VOLUME-6 | ISSUE-1 | JANUARY-2017 • ISSN No 2277 - 8179 | IF : 3.508 | IC Value : 78.46

Case report Dyslipidemia in infancy – Is screening the way forward?

Paediatrics

KEYWORDS: Hyperlipidemia, Hypercholesterolemia,Hypertriglyceridemia

Dr Parimala V Thirumalesh — MD, MRCPCH Senior Consultant in Paediatrics and Neonatology Aster CMI Hospital, Bangalore

Dr Yamuna Subha Tulasi — MBBS, DNB Specialist Paediatrics Aster CMI Hospital, Bangalore

ABSTRACT

Dyslipidemias are disorders of lipoprotein metabolism resulting in elevated levels of cholesterol, triglycerides, low density lipoproteins and decreased or normal high density lipoproteins. These disorders are often asymptomatic and lead to atherosclerosis and early death if undetected. The prevalence is reported to be as high as 20 percent in children between the age of six and nineteen years in the United States where routine screening is in practice. The prevalence in our country and in the age group below six years universally is unknown In this case report the authors describe a 5 month old infant who presented with gastroenteritis and was detected to have dyslipidemia incidentally due to his lipemic blood sample. Since the mainstay of treatment is dietary management, introducing it early on during the weaning period and avoiding the usual practice of high fat diet for weaning will prove to be a key factor in lowering the risk of morbidity and mortality in these children. This article aims to raise the question of whether screening for dyslipidemia should begin before the weaning age universally so that proper dietary management can be initiated early on to prevent morbidity and mortality in these silent sufferers.

Introduction

Hyperlipidaemia involves abnormally elevated levels of any or all lipids and/or lipoproteins in the blood. Hyperlipidemias may basically be classified as either familial (also called primary)[1] caused by specific genetic abnormalities, or acquired (also called secondary)[1] when resulting from another underlying disorder that leads to alterations in plasma lipid and lipoprotein metabolism.[1]

Case report

We report a case of Type 1 hyperlipidaemia in a 5 month old boy who presented with loose stools and hypovolemic shock. When blood was drawn for investigations, it was found to be lipemic (pic 1) hence was .investigated for hyperlipidaemia. Clinically the child had pallor, waxy appearance of the skin and moderate hepatosplenomegaly .Shock was treated with intravenous fluid replacement therapy and baby was well within 12 hours. Child was put on diet control for hyperlipidaemia and was followed up for a period of 3 years. The progressive decline in the lipid levels are shown below in graph 2 and 3..The screening for inborn errors of metabolism was normal in this child.

Fig 1: venous sample collected during cannulation

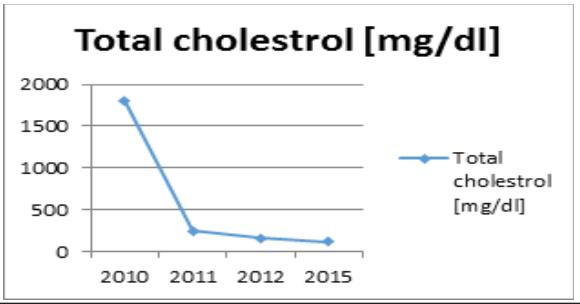

Fig2: Progressive decline in the levels of Total cholesterol over 5 years of follow up

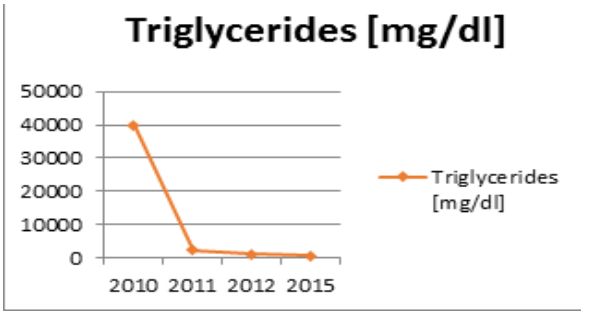

Fig3: Progressive decline in the levels of serum Triglycerides over 5 years of follow

Discussion

Type 1 Hyperlipoproteinemia (T1HLP) presenting in childhood is in most cases due to genetic deficiency of lipoprotein lipase (LPL) or related proteins such as apolipoprotein (apo) C2, apo A5, and glycosylphosphatidylinositol-anchored high-density lipoprotein binding protein 1 (GPIHBP1) [2,3]. Absent or non-functional LPL due to mutations in LPL or other genes encoding the cofactors essential for its activity results in decreased hydrolysis of triglycerides transported in chylomicrons and very low-density lipoproteins at the tissue capillary endothelial surface. These patients suffer from recurrent attacks of acute pancreatitis, eruptive xanthomas, and lipemia retinalis[4]

The full-blown disease is manifested by attacks of abdominal pain, hepatosplenomegaly,

eruptivexanthomas, and lactescence of the plasma.

The main treatment modalities for familial hyperlipedemia remain dietary modifications in children less than 8 years of age and above 8 years of age, a combined approach with diet changes and Pharmacological interventions with Statins and fibrates[5]

Routine screening for hyperlipidemias in children remains controversial and no paediatric organizations have recommended universal screening[6]. National Heart, Lung, and Blood Institute [NHLBI] and American Academy of Pediatrics [AAP] recommends target strategy approach for screening children for hyperlipidaemia, which includes screening children with a known family history of

dyslipidemia or premature onset of cardiovascular disease between the ages of 2 and 8, children with unknown family history but with other risk factors such as obesity, hypertension, and diabetes between the ages of 2 and 8.[7,8]

Screening before
2 years of age is not recommended.[7,8]
As it has been seen form our experience of the above discussed case, an early screening for serum lipid levels would help the paediatrician to identify the children with Familial hyperlipidemia at an early age so as to institute the necessary interventions.

Conclusion and Recommendation
An additional inclusion of serum lipid levels if done along with the newborn screening programme for other congenital disorders will help in identifying children with familial hyperlipidemia at the very early stage and hence more favourable outcome in terms of treatment can be achieved before the morbidity due to hyperlipidemia manifests clinically

References

1. Chait A, Brunzell JD (June 1990). "Acquired hyperlipidemia (secondary dyslipoproteinemias)". Endocrinol. Metab. Clin. North Am. 19 (2): 259–78.
2 Rahalkar AR, Giffen F, Har B et al., 2009. Novel LPL mutations associated with lipoprotein lipase deficiency: two case reports and a literature review. Can J PhysiolPharmacol 87:151–160
3. Péterfy M, Ben-Zeev O, Mao HZ et al., 2007. Mutations in LMF1 cause combined lipase deficiency and severe hypertriglyceridemia. Nat Genet 39:1483–1487
4. Garg A, Simha V. 2007. Update on dyslipidemia. J ClinEndocrinolMetab 92:1581–1589
5. Holt, L. E., Jr., Aylward, F. X., Timbers, H. G. Idiopathic familial lipemia. Bull. Johns Hopkins Hosp. 64: 279-314, 1939.
6. US Preventive Services Task Force. Screening for lipid disorders in children: US Preventive Task Force recommendation statement. Pediatrics. 2007;120(1). Available at: www.pediatrics. org/cgi/content/full/120/1/e215
7. National Heart, Lung, and Blood Institute. Third Report of the National Cholesterol Education Program (NCEP) Expert Panel on Detection, Evaluation, and Treatment of High Blood Cholesterol in Adults. Final Report. Washington, DC: National Institutes of Health; 2002.
8. Lipid Screening and Cardiovascular Health in ChildhoodPediatrics 2008; 122:1 198-208

.