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Abstract Title: - A Case report of neonate with white blood

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Aims: - Hypercholesterolemia is the one of the most important risk factors for coronary vascular disease and persistent high cholesterol level in blood lead to premature coronary heart disease and early age of death [1, 2]. Familial hypercholesterolemia is occurred by mutation in hereditary genes (APOB, LDL-R, PCSK9) manifested as autosomal dominant but if mutation in adapter protein 1(LDL-R AP1) gene inherited as autosomal recessive [3, 4. We are describing a newborn case of familial hypercholesterolemia with second degree family history which was suspected during blood sampling as white blood.

Methods: - A 24 days old male, Hindu of non-consanguineous marriage admitted in NICU with complain of excessive cry with refusal to feed. In birth history, baby had delivered by LSCS and history of immediate cry and no history of NICU admission. Mother had no history of any significant medical disease, drug intake except iron & folic acid, fever. So provisional diagnosis of LONS was made but during sampling, blood was found highly viscous and turning white after few second. Baby was exclusive breast feed. On examination, baby was not dysmorphic or any skin lesion. In vital CRT was <2 sec, RR 34/min, HR 140/min. In systemic examination Anterior fontanelle normal, sucking & rooting poor and neonatal reflex was depressed. Other cardiac, Respiratory and per abdomen was normal. So, it was suspicious about uncommon presentation of familial hypercholesterolemia. In review of family history, parents had no history of any xanthelasma or tendon xanthoma or neither any cardiac disease. But grandfather has history of cardiac disease diagnosed at the age of 30 yrs.

Results: - In septic screen, TLC 12780 (Neutrophil 45.7%& lymphocyte 43.3%) Hb 35.1 g/dl, platelets 5.53 lakh/cmm, CRP 48 mg/L, and in peripheral smear normocytic normochromic blood cell. In blood culture and urine culture was sterile. Other RBS, serum electrolyte and kidney function was normal. Parental Lipid profile was normal. But baby lipid profile was abnormal (Table 1). In ophthalmological examination showed lipemia retinalis.

Serum cholesterol	496 mg/dl
Serum triglyceride	1180 mg/dl
HDL	34 mg/dl
LDL	226 mg/dl

VLDL 216 mg/dl

In genetic study whole exome sequencing showed homozygous pathogenic variant in LPL gene associated with hyperlipoproteinemia, type 1(location Exon 5, variant c.644>A).

Baby was treated with broad spectrum antibiotic inj ceftriaxone and inj amikacin and supportive IV fluid. Baby was also treated with tab atorvastatin. After 10 days of treatment baby was discharge from NICU. Baby was brought in emergency after 17 days as SIDS.

Discussion: Hyperlipidemia is occurred due to increased concentration of plasma lipoprotein and changes resulting from genetic defect are classified as primary disorder of lipoprotein metabolism. FH is a monogenic, autosomal dominant that manifested in heterozygous form in approx. 1 in 500 individuals. Homozygous are occurred approximately 1 in a million [6]. The total cholesterol level is much higher in homozygous variant (500-1000 mg/dl) with respect to heterozygous variant (325-450 mg/dl). Incidence of premature coronary artery disease is an early manifestation in homozygous variant (second decade) and xanthoma occurred in first decade [5].

Conclusions: - This case report highlights the significance of conscious and proactive genetic screening for familial hypercholesterolemia to early diagnosis and treatment to prevent further development of immature coronary heart disease.

Keywords: