Congenital defects of the posterior arch of the atlas: a report of seven cases including an affected mother and son.

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Congenital Defects of the Posterior Arch of the Atlas: A Report of Seven Cases Including an Affected Mother and Son

Guido Currarino, Nancy Rollins, and Jan T. Diehl

PURPOSE: To describe our experience with congenital anomalies of the posterior arch of the atlas, with a review and classification of these defects and a note on their clinical significance. **METHODS:** We report six children and one adult, the mother of one of the children, with an anomalous posterior arch of the atlas. The diagnosis was made on lateral films of the neck. Three patients also had axial CT of the cervical spine. **RESULTS:** The anomalies encountered in the seven patients were absence of the posterior arch of the atlas (four patients), bilateral clefts (two patients), and unilateral cleft (one patient). In three patients the anomaly was discovered as an incidental asymptomatic finding; three other patients presented with transient neck pain or transient neurologic symptoms after head and neck trauma, and one patient (an adult woman) described neck symptoms of 1-year duration. **CONCLUSIONS:** On the basis of these seven cases we conclude that congenital defects of the posterior arch of the atlas may be discovered as incidental asymptomatic findings, but symptoms occurring after trauma to the head and neck or spontaneously also may be encountered.

Index terms: Spine, abnormalities and anomalies; Spine, vertebrae; Atlas and axis; Pediatric neuroradiology

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Congenital abnormalities of the posterior arch of the atlas (C-1) are very uncommon and not widely known. They are many case reports in the literature, but little mention is made of them in radiologic texts. We report seven cases with a review of the literature and an anatomic classification of these defects. It was also our intent to address the issue of the clinical significance and prognosis of these abnormalities of the atlas.

Materials and Methods

Seven patients with an anomalous posterior arch of C-1 were diagnosed between 1982 and 1991; six were children ranging in age from 20 months to 12.5 years, and one was an adult, the mother of one of the children. Of the seven cases, six were originally seen and diagnosed at our hospital, and one (case 4) whose films were sent to us for consultation was studied in another institution. The diagnosis was made in all patients on lateral films of the neck. Three patients also had axial computed tomography (CT) of the cervical spine, which demonstrated the anomaly in more detail. Our study of the clinical significance of these anomalies was based on the initial and follow-up information obtained in our patients and on information gathered from patients described in the literature.

Results

The initial clinical and plain film and CT findings and follow-up information on the seven patients are shown in Table 1. The first four patients had an absence of the entire posterior arch of C-1, except its most anterior part(s) near the lateral masses in two cases (cases 1 and 2) (Figs 1 and 2). Patients 5 and 6 had bilateral clefts in the posterior arches, and patient 7 had a unilateral cleft. In the four patients with absence of the posterior arch of C-1 including posterior tubercle (cases 1 to 4) (Figs 1, 2, 3, and 4) there was an associated cephalad elongation of the spinous process of C-2, and in three of them (cases 1, 2, and 4) a faint radiolucent or dense line across this superior prominence (or a notch in the contour

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TABLE 1: Clinical and plain film/CT findings in the seven patients reported in this paper

