

Vascular Occlusion Associated With Incontinentia Pigmenti

Richard J. Pellegrino, MD* and Ami J. Shah, MD†

A 4½-year-old girl with incontinentia pigmenti developed acute-onset hemiparesis. Magnetic resonance angiography revealed occlusion of the left middle cerebral artery. Many neurocutaneous syndromes have been linked to ischemic stroke syndromes. Incontinentia pigmenti has not been associated with stroke syndromes in the past; however, this report illustrates that the patient and her mother, who also has incontinentia pigmenti, may have had similar ischemic stroke events.

Pellegrino RJ, Shah AJ. Vascular occlusion associated with incontinentia pigmenti. *Pediatr Neurol* 1994;10:73-74.

Introduction

Incontinentia pigmenti (Bloch-Sulzberger syndrome) is a rare neurocutaneous syndrome characterized by congenital skin lesions, dental anomalies, and scalp and ocular abnormalities [1]. About 30-50% of patients have evidence of central nervous system (CNS) involvement, including mental retardation, seizures, microcephaly, and cerebellar ataxia [1,2].

We report 1 patient who presented with an ischemic stroke syndrome related to incontinentia pigmenti.

Case Report

This 4½-year-old white female presented with acute onset of hemiparesis. She had been in her usual state of good health until 2 days prior to admission when she began to complain of nausea. A flu-like syndrome was manifest in the family at the same time. One day prior to admission the patient experienced difficulty with speech. She was unable to respond to questions, but was able to follow commands. On the morning of admission, she was unable to bear weight, falling to the right side. She had no history of fever, abdominal pain, nausea, vomiting, visual disturbance, or trauma.

The patient had been born to a 35-year-old female (1 spontaneous abortion at 11 wks estimated gestational age). No prenatal complications occurred, but the medical history was notable for incontinentia pigmenti

diagnosed at birth. The patient's development was normal. Recent findings included mild alopecia, anodontia, and streaking skin lesions. She also had hip dysplasias which were treated.

Family History. The patient's mother had a paralytic episode as a child which resolved. The episode was attributed to polio, but the mother had not been diagnosed at that time with incontinentia pigmenti which was later determined in adulthood.

Physical Examination. The general physical examination was normal except for alopecia, cone-shaped teeth, and marbled pigmentation of the skin of the right leg. Neurologic examination disclosed weakness of the right limbs, right central facial paresis, and right homonymous hemianopsia. Deep tendon reflexes were absent on the right and 2+ on the left and abdominal reflexes were absent on the right and intact on the left. Hemogram and electrolytes were within normal limits. Lumbar puncture revealed 10 leukocytes/mm³ which were 100% monocytes; no erythrocytes were observed.

CT demonstrated an infarct of the left frontoparietal area. MRI revealed increased intensity in the subcortical area, possibly due to decreased flow (Fig 1). MRA disclosed occlusion of the proximal aspect of the left middle cerebral artery. There appeared to be some flow within the left middle cerebral branches, possibly representing retrograde collateral flow (Fig 2).

Hospital Course. Because of CSF pleocytosis, the patient was initially begun on cefotaxime which was discontinued after 3 days of negative cultures. The patient improved slowly throughout her hospital course. She remained in the hospital for 5 days, during which time she regained some function of her right arm. Her right central facial paresis improved somewhat; however, right homonymous hemianopsia and aphasia persisted.

After being discharged, she experienced 2 right-sided focal seizures which consisted of eyes rolling up and stiffening of the right arm. She also had some staring episodes. She now remains on antiepileptic medication and her seizures are well controlled.

Discussion

Incontinentia pigmenti is an inherited neurocutaneous disorder of uncertain transmission. It has been suggested that it is a sex-linked disorder which is lethal in males [3]. The female-to-male ratio is 35:1 [4]. Of 465 patients studied, 30.5% had CNS disease. Among these, 16.6% had motor abnormalities including slow motor development, psychomotor retardation, spastic tetraplegia, hemiplegia, and diplegia. A few patients also had abnormal EEGs but no specific abnormality was observed [1]. Our patient exhibited many other signs of incontinentia pigmenti, including alopecia (38% of patients), anodontia (43%), and streaking skin lesions (about 93%) [1]. MRA demonstrated partial occlusion of the left middle cerebral artery. Although other neurocutaneous syndromes, such as neurofibromatosis, tuberous sclerosis, Sturge-Weber syndrome, and Klippel-Trenaunay-Weber syndrome, have been associated with ischemic strokes, incontinentia pigmenti has not [5].

