






Exploring metabolic etiologies of MAFLD in children with normal BMI

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ABSTRACT

Objective: Metabolic dysfunction-associated fatty liver disease (MAFLD) is increasingly recognized in children with a normal body mass index (BMI), and its etiology is generally associated with metabolic, genetic, and environmental factors. Although childhood obesity is a well-known cause of MAFLD, the condition can also develop in non-obese children, often due to underlying metabolic disorders.

Material and Methods: This retrospective, cross-sectional study was conducted over a three-year period at a Pediatric Gastroenterology Clinic. Initially, 253 pediatric patients diagnosed with hepatic steatosis by abdominal ultrasound were screened. After excluding overweight children and children with obesity, 20 non-obese patients with hepatic steatosis were included in the final analysis. The study focused on identifying secondary causes of hepatic steatosis in this cohort by collecting demographic, clinical, biochemical, and metabolic data.

Results: Of the 20 non-obese children evaluated by a pediatric metabolic specialist, two were diagnosed with a metabolic disease. Significant biochemical markers, such as elevated serum triglyceride, ALT, and AST levels, prompted further metabolic investigations, enabling early diagnosis and treatment.

Conclusion: This study highlights the critical role of metabolic and genetic screening in the evaluation of hepatic steatosis in non-obese children, particularly during infancy and early childhood. Early identification of underlying metabolic disorders may enable appropriate intervention and help prevent long-term complications. Non-obese pediatric MAFLD requires careful evaluation and targeted metabolic testing to achieve timely treatment and improved outcomes.

Keywords: Metabolic dysfunction, non-obese children, pediatric MAFLD, rare diseases.

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INTRODUCTION

Fatty liver disease, also referred to as metabolic dysfunction-associated fatty liver disease (MAFLD), is an increasing health problem characterized by the accumulation of fat in the liver. Although obesity is one of the most common causes of this disease, fatty liver disease can also be observed in children with a normal body mass index (BMI).^[1,2] It encompasses a spectrum of liver pathology ranging from simple steatosis to metabolic dysfunction-associated steatohepatitis (MASH), fibrosis, and ultimately cirrhosis.^[3]

While childhood obesity is widely recognized as the primary risk factor for fatty liver disease, recent studies emphasize that the condition can also develop in children with a normal BMI, often as a result of underlying metabolic, genetic, or environmental factors.^[4] The global increase in childhood obesity parallels the rise in the prevalence of MAFLD. Schwimmer et al.^[2] reported that MAFLD affects approximately 38% of obese children and approximately 10% of children with normal BMI. However, the etiology of fatty liver disease in non-obese children is often secondary and may include rare but clinically important disorders such as glycogen storage diseases (GSD), Wilson's disease, congenital disorders of glycosylation, and inherited lipid storage disorders such as Chanarin–Dorfman syndrome (CDS).^[5–7]

Importantly, insulin resistance, the hallmark of metabolic syndrome, can occur independently of obesity and plays a significant role in the pathogenesis of MAFLD. Children with inborn errors of metabolism (IEMs) often present at a very early age and may develop fatty liver in the absence of common risk factors such as obesity. Therefore, early diagnosis of IEMs can be challenging. Fatty liver disease in this group may also result from certain medications, infections (e.g., HCV and CMV), and nutritional deficiencies.^[8]

Clinical manifestations of MAFLD in non-obese children are often nonspecific, and routine liver function tests may fail to accurately identify the disease. Abdominal ultrasonography is commonly used for screening but lacks sensitivity for early or mild steatosis. Liver biopsy remains the gold standard for definitive diagnosis and for differentiating simple steatosis from MASH. Even in young patients with normal BMI, histopathological findings such as macrovesicular steatosis, portal inflammation, and varying degrees of fibrosis are frequently observed.^[9,10]

A thorough investigation of the etiology of MAFLD in non-obese children is crucial, given the availability of targeted therapies for many underlying conditions and the potential for progression to severe liver disease. Early intervention, individualized management, and improved long-term outcomes are achievable through identification of these secondary causes.^[10] In addition to obesity and insulin resistance, genetic factors affecting hepatic structure, dietary habits, and environmental influences play important roles in the development of fatty liver disease. In this context, determining the frequency of metabolic diseases in children with normal BMI but fatty liver disease will contribute to a better understanding of this condition.^[11]

The aim of this study was to determine the frequency of secondary and metabolic causes of MAFLD and to characterize its clinical features in children with normal BMI who presented with hepatic steatosis.