Case	Fig	Age	Sex	Reasons for Neck Films	Physical exam and follow-up	Plain Film/CT Findings
1	1	20 months	F	Possible epiglottitis	Normal physical exam	Absence of posterior arch of C-1 except its most anterior part(s) (type E, Fig 8)
2	2	4 years	M	Evaluate size of adenoids	Normal physical exam	Absence of posterior arch of C-1 except its most anterior part(s) (type E, Fig 8)
3	3	4.5 years	M	Evaluate size of adenoids	Normal physical exam	Absence of entire posterior arch of C-1 (type E, Fig 8)
4	4, A and B	5 years	M	Neck pain after fall	Some neck tenderness on pressure, normal neurologic exam, pain resolved in a few days	Absence of entire posterior arch of C-1, shown by films and axial CT (type E, Fig 8)
5	5, A and B	8.5 years	F	Pain and stiffness of neck after fall	Neck discomfort on rotation of head, normal neurologic exam, symptoms resolved in 3–4 weeks. Patient again seen at 15 years without symptoms in the interval	Neck films at 8.5 years (Fig 5A) and at 15 years show bilateral defects of the posterior arch of C-1. Axial CT of C-1 at 15 years shown in Fig 5B (type D, Fig 8)
6	6, A and B	12 years	F	Weakness in all four extremities of half-hour duration after fall, followed by tingling in both arms for 2 days	Normal physical exam	Bilateral clefts in posterior arch of C-1 shown by films and axial CT. Normal MR of spine (type C, Fig 8)
7	7	35 years	F	Mother of case 3, "snapping sensations" in neck during certain motions for previous 1 year	No neurologic abnormalities on physical exam	Bony gap in left side of the posterior arch of C-1 best shown in fluoroscopic spots (type B, Fig 8)

of this process) was observed. The anterior arch of the atlas was larger than normal in at least two patients (cases 4 and 6) and in both of them a midline cleft in the anterior arch of the atlas was shown by CT. Lateral films of the cervical spine in flexion and extension were obtained in the first six patients and showed no signs of atlantoaxial instability. In three patients the anomaly was discovered as an incidental asymptomatic finding. Three patients presented with transient neck pain or transient neurologic symptoms after head and neck trauma, and one patient (an adult woman) described neck problems of a chronic nature at the time her affected son was investigated.

Discussion

Developmental anomalies of the posterior arch of C-1 range from simple clefts to absence of the entire arch. A suggested classification based on present material and descriptions in the literature is shown diagrammatically in Figure 8 and is described in the legend. Some of the diagrams included in this figure are derived in part from those published by Von Torklus and Gehle (1). Five types with variants are included in this classification: *A*) failure of posterior midline fusion of the two hemiarches (2–6); *B*) unilateral defect (1, 2, 5–7); *C*) bilateral defects (8–13); *D*) absence of the posterior arch with preservation of the posterior tubercle (1, 7, 9, 14–20); and *E*) absence of the posterior arch including tubercle (7, 21–26). Our material included one case of type B (case 7), two cases of type C (cases 5 and 6) (Figs 5 and 6), and four cases of type E (cases 1 to 4).

Autopsy specimens and surgical explorations in a few reported cases have shown that the bony gaps described in these anomalies were bridged by connective tissue rather than cartilage (4–6, 13, 18, 24, 27). In two adults with absence of the entire posterior arch of C-1 (21, 23), a dense fibrous membrane was found at autopsy extend-

Fig. 1. Case 1. Absence of the entire posterior arch of the atlas (except the roots of the two hemiarches) associated with a cephalad elongation of the spinous process of the axis. The *arrow* points to a faint line across part of this superior bony prominence. The anterior arch of C-1 appears slightly enlarged.

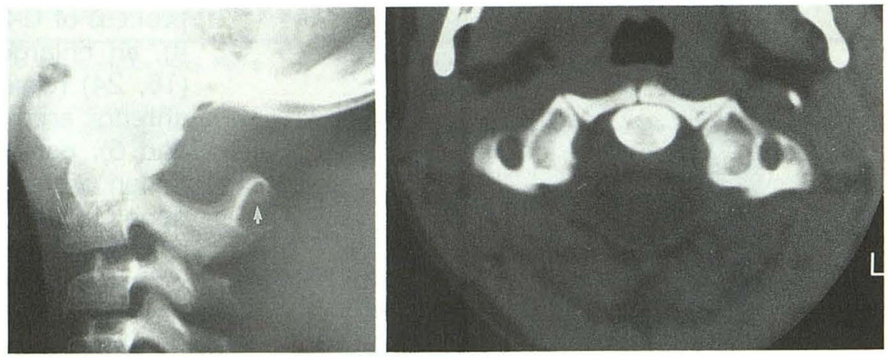
Fig. 2. Case 2. Absence of the entire posterior arch of C-1 (except the roots of the two hemiarches) associated with an upward elongation of the spinous process of C-2. A notch is present in the contour of this superior prominence (*arrows*). The anterior arch of C-1 appears slightly enlarged.

