FAMILIAL HYPERCHOLESTEROLEMIA (FH) IN CHILDREN AND ADOLESCENTS: OPTIMIZATION OF DETECTION FOR EARLY TREATMENT.

Guilherme Redezuk*,Erica I.L. Gomes,Thiago Quinaglia, Vanessa H. S. Zago, Flávia Silva Mothe,Leonardo Staffa, José Roberto Matos Souza, Andrei C. Sposito, Eliana Cotta de Faria.

Abstract: The Familial Hypercholesterolemia (FH) is the most common co-autosomal dominant inherited condition in man. It is mainly caused by mutations in the following three genes: the encoding: the receiver of low-density lipoprotein LDL-C (most common), or apolipoprotein B100 proprotein convertase subtilisin / kexin type 9 (PCSK-9). The resulting chronic elevations in LDL-C levels entail the development of early atherosclerotic cardiovascular disease. The overall estimated prevalence for heterozygous HF ranges from about 1 / 200-250 1/500 and homozygous FH is 1/1 000 000. The childhood is the ideal period of life to discriminate between dyslipidemias FH and not FH. serum LDL-C≥190 mg / dL (2 measures up to 3 months), or LDL-C≥160 mg / dL with a family history of premature coronary disease or high levels of cholesterol increased basal in a parent or LDL -C≥130 mg / dL with positive genetic diagnosis in families may be used as the phenotypic criteria for diagnosis. It is the aim of this study make the early diagnosis of HF in a population sample of children, adolescents and adults Brazilian city of Campinas / SP.

Key words: *Familial Hypercholesterolemia, Dyslipidemia, Atherosclerosis.*

Introduction

The Familial Hypercholesterolemia (FH) as the most common autosomal condition in man, has its consequences present even in childhood , is indispensable, for the efficient management, early diagnosis of cases.

Consequences : chronic elevations in plasma levels of LDL- cholesterol.

Clinical manifestations of FH :

- Early atherosclerotic cardiovascular disease.
- A** - corneal bow ; **B** - tendon xanthoma

RBM Ago V 71 N 8, pgs.:225-245

Objectives: HF screening in a sample of children and adolescents for early diagnosis and treatment of HF.

Results and Discussion

Métodos: Laboratory results of serum analyzes LDL cholesterol by homogeneous method Direct (Roche) done Were selected from consecutively . The sample was 472 050 individuals attended at all basic healthcare systems of Campinas , SP, in the period 2008 to 2015. We assessed the prevalence in the age range of specific subgroups (1 day to 19 years, 1 day to 1 year-364d, 2 to 11 years and 12 to 19 years) . The criterion of LDL C≥190 mg / dL was used for phenotypic screening, according to the I Brazilian Guidelines for Familial Hypercholesterolemia (FH).

- 382 suspected cases of HF (144 children / adolescents 221), prevalence of 0.003, compared to the world of ~ 0.002
- higher prevalence in females, 12-19 years subgroup: likely resulting from secondary causes of not FH hypercholesterolemia

- there were no statistical differences between sexes in the range of 1d-19years, similar results are found in the literature
- after exclusion of secondary causes, the phenotypic diagnosis FH should be confirmed by genetic and treatment initiated

Chart 1: Prevalence of FH suspects by sex and age

Groups (d/y)*	N	Prevalence** (%)	CI 95%	Frequencies ****(%)
T 1d-19y (N=61267)	382	0,62	0,56 - 0,69	-
M 1d-19y (N=27102)	140	0,52***	0,43 - 0,60	0,23
F 1d-19ay(N=34165)	242	0,71***	0,62 - 0,80	0,39
T 1d-1y"(N=1017)	33	3,24	2,14 - 4,35	0,05
M 1d-1y" (N=390)	8	2,05	0,64 - 3,46	0,79
F 1d-1y"(N=627)	25	3,99	2,46 - 5,52	2,46
T 2-11y(N=21623)	111	0,51	0,42 - 0,61	0,18
M 2-11y (N=10458)	51	0,49	0,35 - 0,52	0,24
F 2-11y(N=11165)	60	0,54	0,40 - 0,67	0,28
T 12-19y(N=36062)	221	0,61	0,53 - 0,69	0,36
M 12-19y (N=15004)	71	0,47***	0,36 - 0,58	0,20
F 12-19y(N=21068)	150	0,71***	0,60 - 0,83	0,42

*d/y= days/years; N= with LDL-C ≥ 190 mg/dL; *** test chi-squared= p ≤ 0,05; **1d-1y =1 day a 1 year-364d ; T= total; M= male gender; F= female gender N= number; CI= mean confidence interval (95%); **** calculated from the nT

Conclusions

The results of this study point to the urgent need for continued FH screening in the population of children and adolescents for their early diagnosis and handling.

Acknowledgements

FAPESP; CNPQ; SUS