Klippel Trenaunay syndrome in association with Sturge Weber syndrome about one case

Sturge-Weber Syndrome (SWS) is a sporadic condition of mesodermal phakomatosis characterized by a port-wine vascular nevus of the upper part of the face, leptomeneingeal angiomatosis that involves one of both hemispheres, choroidal vascular lesions associated with glaucoma, seizures, neurological deterioration, and eventual neurodevelopmental delay [1]. Klippel-Trenaunay syndrome (KTS), also a mesodermal phakomatosis, is defined by the triad: port-wine stain, hypertrophy of bone and soft tissue, and varicose veins or venous malformations [2]. Commonly, the diagnosis of KTS can be made when any two of the triad of features is present. It is usually unilateral and almost exclusively involves lower extremities. It is rarely bilateral and involves upper extremities. Overlap between SWS and KTS is recognized**,** and over 40 cases of combined KTS and SWS have been described in the literature [3]. Klippel-Trenaunay-Weber syndrome involves 1 or more extremities and is characterized by a complex of capillary malformations, cutaneous vascular nevi, soft-tissue or bony hypertrophy, and varicose veins or venous malformations [4,5]. The disease is commonly sporadic [6].
This report presented a rare case of the Sturge-Weber syndrome in combination with the Klippel-Trennaunay syndrome in a 4-month-old infant.

Case Report

A 4 month- old male infant with a history of a right congenital glaucoma was presented with left hemi convulsion. His physical examination showed nevus flammeus diffusely involving both sides of his face and extending to his right arm, wrist and leg. His right upper and lower limbs showed a relative soft tissue hypertrophy (Figure 1).

Figure 1 : Bilateral facial nevus flammeus. The right upper limb shows a soft tissue hypertrophy and nevus flammeus.

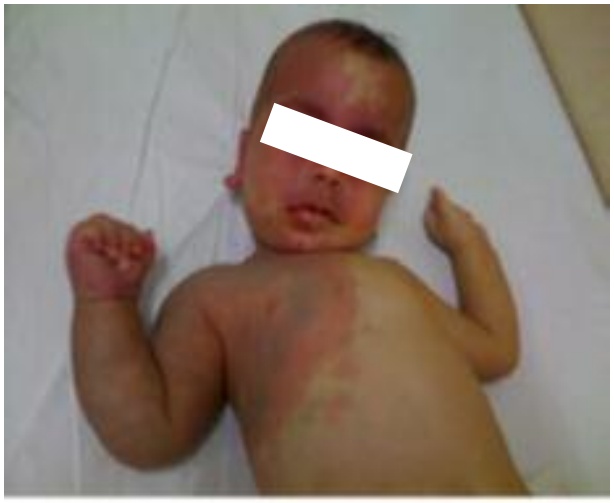
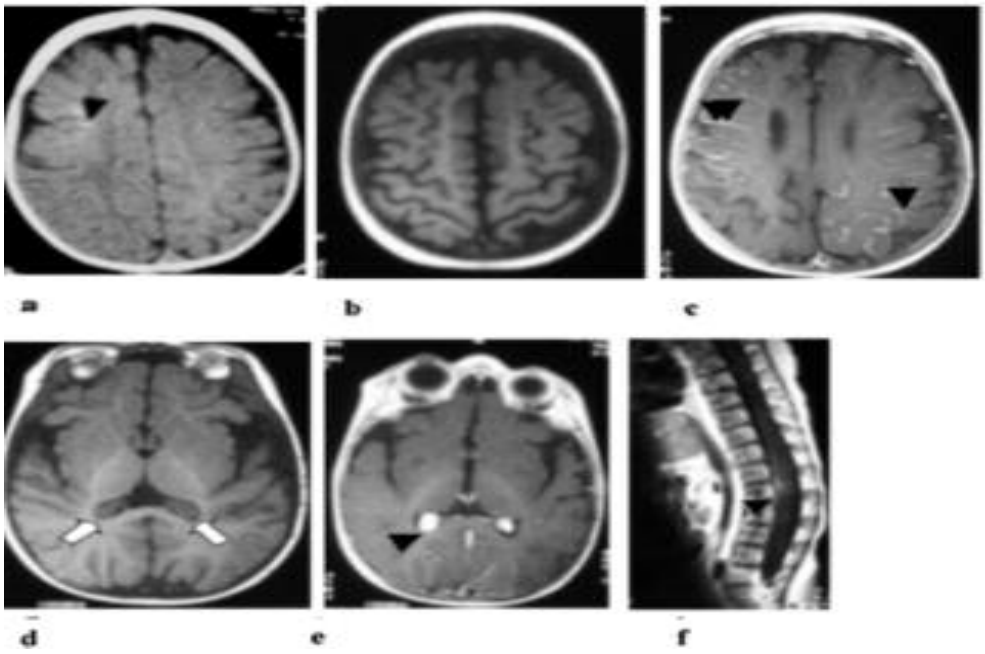