MATERIAL AND METHODS

Study Design and Data Collection

This retrospective, cross-sectional study was conducted at the Department of Pediatric Gastroenterology and Hepatology, Göztepe Süleyman Yalçın City Hospital, and the Division of Inherited Metabolic Disease and Nutrition Clinic at Kartal Dr. Lütfi Kırdar City Hospital between April 2022 and April 2025. Children with a normal body mass index (BMI) who presented with hepatic steatosis during this period were evaluated by a pediatric gastroenterologist and a pediatric metabolic specialist. Clinical, laboratory, and radiological data were retrospectively collected from electronic medical records.

Patient Selection and Inclusion Criteria

Patients with hepatic steatosis detected on ultrasonographic examination were retrospectively screened. To reduce confounding variables associated with obesity, patients with a BMI below the 85th percentile according to national growth reference standards for age and sex were included. Individuals classified as overweight or obese were excluded. Patients who were not evaluated by a pediatric metabolic disease specialist were also excluded from the study.

Although liver biopsy remains the gold standard for diagnosing MASH, histopathological confirmation was not feasible for all participants. Therefore, the diagnosis of hepatic steatosis in this study was based solely on ultrasonographic findings. Liver biopsy was performed in a limited number of patients; however, these data were not consistently documented and were not used as an inclusion criterion.

Inclusion Criteria

- Ultrasonographic evidence of hepatic steatosis
- BMI below the 85th percentile for age and sex according to national growth reference standards
- Evaluation by a pediatric metabolic disease subspecialist

Exclusion Criteria

- BMI \geq 85th percentile (classified as overweight or obese)
- Diagnosis of viral hepatitis (HBV, HCV)
- Diagnosis of autoimmune hepatitis, Wilson's disease, alpha-1 antitrypsin deficiency, or celiac disease
- History of prolonged use of medications known to cause hepatic steatosis (e.g., corticosteroids, valproic acid, methotrexate)
- Presence of known chronic systemic or genetic syndromes unrelated to MAFLD

Comprehensive Clinical and Metabolic Assessment

The age, sex, height, weight, and body mass index (BMI) of all patients, as well as physical examination findings, abdominal ultrasound results, and metabolic and genetic test results, were retrieved from hospital medical records. Biochemical and metabolic assessments obtained from the hospital database included liver function tests (ALT, AST, ALP, GGT, bilirubin, albumin, INR); uric acid levels; lipid profile (cholesterol, LDL, HDL, triglycerides); HOMA-IR calculation based on serum insulin levels; serum amino acid chromatography; urine organic acid analysis

Table 1: Patients' demographic data

	Average	Median	SD	Minimum	Maximum	p
Age (years)	8.83	11.04	6.23	0.08	16.92	0.07
Weight (kg)	35.19	36.70	23.46	4.62	78.00	0.20
Weight (p)	58.90	68.72	29.04	2.22	95.35	0.10
Height (cm)	127.90	142.50	42.56	55.00	180.00	0.11
Height (p)	61.89	67.49	24.15	2.68	97.56	0.20
Waist circumference (cm)	51.18	53.50	5.77	37.00	57.00	0.02
Waist circumference (p)	45.89	49.41	25.49	2.22	91.92	0.20
BMI (kg/m ²)	18.62	18.25	2.98	13.91	25.47	0.04
BMI (P)	51.71	53.59	27.97	4.95	91.77	0.20

p: Kolmogorov-Smirnov Normality Test; *p>0.05: The data are normally distributed. SD: Standard deviation; p: percentile; BMI: Body mass index.

using gas chromatography/mass spectrometry (GC/MS); and plasma acylcarnitine profiling by tandem mass spectrometry.

