

Diagnosis and treatment of familial hypercholesterolemia in children – a preliminary report

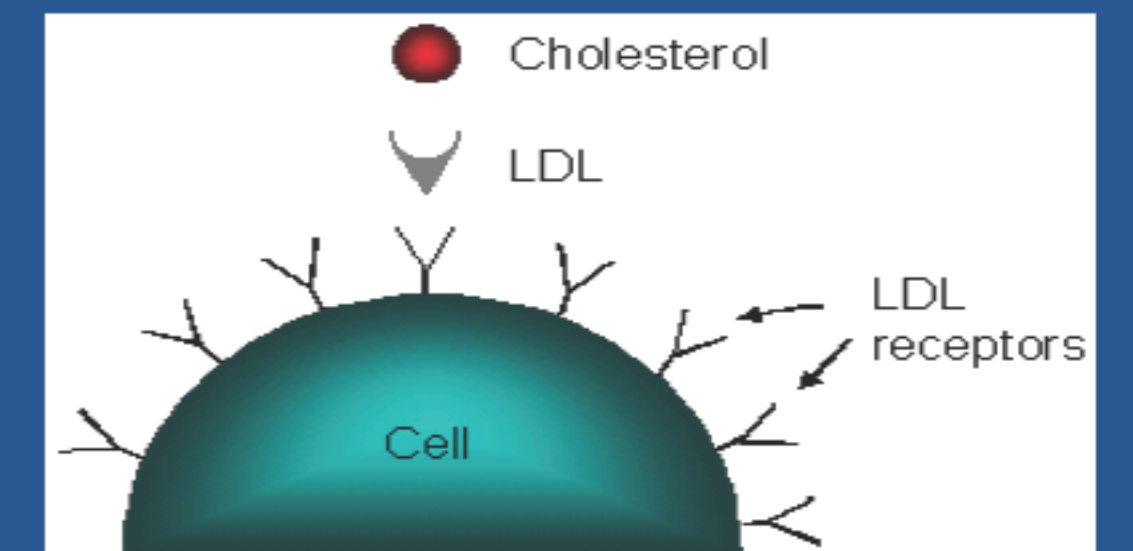
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BACKGROUND

- heterozygous familial hypercholesterolemia (FH) is an autosomal dominant genetic disorder occurring in 1:500 individuals
- patients with FH have a high risk of premature cardiovascular diseases
- effective lipid-lowering therapies are available and it is a chance to extend the life of FH patients

AIMS AND OBJECTIVES

The aim was to analyze the clinical data of children with FH from the Clinic of Pediatrics, Diabetology and Endocrinology and preliminary assesment of the effects of treatment.