ing from the posterior margin of the foramen magnum to the superior border of the axis; this structure was interpreted as the posterior atlanto-occipital membrane and the posterior atlanto-axial ligament in continuity. These dense fibrous bands and membranes probably account for the good general stability of the cervical spine in these patients. The rest of the cervical spine is usually described as normal, with a few exceptions, including an enlargement of the spinous process of C-2 in some cases (23–27) (cases 1 to 4), an enlargement of the anterior arch of C-1 (16, 24) (cases 4 and 6), a midline cleft in the anterior arch of C-1 (2, 3, 14, 21, 28) (cases 4 and 6), partial occipitalization of the atlas (21), fusion of cervical vertebrae (9, 20, 23), unfused dens (19), and hyperplastic dens (24). We found a faint radiolucent or dense line across the upward prominence of the spinous process of C-2 in type E defect (or a notch in the contour of this process) as shown in cases 1, 2, and 4 of this report.

The frequency of only type A (posterior midline cleft) has been determined; it has been said to occur in 3% to 4% of individuals and to comprise more than 90% of all posterior arch defects of C-1 (2–6). This type is difficult to diagnose from lateral films of the cervical spine, and its diagnosis cannot be made with certainty in the first 5 to 10 years of life when the two hemiarches of C-1 may still be "unfused" normally (24). The other types are generally considered quite uncommon. Based on the observation that five children included in this report were diagnosed in this hospital during a period of 9 years, and that about 800 radiographic examinations that include lateral views of the cervical spine are performed yearly at this hospital, it is estimated that the incidence of these anomalies (other than type A) may be 1 in 1440 lateral neck films (0.69%).

The cause of these anomalies is unknown. Hereditary factors may contribute to their origin in some cases, but how frequently this happens is not known. Motateanu et al (10) reported an affected woman and daughter, and an affected mother and her son are described in the present series (cases 3 and 7) (Fig 7).

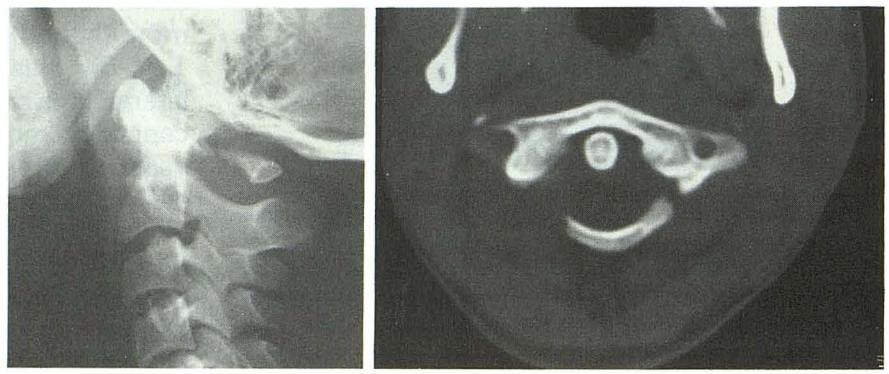
Fig. 3. Case 3. Absence of the entire posterior arch of C-1 associated with an irregular pointed cephalad projection of the spinous process of C-2.



A B

Fig. 4. Case 4. *A,* Lateral view of the cervical spine showing absence of the entire posterior arch of C-1 associated with an upward elongation of the spinous process C-2. A slightly dense line is prsent across this superior prominence (*arrow*). The anterior arch of C-1 is larger than normal.