This study was conducted in accordance with the Helsinki Declaration and was approved by the hospital's Institutional Ethics Committee (approval no: 2024/010.99/9/33). As this was a retrospective study, informed consent was not obtained from the patients' relatives.

Statistical Analysis

The Kolmogorov–Smirnov test was used to assess the normality of variable distributions, and variables were considered to be normally distributed when p>0.05. Qualitative data were expressed as frequencies (%). Relationships between quantitative variables were analyzed using the Pearson correlation test for normally distributed data and the Spearman correlation test for non-normally distributed data. A p-value <0.05 based on two-tailed test results was considered statistically significant. All statistical analyses were performed using SPSS Statistics version 25.00.

RESULTS

A total of 253 pediatric patients who were diagnosed with hepatic steatosis by abdominal ultrasonography (USG) and who presented to the pediatric outpatient clinic within the last three years were screened. Of these, 20 patients with BMI values below the 85th percentile who were evaluated by a pediatric metabolic diseases specialist were included in the final analysis. Overweight and obese patients, as well as those not evaluated by a metabolic specialist, were excluded.

Demographic and Anthropometric Characteristics

The study included 20 pediatric patients diagnosed with hepatic steatosis despite having a normal BMI. The mean age of the cohort was 8.83±6.23 years, ranging from infancy to adolescence. The mean body weight was 35.19±23.46 kg, and the mean height was 127.90±42.56 cm. The mean BMI was 18.62±2.98 kg/m², and all values were within normal limits according to national growth

standards for age and sex. The mean waist circumference was 51.18±5.77 cm. No significant sex-related differences were observed in the cohort (Table 1).

Biochemical and Metabolic Evaluation

Biochemical evaluation revealed that the mean serum alanine aminotransferase (ALT) level was 36.85±70.52 IU/L, while the mean aspartate aminotransferase (AST) level was 50.15±85.31 IU/L. The mean alkaline phosphatase (ALP) level was 252.70±125.61 IU/L, and the mean gamma-glutamyl transferase (GGT) level was 26.15±31.19 IU/L. Total bilirubin levels were below 1 mg/dL in all patients, and no cases of hyperbilirubinemia were observed. Coagulation parameters, including the international normalized ratio (INR), prothrombin time (PT), and activated partial thromboplastin time (APTT), were within normal limits, and no coagulation disorders were detected. The mean triglyceride level was 113.40 mg/dL, and triglyceride concentrations were within normal limits in all patients except those diagnosed with glycogen storage disease. The mean serum albumin level was 4.66±0.24 g/dL. The mean creatine kinase (CK) level was 150.65±100.65 IU/L. The mean serum uric acid level was 4.42±1.33 mg/dL, and the mean total bilirubin level was 1.06±1.26 mg/dL. Except for one patient, ALT, AST, CK, and triglyceride values were within reference limits (Table 2).

Amino Acid and Carnitine Profiles

Furthermore, amino acid analysis, urinary organic acid screening, and plasma acylcarnitine profiling were within normal limits in all patients except one, who required further metabolic evaluation and was diagnosed with a fatty acid oxidation disorder. Free carnitine (C0) levels were also evaluated in all patients. The mean free carnitine concentration was 30.31±10.97 μmol/L, which falls within the reference range for the pediatric population.

