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CONTENTS

RESEARCH

- Serum Zinc and C-Reactive Protein Levels as Risk Factors for Mortality in Systemic Inflammatory Response Syndrome
(Kadar Zinc dan C-Reactive Protein Serum Sebagai Faktor Kebahayaan Kematian di Pasien Systemic Inflammatory Response Syndrome)
Dwi Retnoningrum, Banundari Rachmawati, Dian Widyaningrum 1-5
- Correlations between Mean Platelet Volume and Immature Platelet Fraction to Hemoglobin A1c in Patients with Type 2 Diabetes Mellitus
(Kenasaban antara Mean Platelet Volume dan Immature Platelet Fraction terhadap Hemoglobin A1c di Pasien Diabetes Melitus Tipe 2)
Dian W Astuti, Sony Wibisono, Arifoel Hajat, Sidarti Soehita..... 6-11
- Methicillin-Resistant Staphylococcus Aureus Colonization and Screening Method Effectiveness for Patients Admitted to the Intensive Care
(Kejadian dan Ketepatangunaan Penapisan Kolonisasi Methicillin-Resistant Staphylococcus aureus di Pasien Perawatan Intensif)
Andaru Dahesihdewi, Budi Mulyono, Iwan Dwiprahasto, Supra Wimbari 12-18
- Correlation between Visceral Adipose Tissue-Derived Serpin with Fasting Blood Glucose Level in Obesity
(Hubungan Kadar Visceral Adipose Tissue-Derived Serpin Dengan Kadar Glukosa Darah Puasa Pada Kegemukan)
Novi Khila Firani, Agustin Iskandar, Anik Widijanti, Nonong Eriani 19-23
- Serum Glial Fibrillary Acidic Protein Levels Profile in Patients with Severe Traumatic Brain Injury
(Profil Kadar Glial Fibrillary Acidic Protein Serum di Pasien Cedera Otak Berat)
Arief S. Hariyanto, Endang Retnowati, Agus Turchan 24-28
- Phylogenetic Profile of Escherichia coli Causing Bloodstream Infection and Its Clinical Aspect
(Profil Filogenetik Escherichia coli Penyebab Infeksi Aliran Darah dan Aspek Klinisnya)
Osman Sianipar, Widya Asmara, Iwan Dwiprahasto, Budi Mulyono..... 29-35
- Comparison of Glycemic State in Patients with and without Hyperuricemia
(Perbedaan Status Glikemia pada Pasien dengan dan tanpa Hiperurisemia)
Corrie Abednego, Banundari Rachmawati, Muji Rahayu 36-41
- Analysis of Laboratory Parameters as Sepsis Markers in Neonatals with Hyperbilirubinemia
(Analisis Tolok Ukur Laboratorium Sebagai Petanda Sepsis di Neonatus dengan Hiperbilirubinemia)
Bachtiar Syamsir, Rachmawati Muhiddin, Uleng Bahrn..... 42-46
- Correlation Percentage of S and G2/M with Percentage of Lymphoblasts in Pediatric Acute Lymphoblastic Leukemia
(Kenasaban Persentase S dan G2/M dengan Persentase Limfoblas di Pasien Leukemia Limfoblastik Akut Anak)
Erawati Armayani, Yetti Hernaningsih, Endang Retnowati, Suprpto Ma'at, I Dewa Gede Ugrasena . 47-52

