AJNR

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AJNR Am J Neuroradiol 1992, 13 (1) 291-294
http://www.ajnr.org/content/13/1/291

Cranial CT and MR in the Klippel-Trenaunay-Weber Syndrome

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Summary: This report describes the intracranial CT and MR findings in two cases of Klippel-Trenaunay-Weber Syndrome. The findings are 1) markedly enhancing choroid plexuses, 2) severe cerebral atrophy, 3) cerebral calcifications, and 4) angiomatous leptomeningeal enhancement. The findings may resemble those seen in cases of bilateral Sturge-Weber syndrome. The two diseases should be distinguishable by the external stigmata. The authors raise the question of a spectrum of involvement in the angiodyplasias of Klippel-Trenaunay-Weber syndrome and Sturge-Weber syndrome with considerable overlap.

Index terms: Phakomatoses

The Klippel-Trenaunay-Weber syndrome (KTWS) is a rare congenital anomaly characterized by 1) cutaneous port-wine hemangiomata (usually unilateral and involving an extremity, 2) venous varicosities appearing on the affected body part, and 3) osseous and soft-tissue hypertrophy, also of the affected body part (1–3). A variety of associated central nervous system (CNS) abnormalities, overlapping with those of the more common Sturge-Weber syndrome (SWS), have also been identified (4, 5). We describe the strikingly similar and highly unusual intracranial findings in two cases of KTWS, demonstrated by cranial computed tomography (CT) and magnetic resonance (MR) imaging.

Case Report

Case 1

The first patient was a black male infant born at term who presented at birth with glaucoma, asymmetric upper extremities (Fig. 1), and extensive port-wine nevi. Bilateral cutaneous lesions involved the scalp and face (V_1 and V_2 distribution of cranial nerve 5), the upper and lower extremities, and the shoulders, upper back, and chest. Enhanced cranial CT, obtained on the second day of life, demonstrated enlarged, enhancing choroid plexi (Figs. 2A and 2B). Gd-DTPA-enhanced MR scan, performed later the same day, showed marked choroid plexus enhancement but was otherwise unremarkable (Fig. 2C). Four months later, the patient presented with focal seizures involving the left face and arm. A CT scan demonstrated rapid development of gross cerebral atrophy and bilateral cerebral calcifications, as well as intense choroid plexus enhancement and diffuse angiomatous leptomeningeal enhancement (Figs. 2D and 2E).

Case 2

The second patient was a white male infant born at term who presented at birth with glaucoma and cutaneous hemangiomata involving the face, head, and neck bilaterally, ($V_{1,2,3}$ distribution of cranial nerve 5) as well as the left shoulder, arm, upper chest, and buttock, the backs of both lower extremities, and the lateral aspects of both feet. He developed seizures at 4 months of age that were mostly focal involving the face and extremities (especially the right upper extremity). At age 18 months, the patient presented with phenytoin toxicity, developmental delay, and an enlarging left upper extremity. A cranial CT scan revealed marked bilateral cerebral atrophy, extensive bilateral cortical calcifications, and mild angiomatous leptomeningeal enhancement (Figs. 3A and 3B). An MR scan, performed the same day, demonstrated prominent choroid plexi with high signal on T2-weighted images, asymmetric cerebral atrophy, and extensive areas of decreased signal corresponding to parenchymal calcifications on CT scan. (Figs. 3C and 3D).

Discussion

In 1900, Klippel and Trenaunay described the classic triad of symptoms characterizing a condition that would eventually bear their name (1). Weber, shortly thereafter, identified a number of patients who clearly had the KTWS, as well as several who had associated arteriovenous fistulas (2, 3). While some authors feel that "Parkes-

Received July 5, 1991; revision requested August 10; revision received September 3; final acceptance September 4.

Presented at the 28th Annual Meeting of the ASNR, March 19–23, 1990, Los Angeles, CA.