Radiological Findings

Liver steatosis was evaluated by ultrasonography in all patients. The severity of liver steatosis was predominantly mild (Grade 1), with moderate (Grade 2) steatosis reported in only two patients. No

Table 2: Descriptive statistical information related to measurement values

	Average	Median	SD	Minimum	Maximum	p
Glucose (mg/dl)	89.00	90.00	6.92	70.00	100.00	0.20
ALT (IU/L)	36.85	20.00	70.52	9.00	334.00	0.00
AST (IU/L)	50.15	26.00	85.31	12.00	405.00	0.00
ALP (IU/L)	252.70	230.50	125.61	70.00	618.00	0.20
GGT (IU/L)	26.15	15.50	31.19	10.00	143.00	0.00
Total bilirubin (mg/dl)	1.06	0.54	1.26	0.15	4.57	0.00
Direct bilirubin (mg/dl)	0.21	0.15	0.18	0.05	0.69	0.01
Protrombin time	13.12	12.55	1.53	10.90	16.20	0.05
INR	1.06	1.06	0.08	0.91	1.25	0.12
APTT	28.21	28.85	3.25	20.10	33.60	0.20
Creatinin kinase (IU/L)	150.65	118.00	100.65	60.00	431.00	0.00
Cholesterol (mg/dl)	152.80	151.50	27.80	95.00	203.00	0.20
Triglyceride (mg/dl)	113.40	94.50	89.41	29.00	419.00	0.01
HDL (mg/dl)	51.10	47.00	14.97	20.00	88.00	0.20
LDL (mg/dl)	75.20	82.50	32.00	0.00	127.00	0.20
Albumin (g/dl)	4.67	4.65	0.24	4.20	5.10	0.20
Uric asit (mg/dl)	4.42	4.10	1.33	2.50	7.10	0.16
Lactate (μ mol/L)	1.46	1.30	0.66	0.90	3.70	0.02

p: Kolmogorov-Smirnov Normality Test, * $p > 0.05$: The data are normally distributed. SD: Standard deviation; ALT: Alanine aminotransferase; AST: Aspartate aminotransferase; ALP: Alkaline phosphatase; GGT: Gamma-glutamyl transferase; PT: Prothrombin time; INR: International normalized ratio; APTT: Activated partial thromboplastin time; CK: Creatine kinase; HDL: High-density lipoprotein; LDL: Low-density lipoprotein.

patient was found to have severe (Grade 3) steatosis. The mean follow-up period was 12 months, during which complete resolution of liver steatosis was observed in five patients. No significant changes in steatosis severity were noted in the remaining patients.

When liver and spleen sizes were evaluated according to body size percentiles by abdominal ultrasonography, no hepatomegaly or splenomegaly was detected in any patient, except for the patient diagnosed with glycogen storage disease type 3.

Case Studies and Diagnostic Insights

Notably, one patient was diagnosed with carnitine-acylcarnitine translocase (CACT) deficiency at the age of 2 months after presenting with hepatic steatosis. Fatty liver disease was detected on abdominal ultrasonography performed during evaluation for neonatal hypoglycemia. A diagnosis of CACT deficiency was established by genetic analysis in a patient suspected of having a fatty acid oxidation defect based on an increased C16+C18:1/C2 ratio identified in emergency acylcarnitine analysis. Following diagnosis, a low-fat diet enriched with medium-chain triglycerides (MCT), frequent feeding—particularly during nighttime—and regulated caloric intake were initiated.

Another case involved a 3-month-old female patient with elevated ALT, AST, CK, and triglyceride levels who was found to have Grade 2 fatty liver disease. Physical examination revealed

hepatosplenomegaly. Screening for inherited metabolic diseases and emergency carnitine profiling yielded normal results, and hypoglycemia was not detected. Genetic analysis identified a homozygous mutation in the AGL gene, leading to a diagnosis of glycogen storage disease type 3. The patient was advised to follow a high-carbohydrate diet with frequent feeding.

Follow-up and Disease Progression

The mean follow-up duration for patients in this study was approximately 12 months under pediatric gastroenterology supervision. During this period, ultrasonographic resolution of hepatic steatosis was observed in five of the 20 patients, suggesting that hepatic steatosis may be transient or potentially overdiagnosed when based solely on imaging findings, particularly in patients with normal metabolic evaluations.

DISCUSSION

In this study, we evaluated pediatric patients with ultrasonographically confirmed hepatic steatosis despite having a normal BMI, aiming to identify underlying metabolic or genetic causes. Although obesity remains the most well-known and prevalent cause of MAFLD, our findings support growing evidence that fatty liver disease in non-obese children is neither a benign nor an idiopathic condition but may indicate a significant underlying pathology, particularly in very young infants.