Correlation of Blast Percentage to CD34 of Bone Marrow in All Pediatric Patients (<i>Kenasaban Persentase Blas Dengan CD34 di Sumsum Tulang pada Pasien LLA Anak</i>) Rahmi Rusanti, Yeti Hernaningsih, Endang Retnowati, Mia Ratwita Andarsini, Andy Cahyadi	53–58
Analysis of Decreased Glucose Level in Stored Samples Correlated to Serum Separation and Temperature Storage (<i>Analisis Penurunan Glukosa Dari Sampel Yang Disimpan Dalam Kaitannya Dengan Pemisahan Serum dan Suhu Penyimpanan</i>) Gustamin, Liong Boy Kurniawan, Ruland DN Pakasi	59–63
Diagnostic Concordance between Next Generation and High Sensitive Troponin-I in Angina Pectoris Patients (<i>Kesesuaian Diagnostik Troponin-I Next generation dan High sensitive di Pasien Angina Pectoris</i>) Erna R Tobing, Jusak Nugraha, Muhammad Amminuddin	64–69
Elevated Serum S100B Protein Level as a Parameter for Bad Outcome in Severe Traumatic Brain Injury Patients (<i>Peningkatan Kadar Serum Protein S100B Sebagai Tolok Ukur Keluaran Buruk di Pasien Cedera Kepala Berat</i>) Ridha Dharmajaya, Dina Keumala Sari, Ratna Akbari Ganie	70–75
Analysis of Mean Platelet Volume As A Marker For Myocardial Infarction and Non-Myocardial Infarction in Acute Coronary Syndrome (<i>Analisis Mean Platelet Volume sebagai Pembeda Infark Miokard dan Non-Infark Miokard di Sindrom Koroner Akut</i>) Wandani Syahrir, Liong Boy Kurniawan, Darmawaty Rauf	76–80
Anti-Dengue IgG/IgM Ratio for Secondary Adult Dengue Infection in Surabaya (<i>Rasio IgG/IgM Anti Dengue untuk Infeksi Dengue Sekunder Dewasa di Surabaya</i>) Aryati, Puspa Wardhani, Ade Rochaeni, Jeine Stela Akualing, Usman Hadi	81–85
Analysis of Blood Urea Nitrogen/Creatinin Ratio to Predict the Gastrointestinal Bleeding Tract Site (<i>Analisis Rasio Blood Urea Nitrogen/Kreatinin Untuk Meramalkan Lokasi Perdarahan pada Saluran Cerna</i>) Arfandhy Sanda, Mutmainnah, Ibrahim Abdul Samad	86–90
The Differences of Sodium, Potassium and Chloride Levels in STEMI and NSTEMI Patients (<i>Perbedaan Kadar Natrium, Kalium dan Klorida di Pasien STEMI dan NSTEMI</i>) Freddy Ciptono, Muji Rahayu	91–94

LITERATURE REVIEW

Macrophage Autophagy in Immune Response (<i>Otofagi Makrofag dalam Respons Imun</i>) Jusak Nugraha	95–101
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CASE REPORT

Very Severe Hypertriglyceridemia in Suspected Familial Chylomicronemia Infant (<i>Hipertriglisieridemia Sangat Berat di Bayi Terduga Kausa Familial Chylomicronemia</i>) Fitry Hamka, Liong Boy Kurniawan, Suci Aprianti	102–107
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Rismawati Yaswir, Purwanto AP, Sidarti Soehita, July Kumalawati, Aryati,
Rahayuningsih Dharma, Adi Koesoema Aman, Yolanda Probahoedoso, Puspa Wardhani

CASE REPORT

VERY SEVERE HYPERTRIGLYCERIDEMIA IN SUSPECTED FAMILIAL CHYLOMICRONEMIA INFANT

(Hipertrigliseridemia Sangat Berat di Bayi Terduga Kausa Familial Chylomicronemia)

Fitry Hamka, Liong Boy Kurniawan, Suci Aprianti

ABSTRAK

Seorang bayi perempuan berusia 6 bulan, masuk rumah sakit dengan keluhan utama muntah dialami sejak satu hari sebelum masuk rumah sakit dengan kekerapan lebih dari sepuluh kali dalam sehari dan demam dialami sejak 6 jam sebelum masuk rumah sakit. Pasien ini lahir prematur secara sectio secarea dengan berat badan lahir 1,5 kg. Serum pasien lipemik seperti susu. Kadar kolesterol total serum pasien 477 mg/dL, trigliserida 4370 mg/dL (hipertrigliseridemia sangat berat), LDL direk 135 mg/dL, HDL 5 mg/dL dan kadar hemoglobinnnya 8,0 gr/dL. Pasien ini mendapatkan cairan intravena, antibiotik dan transfusi darah akibat anemia yang diderita. Di kasus ini, hipertrigliseridemia sangat berat diduga terkait dengan kilomikronemia familial. Penapisan profil lipid dari orang tua bayi dan keluarga lainnya sangat penting dilakukan untuk menegakkan diagnosis.

