## MEDICAL GRAND ROUNDS

Wednesday, October 29, 1958

was first seen at the age of 4 years. She was born prematurely, weighing 4 lbs. 11 oz., after a prolonged labor. There was a tendency to vomit feedings frequently in early infancy. At 3 months of age her hair became brittle and would grow no longer than  $\frac{1}{2}$  to 1 inch before breaking off. Her general development was normal except for delayed closure of the anterior cranial fontanelle. At the age of 2 years she had pneumonia complicated by 2 convulsions. At about this time a tentative diagnosis of hypothyroidism was made. Thyroid medication was prescribed, and continued for the next  $1\frac{1}{2}$  years without appreciable effect. She has taken very little milk since that time.

At the age of  $3\frac{1}{2}$  years she began to have generalized convulsions. Each episode lasted about 1 minute, and was characterized by clonic movements of the extremities, clenching of the fists, twitching of the left eyelid, loss of consciousness, and brief postictal weakness. These occurred at the rate of 20 to 30 per day despite the administration of dilantin and phenobarbital, but ceased promptly when a calcium preparation was given intravenously. The exact levels of calcium and phosphorus in the blood serum are not known. Thyroid therapy was stopped.

Treatment with an oral calcium preparation and thrice-weekly injections of parathyroid hormone was started, and the patient remained asymptomatic until about a week prior to admission. At this time she began to lose her hair in large amounts. All treatment was stopped, and she was referred to the hospital for further evaluation.

There is 1 normal sibling. The paternal grandmother and 4 of her siblings have diabetes. No one in the family has difficulties resembling those of the patient.

On admission to the she weighed 13.9 kg. and was 79 cm. tall. There was a striking loss of hair from the eyebrow region and scalp. There were no signs of tetany. The skin was dry, and the nails, teeth, and eyes were normal. The liver edge could be felt  $1\frac{1}{2}$  cm. below the right costal margin. There were no other physical findings of note.

Laboratory examination revealed a normal blood count, negative urinalysis (sp. gr. 1.040), and negative flocculation text for syphilis. The blood urea nitrogen and total serum proteins were normal. The serum calcium was 7.6 mg. per cent and the phosphorus 4.3 mg. per cent; alkaline phosphatase was 10.6 Bodansky units. The 24-hour urinary excretion of calcium was 14 mg. and that of phosphorus 232 mg., and on a subsequent occasion she excreted 45 mg. phosphorus in a 12-hour period. The response in urinary phosphorus excretion to the giving of 200 units of parathyroid

hormone intravenously was definite. Roentgenograms of the skull and long bones were normal. Electroencephalogram revealed generalized high-voltage slow activity, most pronounced in the right posterior region, but without paroxysms.

Other tests showed a 24-hour radioactive iodine uptake of 20 per cent, a 50 per cent drop in circulating eosinophiles after intramuscular corticotropin, and a normal electrocardiogram.

Treatment with A.T. 10 and calcium was recommended. The only follow-up details are that her general condition is good, though she remains completely bald.

Case II (Seen in Cincinnati and reported in the literature by Dr. Harmon.)

A six year old Negro boy was admitted to in 1953. He had listlessness, weakness (especially in the legs) apathy, inertia, abdominal pain, pain in the extremities, and recurrent vomiting. He was losing weight. (36 lbs.)

Physical examination found him to be a dehydrated irritable child, disorientated, with slightly distended and tender abdomen. Blood pressure at times rose to 150/100, pulse 50 (Bradycardia).

Several weeks passed by before the correct diagnosis was made on this patient. The laboratory investigation was found to yield the following data: \_

High W.B.C. (15,000 leucocytes) (81% neutrophils)
EEG showed dysrhythmia. Poor urine concentrating ability
with hyaline casts. Presence of W.B.C. and R. B. C.
Alb. 3+. GFR reduced. Also Tm (p.a.h.) Serum electrolyte
concentrations.

C1 85 mEq/L CO<sub>2</sub> 29 mEq/L Ca excretion less than Na 140 mEq/L Ca 13 to 20 mg./ 300 mg/day. c.s.f. 68 K 4.9 mEq/L P 2 mEq/L mononuclear cells. Mg 0.2 mEq/L BUN 43 mg./ 77 mg/ protein

W.R. 34. All serological and other tests for infection negative. Slightly raised alkaline phosphatase.

Bone X-rays: Homogeneous glass like appearance - osteoporosis.

(numerous osteoblasts seen on histologic examination of bone).

## Course in Hospital: (Briefly)

Became semicomatose, or had occasional bouts of grand mal convulsions - developed blindness (cortical). Developed heart block (EKG) - for one week.

Parathyroid adenoma removed at operation.