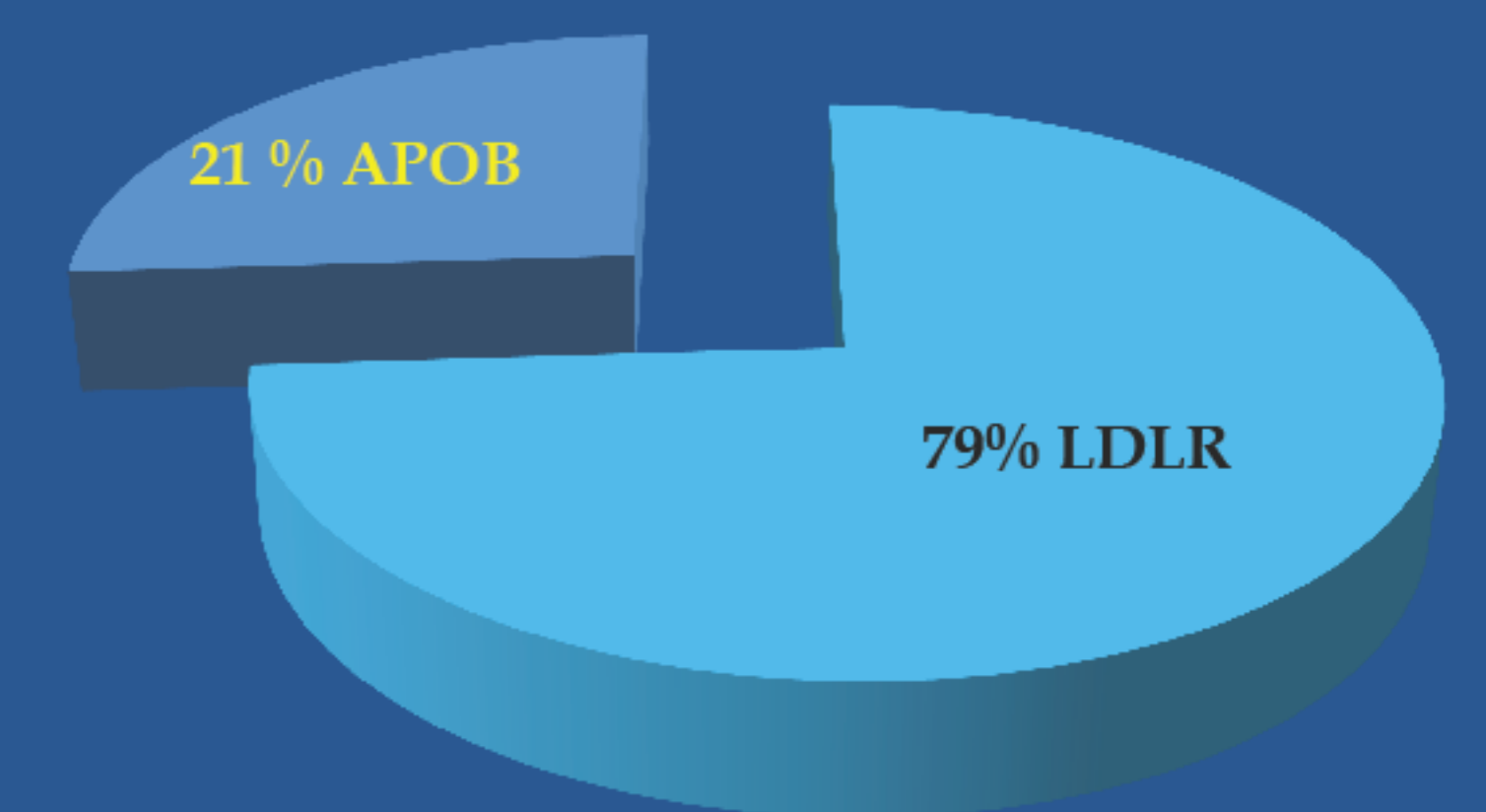


MATERIALS AND METHODS

- The study included children with elevated cholesterol level
- In 210 patients secondary causes of hypercholesterolemia were excluded and molecular testing for mutation in LDLR and APOB genes was performed
- Aged 3 to 18 years

RESULTS

- In group of 210 patients with hypercholesterolemia FH was confirmed in 79 patients
- In all patients, history of cardiovascular diseases in family was positive
- In physical examination no specific symptoms for FH were seen
- initially the average total cholesterol level was 275±40 mg/dl, LDL 223±39 mg/dl, HDL 57±15 mg/dl, triglycerides 99±25 mg/dl
- In 79% of patients in the LDLR gene and in 21% in the APOB gene, mutations were found



- TREATMENT:** All patients with FH started diet and treatment with statins in 59 patients and with statin and ezetimibe in 4 patients with FH was started

- The average level of total cholesterol in the control tests after 12 weeks of treatment was 214±23 mg/dl, LDL 136±18 mg/dl, HDL 55±12 mg/dl, triglycerides 89±27 mg/dl
- In the group of patients treated with pharmacological therapy, no adverse side effects of the treatment were reported

Parameter	Result at baseline
Age of diagnosis [years]	9,8 ± 4,1
F:M	59% : 41%
Positive family history	100%
BMI (10 - 90 percentile)	100%
BP (10-90 percentile)	100%
TC [mg/dl]	275 ± 40
LDL-C [mg/dl]	223 ± 39
HDL-C [mg/dl]	57 ± 15
TG [mg/dl]	99 ± 25
HbA1c [%]	5,36 ± 0,3
TSH	1,9 ± 0,87

Parameter	Before treatment	Ater 12 weeks
TC [mg/dl]	275 ± 40	214 ± 23
LDL [mg/dl]	223 ± 39	136 ± 18
HDL [mg/dl]	57 ± 15	55 ± 12
TG [mg/dl]	99 ± 25	89 ± 27

CONCLUSIONS

- Hypercholesterolemia should be diagnosed and treated as soon as possible
- Therapy consisting of diet, statins and ezetimib is a safe form of therapy in children
- It is necessary to continue monitoring the efficacy and safety of therapy in children with FH