Kata kunci: Hipertrigliseridemia, bayi, kilomikronemia familial

ABSTRACT

A 6-month-old female infant was admitted to the hospital with the main complaint of vomiting since one day before admission with a frequency of more than ten times a day and fever since 6 hours before admittance to the hospital. The patient was born preterm by section secarea with 1.5 kg birth weight. The patient's serum was milky. Serum total cholesterol was 477 mg/dL, triglycerides level was 4370 mg/dL (very severe hypertriglyceridemia), direct Low-Density Lipoprotein (LDL) 135 mg/dL, High-Density Lipoprotein (HDL) 5 mg/dL and hemoglobin level was 8.0 gr/dL. The patient received intravenous fluid, antibiotic and blood transfusion due to anemia. In this case, very severe hypertriglyceridemia is suspected to be related to familial chylomicronemia. Lipid profile screening of baby's parent and other families are important to establish the diagnosis.

Key words: Hypertriglyceridemia, infant, familial chylomicronemia

INTRODUCTION

Fat in the body consists of triglycerides, cholesterol, and phospholipids. The main functions of fat are as a source of calories and as a source of essential fatty acids. Lipoproteins are fats in the blood that bind to apoproteins. Lipoproteins also play a role in transporting fat.^{1,2}

Moreover, fat metabolism is divided into two pathways, namely exogenous and endogenous pathway. In the exogenous pathway, triglycerides derived from the food will form a nucleus of nascent chylomicron.

Meanwhile, in the endogenous pathways, the fat will be carried from the liver to peripheral tissue and then from the peripheral tissue back to the liver. This pathway involves two systems, namely apo B-100 and apoA-1. Chylomicrons with a specific gravity of <0.95 g/mL and a diameter of 100–500 nm actually contain apoB-48, apoA-I, apoA-II, APOC-II/C-III and apoE.¹⁻³

Furthermore, hypertriglyceridemia is defined as an elevated fasting plasma triglyceride level of ≥ 150 mg/dL, which may be accompanied by impaired lipoprotein levels or not. Hypertriglyceridemia can be caused by primary and secondary causes.

Primary hypertriglyceridemia is caused by a genetic defect triggering impaired triglyceride metabolism. Meanwhile, secondary hypertriglyceridemia is caused by a poor diet, alcohol use, obesity, metabolic syndrome, type 2 diabetes mellitus and hypothyroid.^{4,5}

In addition, triglyceride concentrations, based on the National Cholesterol Education Program, Adult Treatment Panel III (NCEP ATP III), are divided into four categories, namely normal at <150 mg / dL (<1.7 mmol/L), borderline high at 150-199 mg/dL (1.7-2.3 mmol/L), high at 200-499 mg/dL (2.3-5.6 mmol/L) and very high at > 500 mg/dL (> 5.6 mmol/L). Meanwhile, the Endocrine Society in 2010 classified hypertriglyceridemia into five levels, namely normal (<150 mg/dL), mild hypertriglyceridemia (150-199 mg/dL), moderate hypertriglyceridemia (200-999 mg/dL), severe hypertriglyceridemia (1000-1999 mg/dL) and very severe hypertriglyceridemia (≥ 2000 mg/dL).^{5,6}

The classification system of Fredrickson, moreover, divides five primer hyperlipoproteinemia phenotypes based on patterns of fractions of lipoproteins, namely type I hyperlipoproteinemia (Familial Chylomicronemia), type IIB hyperlipoproteinemia (Familial Combined Hyperlipidemia), type III hyperlipoproteinemia (Dysbetalipoproteinemia), type IV hyperlipoproteinemia (Primary Hypertriglyceridemia) and type V hyperlipoproteinemia (Primary Mixed Hyperlipidemia).^{5,6}

