[Lipid Storage Disease]

GRAND ROUNDS

January 6, 1960

[redacted]

Birth - FTND 6 lb. 7 oz.
4 wks.- Diarrhea, vomiting, no response to formula change.
5 wks.- Swelling of extremities - rash followed by purpura.
Adm. 6 wks.
Px - Pale lethargic edematous, weak cry, purpura. Scaly, crusting seborrheic scalp lesions. Maclopapular xanthomatous rash with hemorrhagic base over skin, liver down 2 cm, spleen 2 cm, 4+ pitting edema-marked.
Lab.-Hgb. 10.4. WBC 12,500. Platelets 68,000. Urine-neg. Hgb. next a.m. = 5.6 gms. Blood culture, stool culture, nasopharyngeal culture, B. hemolytic strep. BUN = 30. TSP = 1.44 with A/G = 0.93/0.51. Electrophoresis - normal but low. pH = 7.2. CO_2 = 10.8 Cl = 107 Na-120 P=7.7. Bone marrow showed granulocytic hyperplasia.
Rx - Antibiotics, steroids, transfusions.
Hosp. Course - child developed massive GI hemorrhage and died the morning after admission.
Autopsy - moderate to marked infiltration of spleen, lymph nodes, liver, lung, thymus, skin with histiocytes and loss of architecture.
Dx - Acute fulminant L.S. disease complicated by B. hemolytic strep sepsis.

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Birth - FTND - 8 lbs.
11 wks. - Anemic, diarrhea, respiratory infections, Rx - transfused.
13 wks. - Anemic. Rx-transfused.
5½ mos. - Pale, liver down 3 cm-otherwise O.K.
21 mos. - Thrombocytopenia with purpura Rx-steroids-recovery.
3 yrs. - Good health-history of tiring easily-CBC normal.
3½ yrs. - Rash for 1 month which caused pitted scars.
4½ yrs. - Large abdomen - 4 mo. PTA.
1 mo.PTA.-Purpura large liver and spleen.
2 wks.PTA-Hematuria plus large liver and spleen-purpura-Rx.steroids.
3 d.PTA - High spiking temp.
Adm. 4 yrs. 10 mos. -
Px - Temp. 103.6, chronically ill. Purpura L & S down to the pelvic brim. Mild generalized adenopathy.
Lab.-Hgb. - 4.9 WBC=20,300. Platelet = 40,000. Urine = 30 Red RBC's - otherwise negative. Bone marrow showed increased NRBC's with marked decrease in megakaryocytes - Xrays neg. Multiple seralogical tests were neg. Multiple cultures were neg.

Rx - Pcn, chloro, dihydrostrep, sulfa, achro - no chg. temp. curve. Meticorten - temp. to normal. Exp. lap. with splenectomy, liver biopsy, muscle biopsy, node biopsy - Diagnosis-reticuloendothelial hyperplasia with infarcs.

5 yrs. - Suddenly expired PMD's office.

Autopsy -Generalized replacement of normal architecture of lymph nodes, with marked infiltration and distortion of architecture of liver, lungs, bone marrow, some in adrenals and kidneys by a malignant reticuloendothelial process, with multiple infarcs in lungs, lymph nodes, etc.

Imp. - Maligant reticuloendothelialosis - L.S. in type.

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18 mos. - Swelling of jaw plus neck. Fever, irritable.

Adm. 19 mos.

Px - Tender 3 x 6 cm mass rt. submendibular area - cervical adenopathy - rt > left.

Xray-mandible - "Tumor involving mandible".

Rx -Tumor excised, lymph node excised - histological exam. showed marked infiltration and replacement of normal architecture with histiocytes and EOS. The pathological interpretation was EOS gran. of bone and lymph node.

3 yrs. -Diabetes Insipidus.

Adm. 4 yrs. -Meningococcal meningitis.

5 yrs. - ? Tender painful mass left inquinal area which finally cleared.