This patient, like her mother, exhibited an acute onset of hemiparesis, which through noninvasive studies was attributed to partial occlusion of the left middle cerebral artery. Because the patient had no known cause of isch-

From the *Department of Pediatrics; UCLA School of Medicine; †Department of Pediatrics; Children's Hospital of Los Angeles; Los Angeles, California.

Communications should be addressed to:
Dr. Pellegrino; Pediatric Neurology; 18411 Clark Street; Suite 201;
Tarzana, CA 91356.

Received November 11, 1992; accepted August 17, 1993.



Figure 1. MRI performed on day of admission revealed increased intensity in the subcortical area, on the left side (arrow) due to decreased flow in the middle cerebral artery.

emic stroke (e.g., infection, heart disease, trauma, hemoglobinopathies), it seemed plausible to relate her stroke to incontinentia pigmenti, particularly because her mother may have had a similar episode in childhood. Many patients with incontinentia pigmenti have been reported to have seizures [1]. It is possible that some incontinentia pigmenti patients have seizures on the basis of ischemic stroke. Perivascular inflammatory infiltrates have been reported in skin biopsies of patients with incontinentia pigmenti, raising the possibility that a similar inflammatory lesion in the brain is responsible for occlusive stroke in incontinentia pigmenti. Incontinentia pigmenti should be included along with other neurocutaneous syndromes in

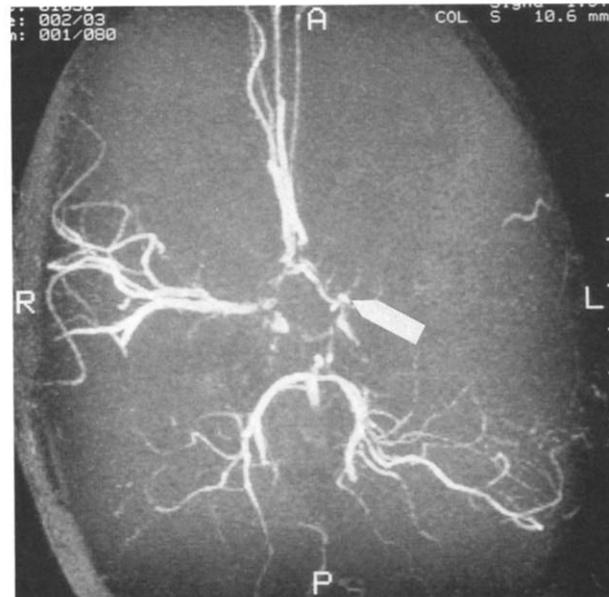


Figure 2. MRA performed 2 days after admission revealed apparent occlusion of the proximal aspect of the left middle cerebral artery (arrow). There appears to be some flow within left middle cerebral branches possibly representing retrograde collateral flow or possibly due to diminished flow through a high-grade stenosis as opposed to total occlusion of the proximal middle cerebral artery.

the differential diagnosis of ischemic strokes in females. Further evaluations will be necessary to determine the mechanism of stroke syndromes in incontinentia pigmenti.

The authors thank Heather MacEwen, Tiffany Bluestone, and the Department of Photography at Children's Hospital of Los Angeles for their help.

References

- [1] Carney RG Jr. Incontinentia pigmenti: A world statistical analysis. *Arch Dermatol* 1976;112:535-42.
- [2] Avrahami E, Harel S, Jurgenson U, Cohn D. Computed tomographic demonstration of brain changes in incontinentia pigmenti. *Am J Dis Child* 1985;139:372-4.
- [3] Lenz W. Half chromatid mutations may explain incontinentia pigmenti in males. *Am J Hum Genet* 1975;27:690-1.
- [4] Cohen BA. Incontinentia pigmenti. *Neurol Clin* 1987;5:361-77.
- [5] Tresher W. Ischemic stroke syndromes in childhood. *Pediatr Ann* 1992;21:375-83.