Familial Chylomicronemia (type I hyperlipoproteinemia) is a very rare disorder, which is autosomal recessive. The cause of this disorder is a deficiency of enzyme lipoprotein lipase and apo C-II. Lipoprotein Lipase Deficiency (LPL) is triggered by mutations in the lipoprotein lipase gene with a prevalence of 1 in 1 million people of a population. Diagnosis of Familial Chylomicronemia can be determined in molecular biology by performing DNA test analysis which shows the presence of mutations in both LPL alleles.^{4,7}

Mixed hyperlipidemia (type 5 hyperlipoproteinemia), on the other hand, is similar to Familial Chylomicronemia, with high fasting serum chylomicron level, high triglyceride level of > 1000 mg/dL (> 11.2 mmol/L) and elevated Very Low-Density-Lipoprotein (VLDL) particle level. Familial chylomicronemia usually occurs in childhood or adolescence, while mixed hyperlipidemia occurs in adulthood. Patients with mixed hyperlipidemia are also associated with an increase in total cholesterol

and other types of lipoproteins, especially VLDL, which are not present in Familial Chylomicronemia.^{4,9}

RESEARCH REPORT

History

A 6-month-old female infant was admitted to the Wahidin Sudirohusodo Hospital on April 26, 2014 with a major complaint of vomiting experienced less than 1 day before the admission. The frequency of the vomiting was more than ten times, containing residual liquid. Fever was also experienced continuously for 6 hours before the hospital admission. This infant had no seizures, but experienced cough and shortness of breath. This infant also had a history of regular bowel movements with yellow color, as well as smooth urinating with yellow color. The photo of this very severe hypertriglyceridemia patient aged 6 months with familial chylomicronemia is shown in Figure 1.



Figure 1. The photo of the infant

This patient, moreover, also had no allergy history. This infant had been fed with breast milk (milk) and formula one. She also had been administrated with Bacille Calmette Guerin (BCG), Polio, Hepatitis B and Diphtheria Pertussis Tetanus (DPT) immunizations. The disease that had been suffered was only diarrhea.

On the other hand, her parents were a 30-year-old father and 25-year-old mother. During the pregnancy, her mother regularly controlled with a midwife, received vitamins, iron supplements and Tetanus

Toxoid (TT) injection twice, never drank herbs, but took blood pressure-lowering drugs and paracetamol drugs from obstetricians.

In the birth history of this infant, the mother gave birth by Sectio Caesarea (SC) for the indication of preeclampsia at 7 months of gestation. The baby was born with a weight of 1.5 kg.

Physically, this infant looked like have moderate pain, lack of nutrition, a composmentis awareness of Glasgow Coma Scale (GCS) of 15, a weight of 4.975 kg, a height of 61 cm, an upper arm circumference of 11 cm, a head circumference of 41 cm, a chest circumference of 42 cm and an abdominal circumference of 40 cm. Her blood pressure was 90/60 mmHg with a pulse rate of 130 x / min, a breathing rate of 32x / minute and a body temperature of 38.7°C.

Results of the physical examination also showed anemic impression and lung impression of bronchovascular breath, but without rhonchi and wheezing. Results of the heart auscultation examination indicated a pure and regular heart sound of I/II. Results of the peristaltic abdominal examination revealed normal impression, palpable liver 1 cm below costal arch, tender consistency, flat surface, no tenderness and sharp edges. The results also showed the spleen was not palpable with a dehydration score of 10 (mild-moderate dehydration) used in the Child Health Sciences Department, Hasanuddin University. Other physical examination results were still within normal limits.