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AJNR 13:291–294, Jan/Feb 1992 0195-6108/92/1301-0291

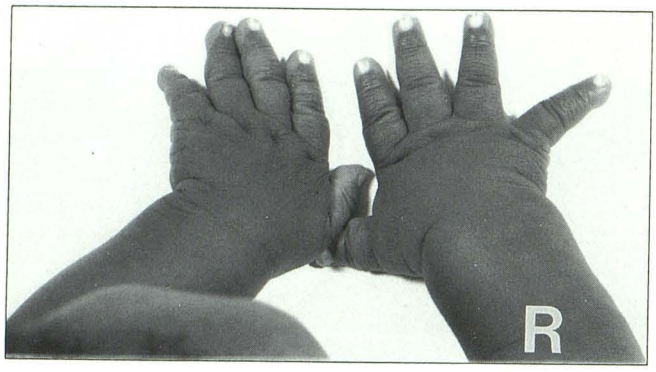
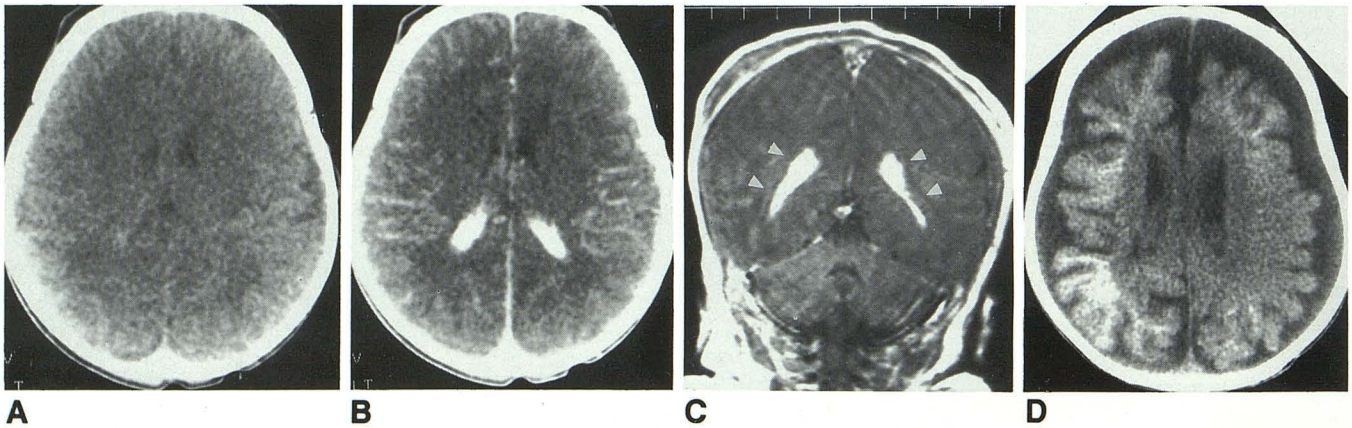


Fig. 1. Patient 1 at age 6 months; note asymmetry of upper extremities with right arm/hand (*R*) slightly larger than left.

Weber syndrome" should be used to designate this latter group of patients, most agree that the Klippel-Trenaunay and Parkes-Weber syndromes are variants of the same condition (4, 6).

The KTWS appears to be a congenital angiodysplasia whose exact pathogenesis and genetic nature is unknown, although prenatal insults (trauma, infection, nutrition, etc) may play some role (4, 5, 7). Because of the wide variety of associated clinical manifestations in this disorder, including neurologic involvement, the KTWS has been classified as one of the phakomatoses (4, 5, 7). In 1973, André stated that the disorder could represent a "forme fruste of the phakomatoses in which the neural element is lacking, but the ectomesodermal components arise from an analogous teratogenic process" (8). We now know that the neural element is very much involved in patients with the KTWS. CNS abnormalities that have been described in this disorder include seizures, mental retardation, migraine headaches, electroencephalogram changes, microcephaly, macrocephaly, hemimegalencephaly, cerebral and spinal arteriovenous malformations, hemangiomas and fistulas, orbitofrontal varices, brain stem angiomas and ischemic infarcts, internal carotid artery aplasia, malformations of the circle of Willis, and optic nerve anomalies (4, 5, 9–19). The inclusion of the KTWS with the other phakomatoses is further supported by the report of multiple cases of this disorder occurring in combination with one of the other neurocutaneous syndromes (4, 5). Over 40 cases of combined KTWS and SWS have been published (4, 5, 19–21). Pietruschka (22) looked at 13 such cases and



