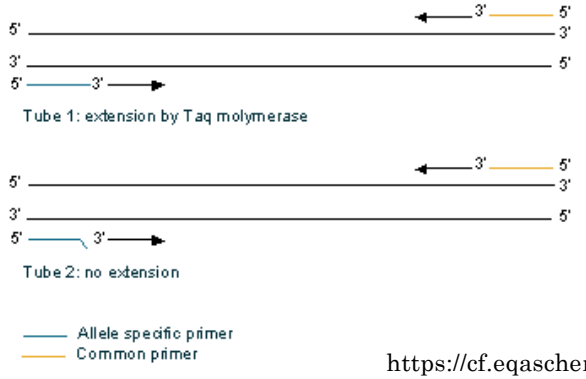
(2) Genotyping

- rs4693608 polymorphism *(HPSE* gene*)* – Endpoint PCR



https://slidetodoc.com

- C282Y and H63D polymorphisms *(HFE* gene*)* – ARMS Multiplex



https://cf.eqascheme.org

Statistical analysis (SPSS)

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Results

Table 1 – Comparison of genotypic distribution of *HFE* and *HPSE* polymorphisms between HF and control populations.

Polymorphism (Gene)	Genotype/ Allele	Controls n (%)	HF Patients n (%)	*p-value*	[OR (95% CI)]	
H63D *(HFE)*	HD	18 (19.8)	22 (27.2)	0.042[1]	2.889[1] (1.041 - 8.018)	Risk
	HH + DD	73 (80.2)	59 (72.8)			
rs4693608 *(HPSE)*	GG	29 (30.5)	23 (20.9)	0.045[2]	0.435[2] (0.193 - 0.982)	Protective
	AA + GA	66 (69.5)	87 (79.1)			
rs4693608 *(HPSE)*	AA + GA	66 (69.5)	87 (79.1)	0.045[2]	2.297[2] (1.018 - 5.179)	Risk
	GG	29 (30.5)	23 (20.9)			

[1]Binary logistic regression, adjusted for age and sex.
[2]Binary logistic regression, adjusted for age.

Table 2 – Comparison of methaemoglobin reductase activity between HF and control populations.

Methaemoglobin reductase activity (µmol/g Hb/min)					
Population	**n**	**Median**	**Minimum**	**Maximum**	***p-value***
Controls	37	20.633	12.418	31.200	0.019[1]
HF Patients	94	15.651	8.370	47.267	

[1]Binary logistic regression, adjusted for age.

Conclusions

HFE, HPSE, methaemoglobin reductase activity: association with HF

Study of iron-related markers

Intravenous iron supplementation

(in some cases)

Better management of HF