RADIOLOGIC CASE PRESENTATION

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The patient was a three-month-old child who was well until the age of two months, at which time he temporarily became limp and unresponsive. He was taken to another hospital where work-up showed an abnormal electroencephalogram and peculiar wormian bones on the skull X ray. He was given oral anti-convulsants. The child subsequently developed excessive drooling and difficulty in swallowing.

Approximately one month later he became unresponsive and developed major motor seizures with twitching of the right side of his face. Physical examination at this time demonstrated a lethargic and hypotonic male child with bilateral sustained ankle clonus. His hair was noted to be short, brittle and kinky. There were wide temperature fluctuations between 97° and 101° and an occasional instance, by past history, of the temperature being 94°. Past history was uncomplicated with a full-term normal pregnancy.

Laboratory findings showed a serum copper level of 11.9 micrograms% (normal, 70-100 micrograms%) and a ceruloplasmin level of 4 milligrams% (normal, 18-52 milligrams%), liver function including alkaline phosphotase of 361, SGOT 26 and LDH 27, jejunal biopsy negative and oral calcium 64; isotope absorption revealed markedly diminished absorption of the isotope.

Radiographs revealed flared and cupped rib ends with hook-like deformities of metaphyses of the long bones, perhaps representing healed metaphyseal fractures or unusual stress upon these bones. There was generalized demineralization present. Cerebral angiography revealed tortuous vessels with considerable narrowing and dilatation along with great separation between brain substance and calvarium, suggesting diminished brain substance (Fig. 1).

Abdominal aortogram demonstrated markedly tortuous and irregularly shaped mesenteric and renal vessels with intermittent areas of constriction and dilatation (Fig. 2). The child's physical and mental status continued to deteriorate and he expired several months later.

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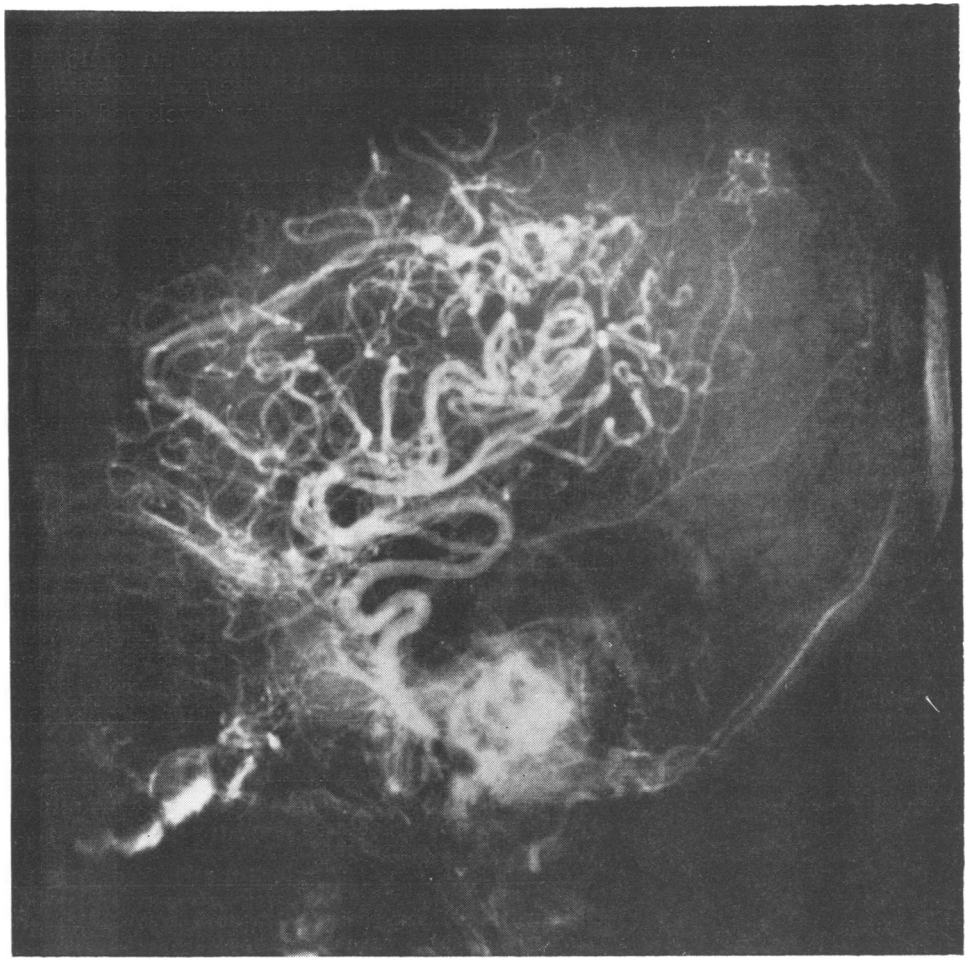

Fig. 1. Cerebral angiography revealing tortuous vessels with considerable narrowing and dilatation along with great separation between the brain substance and calvarium, suggesting diminished brain substance.