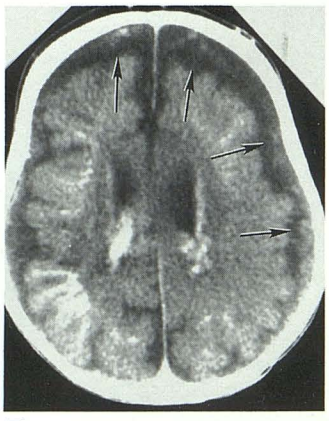


Fig. 2. Patient 1.

A and *B*, Unenhanced and enhanced CT images (GE 9800) on day 2 of life demonstrate prominent, markedly enhancing choroid plexuses.

C, Coronal SE 500/20/2 MR image on same day as CT scan above. Gd-DTPA-enhanced T1-weighted image shows prominent choroid plexuses (*arrowheads*) that markedly enhance following Gd-DTPA administration.

D and *E*, CT scan at age 4 months. Note rapid progression of bilateral cerebral atrophy, extensive calcifications, prominent choroid plexus enhancement, and angiomatous leptomeningeal enhancement (*arrows*).

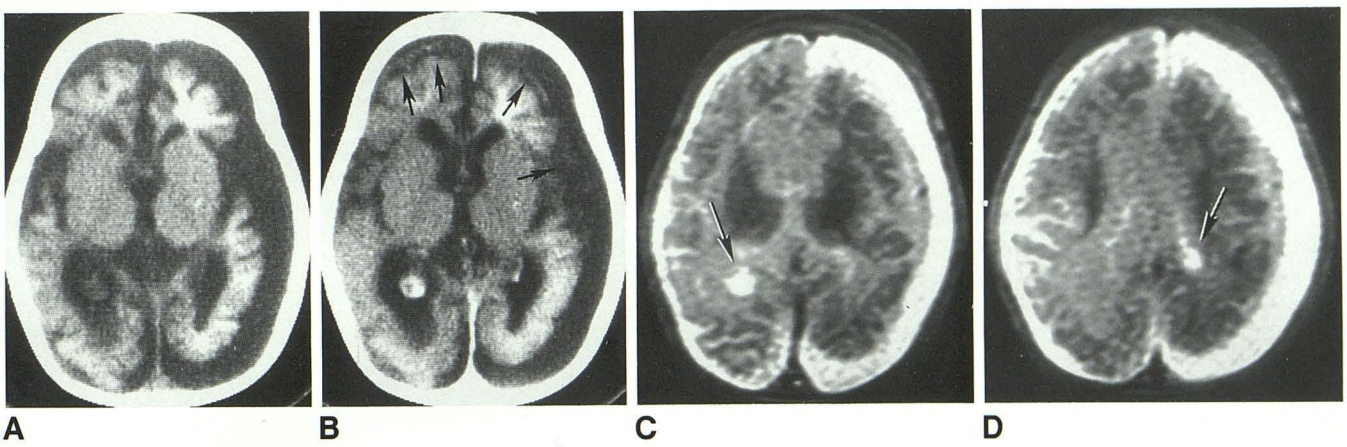


Fig. 3. Patient 2; 18-month-old male with KTWS.

A and *B*, Unenhanced (*A*) and enhanced (*B*) CT scans show severe, bilateral cerebral atrophy, extensive cerebral calcifications, prominently enhancing choroid plexus, and diffuse, angiomatous leptomeningeal enhancement (*arrows*).

C and *D*, Axial T2-weighted (3000/80/1) MR images obtained same day as CT scan demonstrate bilateral asymmetric cerebral atrophy, extensive parenchymal low signal corresponding to cerebral calcifications on CT scan, and increased signal intensity of choroid plexuses (*arrows*).

concluded that KTWS and SWS are probably disembryoplasias that differ only in location of the lesion and in severity of involvement. One of the three patients with SWS reported by Enzmann et al also had clinical features of the KTWS (23). Unlike our cases, the head CT scan in this patient demonstrated unilateral brain involvement.