Laboratory tests were performed at the RSWS laboratory on August 28, 2014, ie routine blood tests, blood chemistry, peripheral blood smears and regular feces. Routine blood tests showed a hemoglobin (Hb) level of 8.0 g/dL, an erythrocyte count of 2.05 million/mm³, a leukocyte count of 17,200/mm³, 12% of hematocrit, MCV of 57 fl, MCH of 39 pg, MCHC of 69 g/dL, a platelet count of 229,000/Mm³, RDW CV of 26.6%, RDW-SD of 49.8 fl and 2.75% of reticulocyte. Results of the calculation of leukocyte types then indicated 31.7% of lymphocytes, 20.2% of monocytes, 47.7% of neutrophils, 0.2% of eosinophils and 0.2% of basophils.

Results of the blood chemistry examination showed a blood glucose level of 93 mg/dL, unreadable urea and creatinine, a total cholesterol of 477 mg/dL, a LDL level of 135 mg/dL, a HDL level of 5 mg/dL and a triglyceride level of 4370 mg/dL. Her serum after the laboratory examination was milky as the image shown in Fig. 2.

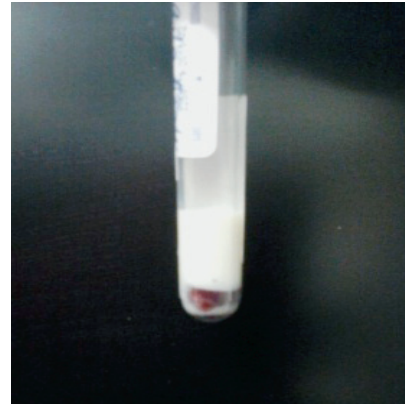


Figure 2. The sample of the patient showed very lipemic and milky serum after separated from blood components during the post-centrifugation

Peripheral blood assessment, furthermore, revealed erythrocytes indicating hypochromic microcytic anemia, anisopoikilocytosis with anulocytes and ovalocytes, pencil cells, target cells, tear drop cells, as well as no inclusions and normoblasts. It also showed sufficient leukocytes, polymorphonuclear (PMN) cells more than Mononuclear cells (MN), toxic granulation and vacuolization, but there was no young cell. Sufficient platelet counts with normal morphology, thus, indicated hypochromic microcytic anemia with Fe/Anemia deficiency of chronic disease accompanied by signs of infection due to the leukocyte counts. Consequently, further examinations should be performed on serum Fe, Ferritin, and Total Iron Binding Capacity (TIBC).

In addition, on macroscopic examination of regular stools, there was a soft consistency, yellow color, no mucus and no blood found in her stools. Microscopically, there were also no worms, parasites, and bacteria found. Laboratory tests then were performed again on April 29, 2014. Results of electrolyte examination showed a sodium level of 134 mmol/L, a potassium level of 2.9 mmol/L and a chloride level of 108 mmol/L. And results of ferritin examination indicated a ferritin level of 45, 35 ng/mL.

Other supporting examinations were also performed. First, echocardiography examination showed normal intracardiac and normal myocardial contractions, suggesting to conduct Multi-Slice-Computerized-Tomography (MSCT) scan for evaluating coronary arteries. Second, Ultrasonography (USG) examination was carried out to observe abdominal, liver, gall bladder, pancreas, spleen, right and left kidneys and vesica urinaria within normal

limits. Third, Computerized Tomography (CT) scan was conducted to evaluate thorax without left bronchopneumonia impression, but with suspected specific bilateral peribronchial lymphadenopathy.

Further laboratory tests were performed on May 5, 2014 after the transfusion of 50 mL PRC. Results of the tests showed a Hb level of 10.2 g/dL, an erythrocyte count of 3.37 million/mm³, a leucocyte count of 8,400/mm³, a hematocrit level of 24%, MCV of 72 fl, MCH of 30 pg, MCHC of 42 g/dL and a platelet count of 389.000/mm³. Results of the calculation of leukocyte types then indicated 20.6% of lymphocytes, 23.2% of monocytes, 50.3% of neutrophils, 5.7% of eosinophils and 0.2% of basophils.

