CASE #8

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This 16-month-old male was born to a gravida III, para III mother after a 38-week gestation and normal delivery. Birth weight was 6 lbs. 7 oz. The head was small and eyes microphthalmic. He was, otherwise, well and discharged on the third hospital day.

At eight weeks, he was hospitalized for UTI. Head circumference was 37.5 cm. Weight was 8 lbs. 12 oz. (3rd %). Eyes were sunken with large non-reactive pupils. Eye movements were conjugate but there was no reaction to threat. Retinal degeneration with macular changes were described. Skull x-rays, LP, CMV, Rubella and toxoplasmosis titers were all normal. Chromosomal studies showed enlarged short arms on some of the D chromosomes.

At 11 months, he was hospitalized for severe weight loss attributed to vomiting and diarrhea recognized from 7 months of age. He weighed 12 lbs. 4 oz. and was hypotonic, lacking head control but responsive to sounds. Spontaneous limb movements were full and tendon reflexes and sensation were preserved. EEG was slow and irregular. Suppression of alpha waves with the eyes open implied some preserved vision. Pneumoencephalogram showed cerebral atrophy (or hypoplasia) without calcifications. Stools were negative for fat, blood and pathogens, BUN, blood sugar, T_4, electrolytes, Ca, P, ALKP and sweat chloride were all normal. Proteins were 4.4 Gm.%, Alb. 3.3 Gm.%. Immunoglobulins and lipoprotein electrophoresis were normal. Xylose absorption was less than 1%. GI series and barium enema showed slow transit time and dilatation of small bowel loops. Hyperalimentation failed to produce consistent weight gain and reintroduction of oral feedings resulted in severe diarrhea.

He remained awake, alert and sociable but never gained useful movements, head control or speech. No focal or specific cerebellar or long tract signs were elicited. He died at 16 months of age weighing 11 lbs. Brain weighed 550 Gm. (nl 1010.)

Microscopic section of cerebellum stained with hematoxylin and eosin.

Points for discussion:

1. Can microphthalmis, retinal degeneration with macular changes and granuloprival cerebellar cortical atrophy be developmental anomalies related to a single teratogenic insult?
2. Is there a relationship between malnutrition and cerebellar cortical atrophy?
3. How is this granuloprival atrophy related to other congenital cases presenting as a static psychomotor retardation?
4. Is this Minamata disease?
5. If analagous to animal and experimental granuloprival degeneration due to viral or chemotherapeutic agents, what is the limiting human perinatal time interval during which granular cell injury might be acquired? What is the likely agent?