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The female infant was the product of an uneventful 36-week pregnancy. Parents were non-consanguineous and healthy. At birth the child was unremarkable and had hepatomegaly with ascites. Laboratory studies revealed the following: total bilirubin 14.5 mg/dl (direct 0.5), albumin 1.7 mg/dl, prothrombin time 30", Factor II 16%, V 22%, VII 16%, X 22%, fibrinogen 114 mg/dl; ammoniemia was normal. Serum ferritin concentration was 1,915 mg/ml. Urinary succinylacetone was absent. Alpha-antitrypsin deficiency was excluded. All viral and serologic studies and cultures were negative. The patient was hospitalized and despite intensive management the child died on day 21 of life of diffuse uncontrolled cutaneous and mucous bleeding. Post mortem evaluation revealed significant hepatomegaly, iron deposits in the liver as well as in other main organs; extensive loss of parenchyma was evident; residual hepatocytes showed iron overload; giant cell transformation was also found. The pathologic examination confirmed the diagnosis of Neonatal Hemochromatosis (NH). NH (OMIM 231100) is an uncommon polycystic iron storage disorder of proratal onset. It is a phenotypically defined disease and it is characterized by insufficient iron stores in the liver and other main organs; NH phenotype is not uncommon. Skin biopsy is an effective screening tool, while demonstration of foamy histiocytes, cholesterol crystals in cytosol. Establishing an early diagnosis is invaluable. With limited treatment options, establishing an early diagnosis is invaluable.

Identification of two novel polymorphisms in the glucocerebrosidase gene region. E. Sidransky1, E. K. Lai1, S. Winfield2, D. Amiel3, M.A. Zimmaro7, N. Tayebe2, and E. I. Gins1. (1) Clinical Neurology Branch, IRP, NIH, Bethesda, MD; (2) Shaare Zedek Medical Center, Jerusalem, Israel.

Niemann-Pick Disease type C (NPC-C) is a lipidosis, is caused by a unique blockage of the intracellular degradation of sphingomyelin. This condition often causes diagnostic confusion in the early stages. We present 3 cases highly suggestive of such phytanic acid retention. Phytanic acid was detected in the urine and faeces of these children. Case 1: A 2 year and 9 month old boy presented with neonatal hepatitis, hepatopatotropamegaly and developmental delay. Initial investigations failed to establish a cause. A repeat study of the bone marrow showed foamy histiocytes, providing a diagnostic clue.

Case 2: A 14 year old boy presented with chronic megaloblastic anaemia, hepatopatotropamegaly and short stature. There were no neurological symptoms. Electron microscopic examination of muscle tissue showed complex lipid storage and cholesterol deposits in cytoplasm.

Case 3: A female infant born at 38 weeks gestation developed neonatal hepatitis at 4 months of age developed respiratory failure requiring ventilatory support until 2 years and 3 months. She had developed devalued general hypotonia and weakness. A muscle, skin and nerve biopsy showed lamellar inclusions suggestive of NPC-C.

In each of the three cases the definitive diagnosis was established by demonstration of impaired cholesterol esterification in skin fibroblasts. In conclusion, these cases illustrate the diverse, but common presentations of a rare disorder. Pulmonary manifestations (as in case 3) are rarely described in classical NPC-C, but believed to exist in infants dying during the neonatal period. Skin biopsy is an effective screening tool, while demonstration of foamy histiocytes, cholesterol crystals in cytosol.

In conclusion, these cases illustrate the diverse, but common presentations of a rare disorder. Pulmonary manifestations (as in case 3) are rarely described in classical NPC-C, but believed to exist in infants dying during the neonatal period. Skin biopsy is an effective screening tool, while demonstration of foamy histiocytes, cholesterol crystals in cytosol. Hepatomegaly and developmental delay. Initial investigations failed to establish a cause. A repeat study of the bone marrow showed foamy histiocytes, providing a diagnostic clue.

A CRITICAL EVALUATION OF COPPER METABOLISM IN INDIAN WILSON'S CHILDREN WITH SPECIAL REFERENCE TO THEIR PHENOTYPES AND RELATIVES. R. Prasad, G. Kaur and B.N.S. Walia. Department of Biochemistry and Paediatrics, Postgraduate Institute of Medical Education and Research, Chandigarh, India.

The female infant was the product of an uneventful 36-week pregnancy. Parents were non-consanguineous and healthy. At birth the child was unremarkable and had hepatomegaly with ascites. Laboratory studies revealed the following: total bilirubin 14.5 mg/dl (direct 0.5), albumin 1.7 mg/dl, prothrombin time 30", Factor II 16%, V 22%, VII 16%, X 22%, fibrinogen 114 mg/dl; ammoniemia was normal. Serum ferritin concentration was 1,915 mg/ml. Urinary succinylacetone was absent. Alpha-antitrypsin deficiency was excluded. All viral and serologic studies and cultures were negative. The patient was hospitalized and despite intensive management the child died on day 21 of life of diffuse uncontrolled cutaneous and mucous bleeding. Post mortem evaluation revealed significant hepatomegaly, iron deposits in the liver as well as in other main organs; extensive loss of parenchyma was evident; residual hepatocytes showed iron overload; giant cell transformation was also found. The pathologic examination confirmed the diagnosis of Neonatal Hemochromatosis (NH). NH (OMIM 231100) is an uncommon polycystic iron storage disorder of proratal onset. It is a phenotypically defined disease and it is characterized by insufficient iron stores in the liver and other main organs; NH phenotype is not uncommon. Skin biopsy is an effective screening tool, while demonstration of foamy histiocytes, cholesterol crystals in cytosol.