During the admission, the patient was temporarily diagnosed with mild dehydration, accompanied by low nutrition and Anemia Deficiency Fe DD/Anemia Chronic Illness. The ultimate diagnosis (final) of this patient then was hyperlipidemia. This patient was also diagnosed (secondary diagnosis) with malnutrition, hypokalemia, mild-moderate dehydration, mixed iron deficiency anemia and chronic disease anemia.

Initial therapy administrated after admission (on April 26, 2014) involved R/IVFD Asering with a dose of 14 drops/minute, paracetamol syrup with a dose of 50 mg/8 hour/oral and domperidone syrup with a dose of 3x1 /2 cth. The second treatment given on the second day (on April 27, 2014) involved R/O2 1 liter/min, IVFD Asering with a dose of 14 drops/min, ampicillin with a dose of 125mg/6hour/IV, paracetamol syrup with a dose of 50 mg/8 hour/oral and domperidone syrup with a dose of 3x1 /2 cth. Therapy on the third day (on April 28, 2014) consisted of R/O2 1 liter/min, IVFD Asering with a dose of 14 drops/min, ampicillin with a dose of 125mg/6h/IV, 50cc Packed Red Cell (PRC) transfusion and paracetamol syrup with a dose of 50mg/8hr/oral. Treatment was given from the fourth day to the ninth day (April 29, 2014-4 May 2014), involved R/IVFD Kaen 3B with a dose of 10 drops/min and ampicillin with a dose of 125 mg/6 hours/IV. The patient returned home on May 5, 2014 at her family own request.

DISCUSSION

This infant was hospitalized with vomiting complaint since one day before the admission to the hospital. She also had fever since 6 hours before the admission. Laboratory results showed that this infant had anemia with a low Hb level of Hb 8.0 g/

dL (the Hb level for infants aged 6 months ranges from 10.4 to 15.6 gr/dL), a hypercholesterolemia level of 477 mg/dL and a severe hypertriglyceridemia level of 4370 mg/dL, a LDL cholesterol level of 135 mg/dL and a HDL cholesterol level of 5 mg/dL (Hypoalphalipoproteinemia). She also was prematurely born with SC due to the indication of preeclampsia of her mother and was born with a weight of 1.5 kg.

Anemia occurring in this infant can be considered as the coexistence of various causes of the disease. This patient suffered from an infection of her lungs (Bronchopneumonia), suspected to have lasted long enough to cause anemia. Fe deficiency actually can also cause anemia. In this infant, the reserves of iron were detected by her ferritin level of 45.35 ng/mL (reference ferritin level of 13–400 ng/mL). Meanwhile, serum Fe and TIBC were not checked to further confirm the diagnosis of anemia due to Fe deficiency.

In the blood chemistry examination, urea and creatinine levels of this infant were unreadable due to a very lipemic sample. Meanwhile, the method of urea and creatinine examinations is actually based on the principle of the spectrophotometer (color intensity). Thus, it can make the instrument disrupted in interpreting the results.

Next, this infant was given oxygen therapy to overcome her shortness of breath, fluid therapy for her dehydration, ampicillin antibiotics with a dose of 125 mg/6 hours/IV for the infection and transfusion of PRC 50 cc for her anemia. This patient was also administrated with 2 grams/day of colestyramine. The indication of colestyramine was to lower cholesterol in the blood.

This infant then returned home at her parent's own request. Unfortunately, the patient's parents refused to have their blood checked so that the exact diagnosis of the cause of hypertriglyceridemia was still unknown, but strongly suspected to be caused by primary causes. Hypertriglyceridemia in this infant was categorized as severe one according to NCEP, ATP III and Endocrine Society.

Severe hyperglycerididemia is usually caused by genetic disorders, but can also be caused by secondary causes, such as poor diet, alcohol use, obesity, metabolic syndrome, type 2 diabetes mellitus and hypothyroidism. Kurniawan, et al¹⁰ reported a very severe case of hypertriglyceridemia in type 2 diabetes mellitus (triglyceride 2.581 mg/dL). In this patient, hypertriglyceridemia was suspected to be caused by primary causes.