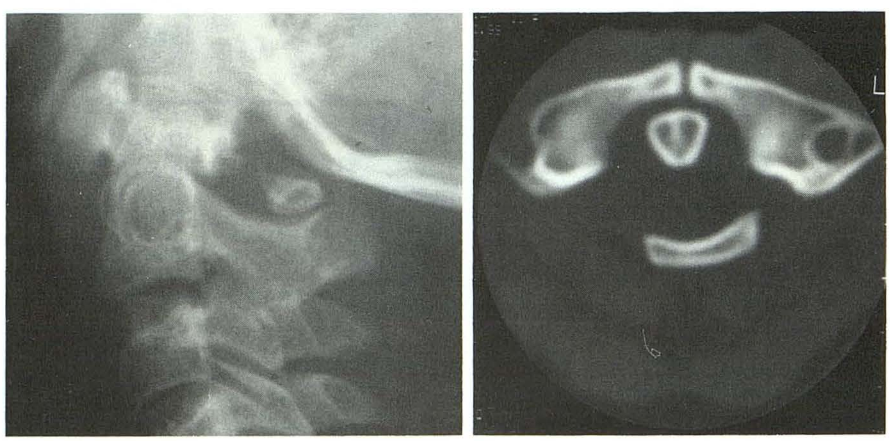
B, Axial CT section of the defective atlas. A midline cleft is present in the anterior arch of C-1, which is still separate from the lateral masses. (Studies performed at Harris Methodist HEB Hospital, Bedford, Tex).



A B

Fig. 5. Case 5. *A,* Lateral view of the cervical spine at 8.5 years showing bilateral clefts in the posterior arch of C-1. The anterior arch of C-1 appears slightly larger than normal.

B, Axial CT sections of the atlas at 15 years clearly shows the extent and location of the defects. The defect on the *right* is larger than the one on the *left.*



A B

Fig. 6. Case 6. *A,* Lateral view of the cervical spine showing bilateral clefts in the posterior arch of C-1. The anterior arch of this vertebra is enlarged.

B, Axial CT section of the atlas clearly shows the extent and location of the defects. The defect on the *right* is larger than the one on the *left.* Note midline cleft of the anterior arch of C-1.

It is commonly believed that the underlying embryologic problem is a local mesenchymal defect leading to a lack of chondrification rather than a primary disturbance of ossification (6, 18, 22, 24). The unattached posterior segment seen in type D is believed to represent a separate ossification for the posterior tubercle. The enlargement of the spinous process of C-2 in patients with absence of the posterior arch of the atlas including posterior tubercle (type E) has been variously attributed to a "compensatory hypertrophy" resulting from the insertion of ligamentous and muscular structures that normally insert on the posterior tubercle of C-1, or to the incorporation of the posterior tubercle of the atlas on the developing spinous process of the axis (15, 23–25). Supporting evidence for the latter interpretation, in some cases, is provided by instances of absence of the posterior arch with an intact tubercle in which this tubercle is very close

Fig. 7. Case 7. Mother of case 3. Fluoroscopic spot films of the cervical spine showing a small gap in the left branch of the posterior arch of C-1 (*arrow*).

to, or is in contact with, the spinous process of C-2 (see Fig 8, type D). Also, in some cases, a transverse radiolucent or dense line or a contour irregularity is present in the superior prominence of C-2, possibly representing the site of fusion of the two elements (see cases 1, 2, and 4 of this report).

Information on the mode of presentation and clinical symptoms is available in the seven patients reported here and in 39 other cases described in the available literature. These patients may be divided into five clinical groups as follows: 1) In 10 previous cases (7, 10, 11, 18, 24, 26, 28) and in cases 1, 2, and 3 of this report, the lesion was discovered as an entirely incidental and asymptomatic finding in films of the cervical spine obtained for unrelated reasons. 2) In eight previous cases (7–9, 12, 17, 18) and in cases 4 and 5 of this report, the diagnosis was made on films of the cervical spine obtained because of neck pain or stiffness or lower back pain of transient nature after trauma to the head or neck. 3) Three patients developed overt neurologic symptoms after head or neck trauma, including case 6 of this series, a case reported by Holsten et al (15), and another reported by Richardson et al (13). Holsten et al (15) described a young man with type D anomaly who developed a quadriplegia that slowly subsided after a long period of head traction. Richardson et al (13) reported a 15-year-old boy with the history of episodes of shock-like paresthesia in all four extremities for 2 years, who developed quadriplegia of 1 hour duration after striking his head. Films of the cervical spine showed absence of the posterior arch of C-1 with intact posterior tubercle. The finding at time of surgery suggested impingement of the cord by the persistent posterior tubercle of C-1, which was surgically removed. 4) Several patients have been described with various neu-