The intracranial abnormalities demonstrated on cranial CT and MR in our two patients are quite similar in appearance (Figs. 2 and 3) and consist of prominent, markedly enhancing choroid plexuses, severe cerebral atrophy, cerebral calcifications, and angiomatous leptomeningeal enhancement. Although a few cases of KTWS have been studied by cranial CT (15, 16, 18, 19) and at least one patient has undergone both CT and MR imaging (17), the constellation of findings that we report in our two cases have not been previously reported. These intracranial findings are similar to, although more extensive than those typically described in the SWS. SWS, also called encephalotrigeminal angiomatosis, is a congenital neurocutaneous syndrome characterized by facial vascular nevus and multiple intracranial abnormalities (24, 25). These include leptomeningeal angiomatosis (usually ipsilateral to the facial nevus), angiomatosis malformations of the choroid plexus, parenchymal calcifications, cerebral atrophy, buphthalmos, glaucoma, mental retardation, and seizures (24, 25). These intracranial abnormalities have been well demonstrated by cranial CT (23, 26–29) and, more recently, by MR (27–31). Within the past 2 years, the usefulness of DTPA and gradient echo imaging has been demonstrated as well (29–31).

The two cases we describe in this report clinically have the KTWS. We cannot be certain that they do not also represent examples of combined KTWS and SWS. In fact, the intracranial findings in a severe bilateral case of SWS could, conceivably, appear identical to those present in our two cases. Bilateral SWS has been reported (24–26, 28, 30, 31). This fact should not be unsettling, since there is probably a spectrum of involvement in the angiodysplasias of KTWS and SWS with considerable overlap.

References

1. Klippel M, Trenaunay P. Du naevus variqueux ostéo-hypertrophique. *Arch Gen Med (Paris)* 1900;3:641–672
2. Weber PF. Angioma formation in connection with hypertrophy of limbs and hemihypertrophy. *Br J Dermatol* 1907;19:231–235
3. Weber PF. Haemangiectatic hypertrophy of limbs: congenital phlebarteriectasias and so-called congenital varicose veins. *Br J Child Dis* 1918;15:13–17
4. Stickler GB. Klippel-Trenaunay syndrome. In: Gomez MR, ed. *Neurocutaneous disorders: a practical approach.* Boston: Butterworths, 1987:368–375
5. Kramer W. Klippel-Trenaunay syndrome. In: Vinken PJ, Bruyn GW, eds. *Handbook of clinical neurology: the phakomatoses.* New York: Elsevier, 1972;14:390–404
6. Mullins JF, Naylor D, Redetski J. The Klippel-Trenaunay-Weber syndrome. *Arch Dermatol* 1962;86:202–206
7. Koch G. Genetic aspects of the phakomatoses. In: Vinken PJ, Bruyn GW, eds. *Handbook of clinical neurology: the phakomatoses.* New York: Elsevier, 1972;14:488–561
8. André J-M. Syndrome de Klippel-Trenaunay-Weber: dysplasies veineuses congenitales. In: *Les dysplasies vasculaires systematisees.* Paris: Expansion Scientifique Francaise, 1973;179–189

