

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

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**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 $\geq$ 190 mg / dL (2 measures up to 3 months), or LDL-C $\geq$ 160 mg / dL with a family history of premature coronary disease or high levels of cholesterol increased basal in a parent or LDL -C $\geq$ 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



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**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 $\geq$ 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 **** (%)
<b>T 1d-19y (N=61267)</b>	<b>382</b>	<b>0,62</b>	<b>0,56 - 0,69</b>	<b>-</b>
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
<b>T 1d-1y''(N=1017)</b>	<b>33</b>	<b>3,24</b>	<b>2,14 - 4,35</b>	<b>0,05</b>
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
<b>T 2-11y(N=21623)</b>	<b>111</b>	<b>0,51</b>	<b>0,42 - 0,61</b>	<b>0,18</b>
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
<b>T 12-19y(N=36062)</b>	<b>221</b>	<b>0,61</b>	<b>0,53 - 0,69</b>	<b>0,36</b>
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  $\geq$  190 mg/dL; \*\*\* test chi-squared=  $p \leq 0,05$ ; \*\*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.

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