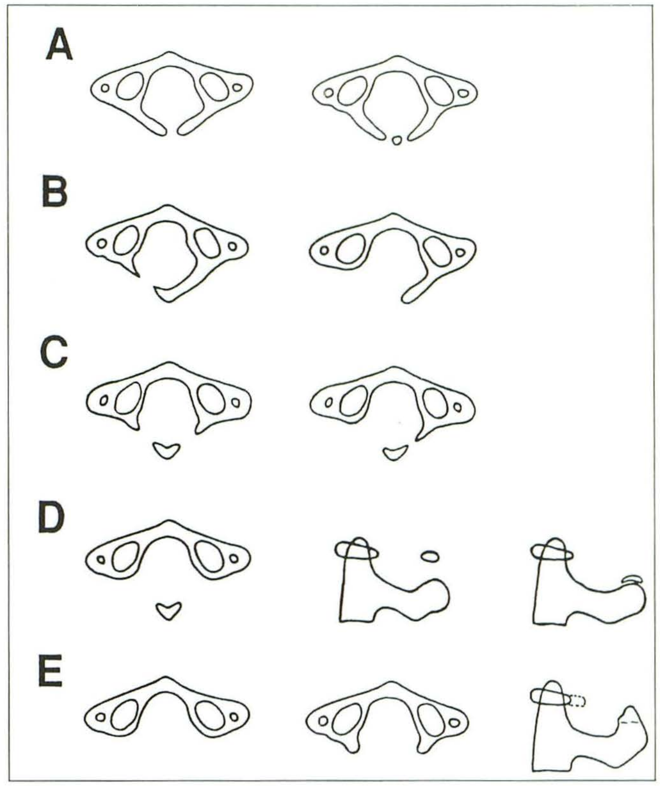


Fig. 8. Congenital anomalies of the posterior arch of the atlas, arranged in 5 types.

A, Failure of posterior midline fusion of the two hemiarches. A bony defect, commonly a fissure or a small gap, is present in the midline posteriorly. Sometimes a small separate ossicle, said to represent a persistent center for the posterior tubercle, is seen within the gap.

B, Unilateral clefts (case 7 of this report). A defect is present in one of the two arms of the posterior arch ranging from a small gap to a complete absence of the half-arch including the posterior tubercle.

C, Bilateral clefts (cases 5 and 6 of this report). A bony defect is present in the lateral aspect of the arch bilaterally with preservation of the most dorsal part of the arch.

D, Absence of the posterior arch with persistent posterior tubercle. In this anomaly, conceivably a more extensive form of bilateral clefts, the lateral parts of the posterior arch are absent except for the posterior tubercle. The defects are frequently asymmetric. The unattached tubercle is usually in the midline and single, but is occasionally bifid with one bony structure on each side of the midline (1), or it may consist of an isolated, unilateral paramedian center (14, 15, 18). The unattached tubercle is usually located well above the spinous process of C-2 but may be low lying and sometimes is in contact with the spinous process of C-2 (17, 18).