9. Matsubara O, Tanaka M, Takashi I, Okeda R. Hemimegalencephaly with hemihypertrophy (Klippel-Trenaunay-Weber syndrome). *Virchows Arch (A)* 1983;400:155–162
10. Stephan MJ, Hall BD, Smith DW, Cohen MM. Macrocephaly in association with unusual cutaneous angiomatosis. *J Pediatr* 1975; 87:353–359
11. O'Connor PS, Smith JL. Optic nerve variant in the Klippel-Trenaunay-Weber syndrome. *Ann Ophthalmol* 1978;10:131–134
12. Djindjian M, Djindjian R, Hurth M, Rey A, Houdart R. Spinal cord arteriovenous malformations and the Klippel-Trenaunay-Weber syndrome. *Surg Neurol* 1977;8:229–237
13. Kojima Y, Kuwana N, Sato M, Ikeda Y. Klippel-Trenauney-Weber syndrome with spinal arteriovenous malformation. *Neurol Med Chir (Tokyo)* 1989;29:235–240
14. Alberti E. Ischaemic infarct of the brain stem combined with bisymptomatic Klippel-Trenaunay-Weber syndrome and cutis laxa. *J Neurol Neurosurg Psych* 1976;39:581–585
15. Goldstein SJ, Lee C, Young AB, Guidry GJ. Aplasia of the cervical internal carotid artery and malformation of the circle of Willis associated with Klippel-Trenaunay syndrome: case report. *J Neurosurg* 1984;61:786–789
16. Oyesiku NM, Gahm NH, Goldman RL. Cerebral arteriovenous fistula in the Klippel-Trenaunay-Weber syndrome. *Dev Med Child Neurol* 1988;30:245–251
17. Jaksch H, Bewermeyer H, Dreesbach HA, Heiss W-D. Cerebral hemorrhage in arteriovenous malformation associated with Klippel-Trenaunay syndrome. *J Neurol* 1986;233:48–50
18. Anlar B, Yalaz K, Erzen C. Klippel-Trenaunay-Weber syndrome: a case with cerebral and cerebellar hemihypertrophy. *Neuroradiology* 1988;30:360
19. Meyer E. Neurocutaneous syndrome with excessive macrohydrocephalus (Sturge-Weber/Klippel-Trenaunay syndrome). *Neuropaediatrie* 1979;10:67–75
20. Deutsch J, Weissenbacher G, Widholm K, Wolf G, Barsegar B. Combination of the syndrome of Sturge-Weber and the syndrome of Klippel-Trenaunay. *Klin Paediatr* 1976;188:464–471
21. Haberland C, Perou M. Bone involvement in Sturge-Weber-Dimitri syndrome: combined Sturge-Weber-Dimitri and Klippel-Trenaunay-Weber syndrome. *Confin Neurol* 1966;28:413–422
22. Pietruschka G. Zur symptomatik der syndrome nach Sturge-Weber und Klippel-Trenaunay. *Klin Monatsbl Augenheilkd* 1960;137:545–557
23. Enzmann DR, Hayward RW, Norman D, Dunn RP. Cranial computed tomographic scan appearance of Sturge-Weber disease: unusual presentation. *Radiology* 1977;122:721–724
24. Gomez MR, Bebin EM. Sturge-Weber syndrome. In: Gomez MR, ed. *Neurocutaneous disease: a practical approach.* Boston: Butterworths, 1987:356–367
25. Coulam CM, Brown LR, Reese DF. Sturge-Weber syndrome. *Semin Roentgenol* 1976;11:55–60
26. Boltshauser E, Wilson J, Hoare RD. Sturge-Weber syndrome with bilateral intracranial calcification. *J Neurol Neurosurg Psychiatry* 1976;39:429–435
27. Stimac GK, Solomon MA, Newton TH. CT and MR of angiomatous malformations of the choroid plexus in patients with Sturge-Weber disease. *AJNR* 1986;7:623–627
28. Chamberlain MC, Press GA, Hesselink JR. MR imaging and CT in three cases of Sturge-Weber syndrome: prospective comparison. *AJNR* 1989;10:491–496
29. Wasenko JJ, Rosenbloom SA, Duchesneau PM, Lanzieri CF, Weinstein MA. The Sturge-Weber syndrome: comparison of MR and CT characteristics. *AJNR* 1990;11:131–134
30. Elster AD, Chen MYM. MR imaging of Sturge-Weber syndrome: role of gadopentetate dimeglumine and gradient-echo techniques. *AJNR* 1990;11:685–689
31. Lipski S, Brunelle F, Aicardi J, Hirsch JF, Lallemand D. Gd-DOTA-enhanced MR imaging in two cases of Sturge-Weber syndrome. *AJNR* 1990;11:690–692