First, the majority of our patients exhibited normal liver enzyme levels and metabolic parameters, reinforcing the notion that incidental ultrasonographic detection of hepatic steatosis does not always reflect clinically significant metabolic pathology in non-obese children. Mean ALT and AST levels were mildly elevated but remained within reference ranges in all but one patient, suggesting that substantial hepatocellular injury is uncommon in this subgroup. Notably, the mean ALT (36.85 ± 70.52 IU/L) and AST (50.15 ± 85.31 IU/L) values demonstrated wide standard deviations, reflecting heterogeneity in hepatic involvement. Although elevated ALP and GGT levels were observed in some patients, no cases of hyperbilirubinemia or coagulopathy were documented, further supporting the generally benign biochemical profile of this cohort.

Our study highlights the critical role of metabolic investigations in non-obese children with hepatic steatosis. While most patients had normal amino acid, organic acid, and acylcarnitine profiles, two notable cases underscore the diagnostic value of a comprehensive metabolic workup. One patient was diagnosed with carnitine-acylcarnitine translocase (CACT) deficiency at 2 months of age following presentation with hepatic steatosis. CACT deficiency is a rare but severe disorder of mitochondrial long-chain fatty acid oxidation that may present with hypoglycemia, cardiomyopathy, and liver dysfunction during the neonatal period.^[12,13] In this patient, hepatic steatosis was the sole initial finding prompting further metabolic evaluation, ultimately enabling early diagnosis and treatment. Similarly, another infant was diagnosed with glycogen storage disease (GSD) type III after presenting with elevated liver enzymes, hypertriglyceridemia, and fatty liver changes on ultrasonography. Genetic analysis revealed a homozygous mutation in the AGL gene, confirming the diagnosis. This finding is consistent with previous reports indicating that hepatomegaly and steatosis may represent early manifestations of GSD, often preceding overt clinical symptoms.^[14] The absence of hepatosplenomegaly in most of our cohort, except in the patient with GSD type III, suggests that organomegaly remains an important clinical indicator for distinguishing metabolic hepatopathies from isolated or benign steatosis.

These two cases demonstrate that the earlier hepatic steatosis is detected in infants, the higher the likelihood of an underlying metabolic disorder. While fatty liver in an obese adolescent may suggest classic MAFLD, fatty liver in a non-obese infant or toddler should prompt a thorough metabolic and genetic evaluation. This concept is supported by prior literature. Vimalasvaran et al.^[7] reported that in children younger than 10 years with biopsy-confirmed fatty liver disease, glycogen storage disease was the leading cause, accounting for more than 30% of cases.^[2,7] Schwimmer et al.^[2] also demonstrated that NAFLD may affect approximately 10% of non-obese children in autopsy-based studies and that histological features cannot be distinguished from those observed in obese patients.^[2,7] Yıldız and Sivri^[6] further emphasized that a substantial proportion of non-obese children undergoing liver biopsy for suspected NAFLD have underlying metabolic or genetic etiologies. This observation highlights that metabolic disorders may be misdiagnosed as idiopathic fatty liver disease in the absence of a comprehensive metabolic evaluation, underscoring the importance of early diagnosis. In our study, a patient diagnosed with carnitine-acylcarnitine translocase (CACT) deficiency exhibited elevated serum triglyceride, ALT, and AST levels.

These abnormalities served as the initial indicators prompting further diagnostic investigations. This case emphasizes the importance of early identification of rare metabolic disorders, as timely intervention can prevent long-term complications such as hypoglycemia, liver dysfunction, and cardiomyopathy in CACT deficiency.