Figure 2 : a: Axial CT image shows calcification of the right frontal cortex (black arrow).
b: Axial SE T1 image shows normal brain parenchyma.
c: Post contrast axial SE T1 image shows the "pial angioma" identified by the presence of bilateral fronto-parietal enhancement that seems to fill the subarachnoid space, covering the surfaces of the gyri and filling the cortical sulci (arrowheads).
d: Axial SE T1 image shows normal brain parenchyma but enlarged choroid plexus (arrows).
e: Post contrast axial SE T1 image shows marked enhancement of the right choroid plexus (arrowhead).
f: Post contrast sagittal SE T1 image shows the pial enhancement at the medullary surface (black arrow).



There was no difference in length of both upper limbs. But the circumference of his right thigh was 26 cm and that of his left thigh was 22 cm. The haematological, biochemical, and urinary laboratory tests and the chest and skull radiography were normal. Abdominal ultrasonography showed no pathological findings of the visceral organs. On plainradiography we detected soft tissue hypertrophy, and in colour Doppler imaging no arterio venous fistula was detected. Cranial computed tomography scan showed calcification of the right frontal cortex. MRI showed normal brain parenchyma but enlarged right choroid plexus (Figure 2).

A diagnosis of Overlap between SWS and KTS was made on the basis (according to association) of port-wine vascular nevus, hypertrophy of bone, leptomeningeal angiomatosis that involves the right hemisphere, and choroidal vascular lesions.

Treatment with anticonvulsant was begun, but hemi seizures recurred several times. His electroencephalography revealed depression ofbackground activity and polyphasic spikes and waves activity in the right side. The patient died at the age of 7 months secondary to a refractory status epilepticus.

Conclusion

Klippel-Trenaunay-Weber syndrome is a rare vascular syndrome sometimes associated with neurologic pathologies. In our case evolution was rapidly fatal secondary to a refractory status epilepticus.

Conflict of interest: No conflict of interests.

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Une tumeur inhabituelle du canal cholédoque

Le carcinosarcome est une tumeur maligne rare, associant un contingent épithélial carcinomateux et un contingent mésenchymateux malin pouvant renfermer un tissu hétérologue [1]. Il survient au niveau de plusieurs organes tels que l'utérus, la vessie, le poumon, l'œsophage. La localisation au niveau de l'arbre biliaire est rare. Elle a été décrite pour la première fois en 1907 par *Landsteiner* [2]. Environ 36 cas ont été rapportés au niveau de la vésicule biliaire et seulement 6 cas au niveau des voies biliaires extra-hépatiques [3].

Le but de notre travail est d'étudier les caractéristiques anatomo-cliniques et évolutives du carcinosarcome du canal cholédoque.

Observation

Mr AH, âgé de 71 ans, consultait pour des douleurs de l'hypochondre gauche évoluant depuis un mois avec apparition récente d'un ictère cutanéo-muqueux. Il présentait par ailleurs des urines foncées, des selles décolorées et une altération de l'état général. L'examen biologique mettait en évidence des signes de cholestase. L'échographie abdominale montrait un pancréas hétérogène au niveau de sa portion céphalique avec une dilatation du bas cholédoque, des voies biliaires intrahépatiques et extrahépatiques. La tomodensitométrie abdominale objectivait une dilatation du cholédoque jusqu'à sa portion rétropancréatique et une dilatation des voies biliaires intrahépatiques. La tête du pancréas était globuleuse dans sa partie basse avec présence d'une plage hypodense de 15 mm. A l'IRM, il existait une dilatation cholédocienne marquée et débutante au niveau du canal de Wirsung en amont d'un épaississement pariétal cholédocien et d'une masse centrée sur le bas cholédoque. Cette masse et cet épaississement étaient en hyposignal T2 et de rehaussement plus marqué en temps tardif. Ces aspects évoquaient un cholangiocarcinome du bas cholédoque. Une duodénopancréatectomie céphalique était réalisée. A l'examen macroscopique, le cholédoque était le siège d'une formation tumorale intraluminale, végétante et sténosante, infiltrant le pancréas et la paroi duodénale. Histologiquement, cette masse correspondait à une prolifération tumorale biphasique, à double composante [Figure 1].

Figure 1 : HEx10) : Prolifération tumorale biphasique associant un contingent épithélial et mésenchymateux