E, Absence of the entire arch including posterior tubercle (cases 1 to 4 of this report). The entire posterior arch is missing, but occasionally one or both roots of the arch near the lateral masses are preserved. The spinous process of the axis (C-2) is frequently hypertrophied with a cephalad extension of variable size (23–27). There may be a faint radiolucent or dense line across this prominence (or a notch in its contour).

rologic problems of a chronic nature before the discovery of the C-1 anomaly, including the patient reported by Richardson et al (13), referred to above. Dalinka et al (7) reported a woman with absence of the entire posterior arch of C-1 who was investigated because of "numbness of the head of one year duration." Spadaro et al (20) described seven adults with aplasia of the posterior arch of C-1 with persistent posterior tubercle who presented with a variety of neurologic symptoms or signs. 5) Finally, several adult patients with an anomalous posterior arch of C-1, including case 7 of this report, complained of chronic symptoms referable to the neck or cervical spine. Degrez et al (2) mentioned nine patients, and three other patients were described, one by Fiorani-Gallotta et al (24), the second by Ghislanzoni (29), and the third by Schultze et al (26), who presented with chronic symptoms which were attributed to cervical osteoarthritis.

In conclusion, seven instances of congenital anomalies of the posterior arch of the atlas are described including a mother and her son. An anatomic classification of these anomalies is presented. A review of the clinical symptoms described in affected patients showed that these abnormalities of C-1 are not always asymptomatic and benign as suggested by some authors (7), although it is recognized that often it is not clear whether the symptoms described are in fact attributable to the C-1 defect. As far as we could determine, no fatalities or permanent paralysis have been described in any of these patients, and no particular type of C-1 defect seems to be more prone than others to develop symptoms spontaneously or after trauma. These observations point out the importance of recognizing the possible complications after trauma and other problems that may be encountered in these patients. Normal activity should be encouraged, but contact sports and other strenuous athletic endeavors probably should be avoided in severe cases. Although flexion and extension films of the cervical spine have shown no atlantoaxial instability except in a mild case reported by Schultze et al (26), such films are recommended in all patients with anomalies of the atlas, because the prognosis in patients with atlantoaxial instability is necessarily less certain.

Acknowledgments

We thank Dr. Jerome Novotny (Harris Methodist HEB Hospital, Bedford, Tex) for allowing us to include case 4 in this series.

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Errata

Several references published in the paper "Congenital Defects of the Posterior Arch of the Atlas: A Report of Seven Cases Including an Affected Mother and Son" by Guido Currarino, Nancy Rollins, and Jan T. Diehl, which appeared on pages 249–254 of the February 1994 issue of the *AJNR*, were incorrect because of an error made by the printer. The references were submitted correctly by the author. The references are listed correctly below. The printer regrets the error.

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14. Brocher JE. Konstitutionell bedingte Veränderungen des Wirbelogen. *Fortschr Röntgenstr* 1960;92:363–380
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Figures 2 (*A–D*) and Figures 4 (*A–D*) were inadvertently switched by the printer in the article "MR Imaging of the Middle Cerebral Artery Stenosis and Occlusion: Value of MR Angiography," by Norihiko Fujita, Norio Hirabuki, Keiko Fujii, Tsutomu Hashimoto, Takashi Miura, Tadayuki Sato, and Takahiro Kozuka, published on pages 335–341 of the February 1994 issue. The correct figures, as they should have appeared, are shown below. The printer regrets the error.