In patients with familial chylomicronemia, triglyceride level will be very high, more than 1000 mg/dL (> 11.2 mmol/L) and almost always caused by a high blood chylomicron level. The serum of this patient in this research was very lipemic and milky. when it was settled at 4°C (39.2°F) for several hours, the chylomicron had settled at the top of the serum, and then formed supernatant creamy. Serum triglyceride concentration is generally at > 1000 mg/dL (> 11.2 mmol/L), but can sometimes exceed into 10,000 mg/dL (112 mmol/L). Another lipid abnormality includes an increase in serum total cholesterol, whereas both LDL cholesterol and HDL cholesterol may be decreased or normal.

The ratio of cholesterol to triglycerides in patients with Familial Chylomicronemia (type I Hyperlipoproteinemia), according to lipoprotein patterns in the characteristics of inherited hyperlipidemia in Atlas of Metabolic Diseases,¹¹ is <0.2. In this patient, the ratio of cholesterol to triglycerides was 0.1, in line with the criteria of Familial Chylomicronemia (type I Hyperlipoproteinemia).

Familial Chylomicronemia often appears in infancy or childhood, and manifests during adolescence. Clinical symptoms of Familial Chylomicronemia are failure to thrive, eruptive xanthomas on extensor and buttock surfaces, retinal lipemia, hepatosplenomegaly, recurrent abdominal pain, nausea and vomiting, as well as acute pancreatitis. Less common clinical symptoms of Familial Chylomicronemia are intestinal bleeding, pallor, anemia, irritability, diarrhea, seizures and encephalopathy.

Based on the above explanation, it can be said that this infant could suffer from Familial Chylomicronemia. Some of its clinical symptoms were also found, such as anemia, vomiting and diarrhea. As a result, it is necessary to examine the electrophoresis of chylomicron and other lipid markers, such as apo C-II and VLDL of infants and other family members to strengthen further the diagnosis.

Familial chylomicronemia is a disease found in late childhood and adolescence, but has been reported also in infants and neonates. Early diagnosis is very important to prevent complications, such as acute pancreatitis, chronic pancreatitis, pancreatic necrosis,

and cardiovascular disease. Early diagnosis, drug administration and diet modification (15 grams of fat intake per day as well as the administrations of fish oil, fat-soluble vitamin supplements and lipid-lowering drugs) can improve prognosis and prevent complications in children with this disease.

The closest differential diagnosis to Familial Chylomicronemia (type I hyperlipoproteinemia) is usually mixed hyperlipidemia (Hyperlipoproteinemia type V). Examination of lipid profiles on the mixed hyperlipidemia will indicate milky serum with a very high triglyceride level of > 1000 mg/dL and a cholesterol-triglyceride ratio of 0.15: 0.60. The ratio of cholesterol to triglycerides in this patient was 0.1. Therefore, it did not fit the Mixed Hyperlipidemia category. Clinical symptoms of the Mixed Hyperlipidemia are actually similar to those of Familial Chylomicronemia. But, the difference is that Familial Chylomicronemia is more common in childhood or adolescence, while Mixed Hyperlipidaemia is more common in adulthood.

Unfortunately, there were some limitations in this research report. First, in this research, other lipid profiles of this infant, such as apoC-II and VLDL were not examined since she returned home at her family's own request. Second, lipid profiles of her parents and other families were also not checked since they refused to have their blood checked.

CONCLUSION

A 6-month-old female infant with a diagnosis of very severe hypertriglyceridemia (triglyceride 4370 mg/dL) was suspected to suffer from familial chylomicronemia. This infant had symptoms of vomiting, a history of diarrhea and anemia with a total cholesterol of 477 mg/dL, a direct LDL of 135 mg/dL and a HDL of 5 mg/dL. Next, parents and other family members of this infant should be screened to assess genetic inheritance patterns in establishing a definitive diagnosis of the cause of severe hypertriglyceridemia in this baby. Severe hyperglyceridemia can be caused by genetic and familial abnormalities, and can be suffered by several people in a family lineage.

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