Another important finding of our study is that lipid metabolism should be evaluated even in non-obese children. Particularly in the absence of obesity, elevated serum triglyceride and cholesterol levels may indicate underlying conditions such as familial combined hyperlipidemia, glycogen storage diseases, or fatty acid oxidation defects. Therefore, a routine metabolic work-up, including serum cholesterol, triglycerides, uric acid levels, and extended newborn screening panels (if not previously performed), should be considered in all non-obese children with hepatic steatosis. Ultrasonography remains a valuable, non-invasive tool for detecting hepatic steatosis; however, its ability to determine the underlying etiology is limited. Biochemical markers and enzyme panels alone may also fail to provide a complete assessment. In such cases, liver biopsy remains the gold standard, particularly in patients with unexplained or progressive liver dysfunction. In our cohort, biopsy findings further supported the presence of storage disorders in selected cases and reinforced the need for tissue-based diagnosis when clinically indicated.

Taken together, our findings suggest a paradigm shift in the evaluation of pediatric fatty liver disease. While obesity-related fatty liver disease is increasingly prevalent and well characterized, fatty liver disease in non-obese children—especially in infants—requires careful and early investigation for underlying, potentially treatable disorders. Early diagnosis not only guides appropriate management but also has important implications for genetic counselling, family screening, and long-term prognosis.

Importantly, two of the 20 patients were diagnosed with underlying metabolic diseases, and both were younger than one year at the time of diagnosis. This finding highlights the significance of age at diagnosis as a potential indicator of metabolic liver disease. In contrast, the remaining 18 patients exhibited normal or nonspecific metabolic profiles during the follow-up period.

This observation supports previous studies, such as that by Yıldız and Sivri, which emphasized that non-obese children with hepatic steatosis diagnosed at a younger age are more likely to have an underlying genetic or metabolic condition.^[6] Conversely, older children with normal BMI and no significant metabolic abnormalities may have a more benign form of fatty liver disease that may resolve or remain stable without requiring invasive interventions. These findings suggest that early-onset steatosis in infants may warrant a more aggressive metabolic work-up, whereas a strategy of careful observation may be more appropriate in older children with normal BMI and unremarkable laboratory findings. This distinction may help avoid unnecessary anxiety, invasive testing, or overtreatment in a subset of patients while ensuring timely diagnosis in high-risk infants. Further prospective studies are needed to identify predictive factors for the persistence or resolution of steatosis in this population and to determine clinical or biochemical indicators that should prompt metabolic evaluation. Our findings contribute to the growing body of evidence supporting the need for age- and risk-stratified algorithms in the assessment of pediatric hepatic steatosis.

This study has several limitations. First, due to its retrospective design, the analysis was restricted to available clinical and laboratory data, and standardized assessment protocols could not be uniformly applied. Second, genetic testing and liver biopsy were not performed in all patients but were limited to selected cases based on clinical suspicion; therefore, some underlying metabolic or genetic conditions may have been underdiagnosed. Another limitation is that potential confounding factors, such as dietary habits, physical activity levels, and socioeconomic status, were not assessed in detail and thus could not be fully controlled. These variables are known to influence the development and progression of hepatic steatosis and may have affected the interpretation of our results. Finally, the relatively short follow-up period limited our ability to evaluate the long-term progression or resolution of hepatic steatosis in this population.

CONCLUSION

Our findings emphasize the importance of considering underlying metabolic and genetic disorders, particularly in non-obese children presenting with hepatic steatosis during early infancy. When fatty liver is detected at a very young age, accompanied by significant biochemical abnormalities or associated with parental consanguinity, the likelihood of an inborn error of metabolism increases substantially. Therefore, patients with early-onset or severe hepatic steatosis—especially in the presence of parental consanguinity or hepatomegaly—should undergo comprehensive metabolic screening without delay. Referral to a pediatric metabolic specialist is essential to ensure accurate diagnosis, timely intervention, and improved clinical outcomes.

Statement

Ethics Committee Approval: The Kartal Dr. Lütfi Kırdar City Hospital Ethics Committee granted approval for this study (date: 25.10.2024, number: 2024/010.99/9/33).

Informed Consent: As this was a retrospective study, informed consent was not obtained from the patients' relatives.

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