5 yrs. - ? Tender painful mass of left scapula which finally cleared.

5½ yrs.-Mass of left side of neck- slowly progressive in size.

Adm. 6 yrs. -Adm. with history of trauma to masses left side of neck, fever for two days.

Px - Large, tender lymph nodes 1 to 3 cm, anterior and posterior cervical chains.

Rx - Lymph nodes excised. Exam. of the lymph node showed a moderate infiltration of the lymph node with cells containing small and large vacuoles, eosinphilic infiltration was again noted, stain of the sections with Sudan IV showed many of the macrophages with droplets. Typical foam cells were not seen. Diagosis was granulomatous inflamation of lymph nodes, HSCD in type. No follow-up on this patient.

Imp. - EGB initially followed by HSCD involvement of lymph node.

BIBLIOGRAPHY

Etiology

Siperstein-1958. Discussed these diseases as a possible lipoid disturbance pointing out that the etiology of the cholesterol found in the HSCD lesions might be due to an increase in output of cholesterol by histiocytes or, of course, an increase in utilization of cholesterol by histiocytes. He also pointed out that the serum cholesterol was usually normal.

Adv Ped 6:209-1953. These diseases reviewed under lipoidosis but the author was unsure as to their etiology. A good general review.

Ped 8:573-1951. Gives good differential diagnosis of xanthomathoses and pointed out that xanthoma may be due to multiple etiology.

Acta Ped 46:471-1957. Pointed out the occa.. familial occurrance of LSD and discussed whether it was a lipoid storage disease or possibly a leukemic-like manifestation.

Pathology

Ped 8:573-1951. A good pathological description is given with a differential diagnosis of the xanthomathoses.

Adv Ped 4:117-1949. Presents these as 3 separate diseases and feels the diseases should be classified according to the clinical manifestations rather than the pathological appearance until the etiology of the disease is known.

AMA Arch Path 56:84-1953. Extensive review comparing findings in all 3 diseases. Proposes term "Histiocytosis X." He feels that the diseases may be varying manifestations of an unknown histiocytic disease.

NEJM 257:1082-1952. Report of eosinophilic granuloma of lung with review of literature.

Am J Path 37:99-1944. Discusses EGB and HSCD. Believes that they are the same disease.

AJDC 60:471-1940. Presents 2 cases LSD with HSCD characteristics. Believes they are the same.

Arch Patho 63:49-1957. Reports the occurrence of LSD in a 57 year old white male with review of other similar cases in adults.

Diagnosis

Ped 8:573-1951. Good DDX of xanthomatous diseases.

AJM 22:636-1957. Reports 40 cases of EGB, HSCD, and LSD, with physical findings, course and prognosis.

AJDC 90:323-1955. Excellent review of LSD with 15 cases reported.

RAD 71:525-1958. Reports incidence of GI involvement. 7 of 11 LSD (6 symptomatic), 2 of 5 transitional forms showed GI involvement.

AJM 22: 834-1957. Importance of radiological survey with extensive bilateral pulmonary disease of unknown origin.

Ped 19:438-1957. Discusses skin scraping as a method of diagnosis in all suspicious eczematoid-like lesions with biopsy if indicated.

AMA Arch of Derm 78:662-1958. A case of HSCD in a 43 year old negro female with initial symptoms of eczema.

Treatment

J Bone & Jt Surg 24:499-1942. Describes 9 cases of EGB all of whom healed spontaneously or with Xray.

Post Grad Med 12:427-1952. Use of nitrogen mustard in treatment of HSCD and LSD.

J Ped 40:269-1952. Treatment of LSD in twins with Aureo and Strep with recovery.

SLCH-1955. Treatment of HSCD with INH with good results.

Gr Or St J 10:104-1955. Treatment of LSD with Cortisone with good results.

AJDC 90:325-1955. Treatment of LSD with Chloro with recovery in 2 cases.

AJM 22:834-1957. Xray therapy for pulmonary lesions.