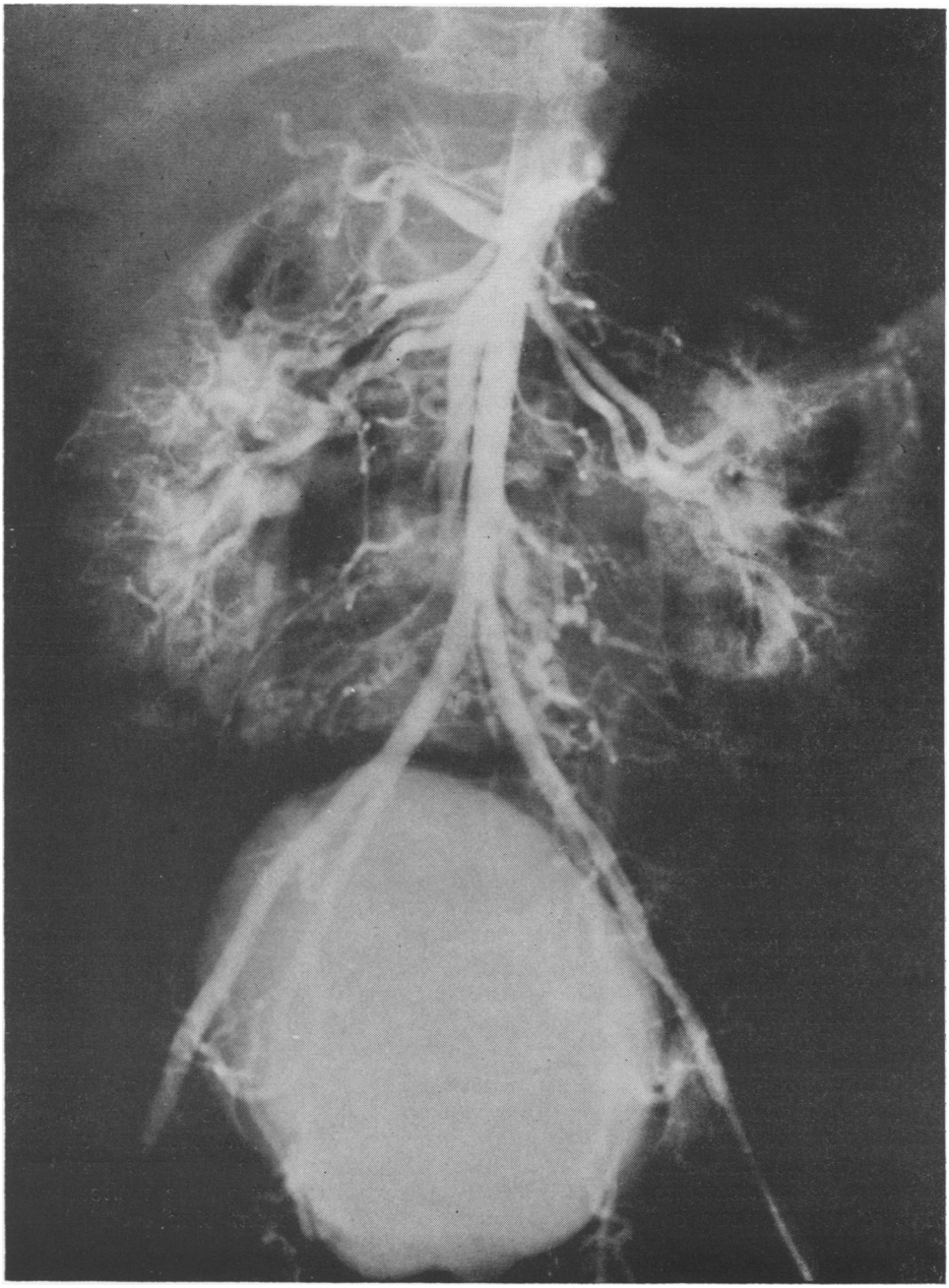

Fig. 2. Abdominal aortogram demonstrating markedly tortuous and irregularly shaped mesenteric and renal vessels with intermittent areas of constriction and dilatation.

ANSWER

Menkes Kinky Hair Syndrome

The Kinky Hair syndrome was originally described by Menkes in 1962 and is a sex-linked, recessive, neurodegenerative disorder. The syndrome includes seizures, mental retardation, skeletal deformities, hypothermia, abnormal hair, and peculiar tortuous arteries. The skeletal abnormalities, which may simulate the battered child syndrome or rachitiform diseases, are manifestations of copper deficiency, an element necessary for normal skeletal growth.

It is unknown whether the abnormal appearance of the arterial system precedes the nervous system degeneration or is a result of this deterioration. However, because the arterial pattern does not correspond to any of the known patterns in cerebral atrophy, the arterial aberration presumably is a congenital malformation. This case illustrates the value of angiography in more definitively diagnosing specific degenerative diseases.

REFERENCES

1. Danks DM, Campbell PE, Stevens BJ, Mayne V, and Cartwright E: Menkes kinky hair syndrome: An inherited defect in copper absorption with widespread effects. Pediatrics, 50:188-201, 1972
2. Graham GG, and Cordano A: Copper depletion and deficiency in the malnourished infant. Johns Hopkins Med J, 124:139-150, 1969
3. Menkes JH, Alter M, Steigleder GK, Weakley DR, and Sung JH: A sex-linked recessive disorder with retardation of growth, peculiar hair and focal cerebral and cerebellar degeneration. Pediatrics, 29:764-779, 1962
4. Wesenberg RL, Gwinn JL, and Barnes GR Jr: Radiological findings in the kinky hair syndrome. Radiology, 92:500-506, 1969

LETTER TO THE EDITOR

Because of the rarity and unique features of the mucocutaneous lymph node syndrome and because only recently has this condition been recognized in the United States, we think it necessary to provide additional information regarding the Radiologic Case Presentation in Cardiovascular Diseases, Bulletin of the Texas Heart Institute, Volume 2, Number 4, 1975.

The diagnosis and angiographic studies were made by Michael Nihill, M.D., and Dan G. McNamara, M.D., of the Cardiology Service of Texas Children's Hospital. We regret that this was not acknowledged at the time of publication. Also, the complication of "gangrene of right arm and leg" were complications of the patient's disease and not of the catheterization, as might have been misunderstood by our presentation.

Sincerely yours,

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