A **B** **C**

D

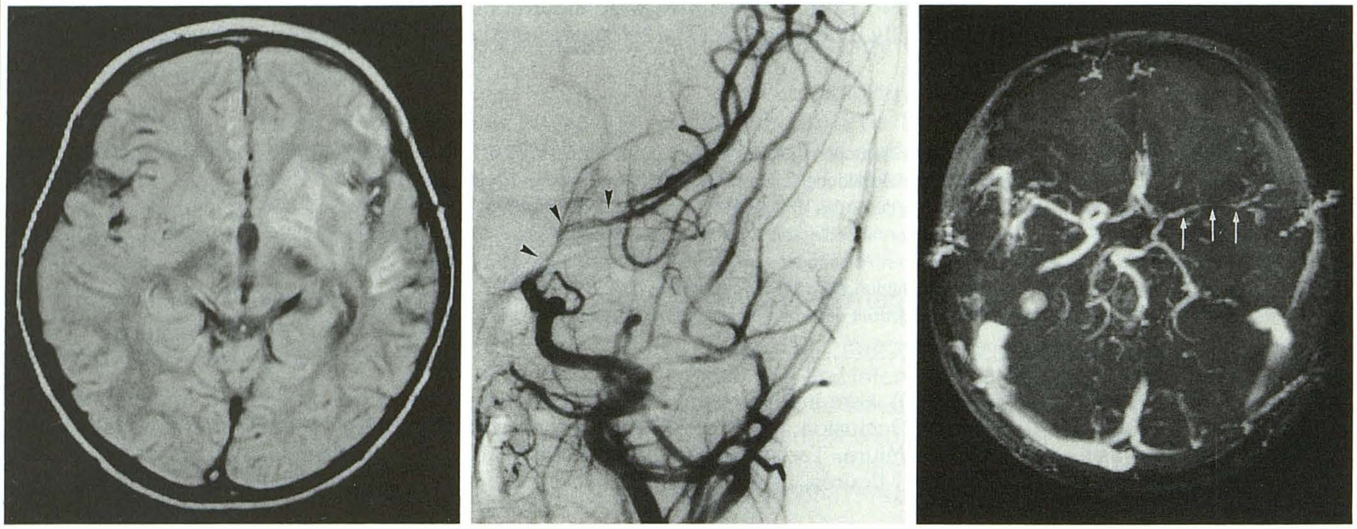
Fig. 2. Case 1: 2-year-old boy with left hemiplegia.

A, T2-weighted spin-echo image (2500/90) shows right basal ganglionic infarction. Normal flow voids are present within the ipsilateral sylvian fissure.

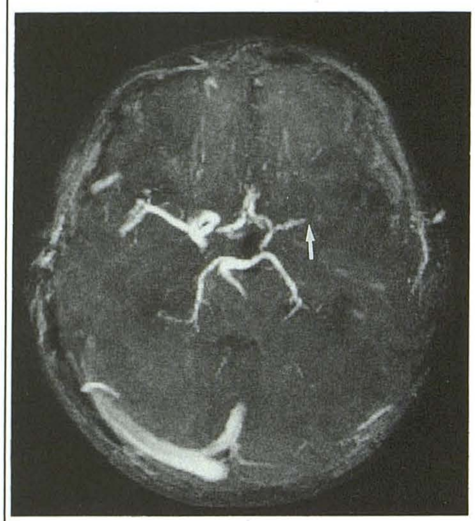
B, Right internal carotid angiogram reveals minimal narrowing of the proximal right MCA (*arrowhead*).

C, Two-dimensional MR angiogram (80/7, 90° flip angle) demonstrates an apparent discontinuity (*arrow*) immediately distal to the right MCA origin and a decreased caliber of the horizontal segment.

D, Three-dimensional MR angiogram (60/7, 35° flip angle) also shows an apparent discontinuity (*arrow*) immediately to the right MCA origin. The apparent discontinuity is less prominent than that on the 2-D angiogram.



A B C



D

Fig. 4. Case 5: 2-year-old boy with right hemiplegia.

A, Proton-density-weighted spin-echo image (2500/15) shows slightly hyperintense signal within the MCA branches in the left sylvian fissure.

B, Left common carotid angiogram reveals severe stenosis from the distal internal carotid artery to the proximal MCA (*arrowheads*).

C, Two-dimensional MR angiogram (80/7, 90° flip angle) shows a decreased caliber of the horizontal segment of the left MCA with a focal discontinuity (*arrows*).

D, Three-dimensional MR angiogram (50/7, 30° flip angle) demonstrates nonvisualization distal to the midportion of the horizontal segment of the left MCA (*arrow*), leading to an incorrect diagnosis of occlusion.