CASE 4

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This female, first-born infant, was delivered two months prematurely (wt 1900 gm.) Family history negative. The first year of life was uneventful, although in retrospect the veins of the left forehead always were rather prominent. Fever and the first of several 30-minute episodes of deep lethargy led to hospitalization at 13 months of age. OFC then was 48 cm, and there were somewhat large, non-pulsating veins in the optic fundi; otherwise the child was normal. At 17 months the left eye had become prominent; OFC was unchanged, and psychomotor impairment was now obvious. Irregular calcifications of the frontal lobes of a peculiar, punctate, and serpiginous type were first seen at 19 months. The left superior orbital fissure was widened. Air study revealed normal ventricles; angiography was not helpful. Cerebrospinal fluid protein: 73 mgm, slight increase in globulin. Chemical studies, including Ca and P, revealed no changes. The further course was one of steady retrogression with increase in calcifications. A serum test for toxoplasmosis was negative. The child died early in 1966, 3 years old.

Autopsy findings: The left eye was protuberant and enlarged veins, normal in distribution, were noted in the left orbit. The brain was small (700 gm) but normal in shape. There were large and abnormal veins over the surface and two frank small hemangiomas in the right uncus and in the cerebellum. The basal ganglia contained many enlarged veins. There were cavities of the white matter, mostly in the frontal lobes with white slivers of calcium. Small specks of calcific material were also visible, especially under slight magnification, at or slightly below the corticomedullary junction -- a finding confirmed in low KV x-rays of the specimen. The remainder of the autopsy was non-contributory.

Diagnostically, we consider the possibility of a widespread, transcortical arteriovenous shunting with calcifications in and around the abnormal vessels.

Dr. Muller's diagram

2 sections are submitted, one stained with H & E, the other with a modified reticulin method.