Histiocytosis X and Caroli’s Disease

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ABSTRACT

Both Caroli’s Syndrome and Langerhan’s cell histiocytosis can be associated with a variety of congenital anomalies. However, the association of the 2 diseases together has not been reported. We report a case of a 3 year old boy who had Caroli’s disease as diagnosed radiologically and Langerhan’s cell histiocytosis as diagnosed histologically.

Keywords: Caroli’s syndrome, Langerhan’s cell histiocytosis, pediatric, malignancy.

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Carolí’s Syndrome is a congenital non-familial malformation characterized by multifocal dilatation of the segmental bile ducts. Langerhan’s cell histiocytosis (LCH), Class I, previously called histiocytosis X, is a disease characterized by idiopathic proliferation (and accumulation) of specific cells of the monocyte-macrophage system producing focal and systemic manifestation. Both diseases can be accompanied by other congenital anomalies; but we are unaware of a case in which they were associated. We report a case of Caroli’s Syndrome and LCH which occurred in a 3-year old Saudi boy.

Case Report. A 3-year old Saudi boy was referred to our hospital with a chief complaint of failure to thrive and abdominal distension. This child was a full term baby with uneventful natal and postnatal period; at the age of 7 months he started to have bilateral otorhea, loss of appetite, loss of weight and seborrheic skin rashes over his scalp; at the age of 1 year he was diagnosed with diabetes insipidus and was treated with nebulizer vasopressin. After that the child’s general condition started to deteriorate with a low grade fever, jaundice and distended abdomen; a liver biopsy was carried out at this stage at the referring hospital which suggested cholestasis probably due to Caroli’s disease.

On examination, the child was underweight, jaundiced and had distended abdomen, due to hepatosplenomegaly; Liver function tests showed a cholestatic picture; ultrasound and CT scan documented the presence of cystic masses in intimate contact with the liver with intrahepatic dilatation, the diagnosis was that of Caroli’s diseases. A laparotomy was carried out to excise the cyst and carry out biliary drainage; the cyst was removed and operative cholangiogram through a butterfly needle in the common bile duct (CBD) showed the presence of 3 stones in the CBD. They were removed and T-tube was inserted. No permanent biliary enteric anastomosis was carried out. After the removal of the T-tube a permanent fistula was formed which was resistant to all types of treatment including cholecystojejunostomy carried out on 2nd surgery. It was only controlled by Sandostatin. Histopathology of the excised cyst supported the diagnosis of Carolí’s disease.

A year later generalized seborrhic skin rashes on the scalp and all over the body suggested LCH. Skin biopsy proved the diagnosis by showing infiltration of the superficial dermis by extensive histiocytic infiltration. The infiltrate invaded the epidermis. The histiocysts appeared long, rounded with abundant eosinophilic cytoplasm and eccentric indented nuclei. Immunohistochemical staining confirmed the
diagnosis of LCH, and bone marrow aspiration showed marked hypercellular marrow with increased iron stores, megakaryocytes increased in number; normoblastic erythropoiesis and active granulopoiesis. There was a slight increase in band forms with toxic changes. There was proliferation of LCH with moderate grayish blue cytoplasm not showing any phagocytotic picture. The nuclei contained 1-2 nucleoli, plasma cell were prominent, PAS stain was negative but acid phosphatase was positive. This bone marrow picture is confirmation of generalized hyperplastic bone marrow with LCH involvement. The liver biopsy taken at the referring hospital was negative for LCH. The general condition of the patient deteriorated, he received 2 doses of Vinblastin I.V. before he succumbed from fulminant liver failure.

**Discussion.** Langerhan's cell histiocytosis, Class I, previously called histiocytosis X, refers to a spectrum of diseases characterized by idiopathic proliferation of a specific cell of the monocyte-macrophage system producing focal or systemic manifestation. Causes and pathogenesis remain unclear. However, recent studies suggest abnormal immune regulation as an important factor. The disease includes 3 recognized clinical syndromes, which may have considerable clinical overlap: eosinophilic granuloma, in which the disease is limited to bone in patients usually 5-15 years old; Hand-Schuller-Christian disease occurs in young children, and the classic triad of osteolytic defect in membranous bones, exophthalmos and diabetes insipidus; the disseminated form of LCH, the Letterer-Siwe-Syndrome, which is usually encountered in infants. The coexistence of congenital anomalies with LCH was noted by Greenburger et al.; such coexistence was documented by Shells; compared to 2 control groups, they found that there is increased frequency of congenital anomalies in histiocytic patients. Most of the congenital anomalies are discovered at the time of the diagnosis of histiocytosis unlike our patient, when the congenital anomaly was discovered about a year before the disease. The anomalies can be multiple or single; although various types of congenital anomalies were found to coexist with LCH; no case of Caroli's disease was mentioned in the literature from 1980. Children with LCH and congenital anomalies have a greater chance of developing organ dysfunction in the course of their diseases.

The outcome of LCH is extremely variable, but in most cases it is a self-resolving process. The 2 prognostic factors appear to be age and degree of organ involvement. Children under 2 years have a higher mortality. Presence of organ dysfunction (e.g. liver, lungs, bone marrow) was found to be a poor prognostic sign, children with these poor prognostic features constitute fewer than 15%. Chemotherapy is only used in those patients who have major organ dysfunction or progressive disease. The reported patient received 2 doses of vinblastin intravenously because he had bone marrow involvement, we were not sure if he had liver involvement, or not since the liver biopsy taken at the beginning of his disease was negative and another biopsy at the later stage was not taken. The reason for his permanent fistula after his first surgery was probably because of the involvement of the tract by LCH. We think we report the first case in world literature in which LCH and Caroli's disease coexisted.

**References**