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ABSTRACT FORM

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1. GASTROENTEROLOGY
2. **HEPATOLOGY**
3. NUTRITION

Abstract Title - Study of Family Clustering and PNPLA3 Polymorphism in Pediatric Non Alcoholic Fatty Liver Disease

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Body of Abstract

Background/Aims

Nonalcoholic fatty liver disease (NAFLD) is one of the most common liver disease worldwide both in adults as well as in children. There is a strong suggestion that familial/genetic factors (obesity, insulin resistance, NAFLD, type 2 diabetes mellitus, PNPLA3 polymorphism) are a major determinant of whether an individual will have NAFLD or not. No data on family clustering and PNPLA3 polymorphism in pediatric population is available from Indian subcontinent. We, therefore, aimed to establish correlation of pediatric NAFLD with predictive metabolic risk factors and PNPLA3 polymorphism in families.

Methods

As observational, prospective study was performed including children (aged 5-18 years) with diagnosed NAFLD (ultrasonographic evidence of fatty liver) and their parents. Detailed evaluation of subjects was done including anthropometry, metabolic screening, PNPLA3 I148M polymorphism and liver biopsy (as applicable).

Results

A total of 39 children (33 males and 6 females) were included along with their parents suggestive of predominant male predilection. The mean age of children was 154.5 $\pm$ 38.3 months. In these children, a family history of NAFLD/Dyslipidemia, Type 2 Diabetes Mellitus (DM), Hypertension (HTN) and Hyperuricemia could be elicited in 19 (48 %), 10 (25 %), 18 (46 %) and 1 subject respectively.

Frank metabolic syndrome (in those aged $\geq$ 10 years, with obesity + 2 out of HTN/ high triglycerides/ low HDL/ high blood sugar) was seen in 4 (10 %) patients. HTN, Pre-HTN, Obesity, Overweight, Hyperuricemia, Dyslipidemia (high TG/low HDL), Insulin resistance and DM was seen in 4 (10 %), 9 (23 %), 21 (54 %), 11 (28 %), 7 (18 %), 28 (72 %), 9 (23 %) and 4 (10 %) pediatric subjects respectively. PNPLA3 polymorphism was detected in 21 children (9 homozygous and 12 heterozygous) i.e > 50 % of the cases. None of the children had higher grades of fatty liver ($\geq$ grade 3) on USG. In 6 patients in whom liver biopsy could be done, 3 patients were diagnosed to have NASH with F1, F2 and F3 fibrosis in 1 patient each.

In the families' workup, PNPLA3 polymorphism was detected in 26 fathers (6 homozygous and 20 heterozygous) and 16 mothers (4 homozygous and 12 heterozygous). Frank metabolic syndrome was detected in 13 fathers and 11 mothers.

Conclusion

Pediatric NAFLD is not an uncommon problem in Indian population with upto 10 % showing evidence of frank metabolic syndrome. Presence of PNPLA3 polymorphism could be confirmed in more than half of the cases. There is significant prevalence and family clustering of metabolic risk factors and PNPLA3 polymorphism in such children. This data could guide us to prognosticate the families so as to allow better management and prevent future risk of various metabolic complications.

Key Words: Pediatric Non Alcoholic Fatty Liver Disease, Family Clustering, PNPLA3 Polymorphism