Ischaemic infarct of the brain stem combined with bisymptomatic Klippel-Trénaunay-Weber syndrome and cutis laxa

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SYNOPSIS The combination of Klippel-Trénaunay-Weber syndrome (KTWS) with intracranial vascular malformations is documented by angiography in a 25 year old man with the classical features of KTWS: systematized naevi, hypertrophy of the right side, and varicosity. In addition, the syndrome was associated with ‘cutis laxa’, a coincidence of some rarity. The patient suffered from an ischaemic infarct of the brain stem with several neurological deficits.

In 1900, Klippel and Trénaunay described a triad of symptoms consisting of a naevus telangiectaticus in metameric distribution affecting the entire lower extremity, unilateral varicectasia, and hypertrophy of the affected side (unilateral macrosomia).

In combination with the syndrome described above, Parkes Weber in 1918 observed arterio-venous deformities occurring in addition to varicectatic changes. Therefore, this clinical entity is now generally called the Klippel-Trénaunay-Parkes-Weber syndrome (KTPWS). Other synonyms are: naevus osteohypertrophicus, macrosomia, angiomatosis osteohypertrophica, haemangiectasia hypertrophiicans, telangiectasia and angioelephantiasis, angio-plastic macrosomia, angio-oste-hypertrophic syndrome, phlébectasies ou phlébartérietiases étendues congénitales complexes (Kramer, 1972).

In 1953, Fegeler et al. differentiated between the three cardinal symptoms, systematized haemangiomas, hypertrophy, and varicosity, and the secondary symptoms which include changes in the peripheral nervous system such as perception and vegetative disorders. Epileptic fits, changes in the electroencephalogram, or oligophrenia have been described in this context only very infrequently (André, 1973). This paper describes a rare combination of the Klippel-Trénaunay-Parkes-Weber syndrome with ‘cutis laxa’ and intracranial vascular deformities leading to the clinical picture of an ischaemic infarct of the brain stem. The author believes that this is the first description of a clinical course of this kind.

CASE HISTORY

The patient K.B., aged 25 years, was admitted on 3 March 1974 and discharged home on 20 April 1974.

There was no history of previous serious illness. The patient had performed hard physical work for eight years. Headaches, limited to the occipital region for three days, started eight days before admission. After four days without complaint, the patient woke up with splitting headache; vomiting occurred some hours later. This had been preceded by particularly strenuous work the day before. One day later, the patient complained of dryness in the mouth and continuing occipital headache, later followed by a sensation of numbness in the legs spreading to the upper extremities. The patient was admitted for treatment.

NEUROLOGICAL FINDINGS ON ADMISSION The patient had to be supported on both sides when walking. There was an ataxic gait, predomin-
E. Alberti

stantly of the right leg, sensorimotor proportional hemiparesis of the right side and impaired co-
ordination of the right. There was anisocoria (left pupil more dilated than right pupil); paralysis of
the right 6th cranial nerve, dysadaptation and course and jerking end position nystagmus in both eyes. There was also nuclear paresis of the left facial nerve, strangled voice, paralysis of
deglutition and profuse salivation. Slight drowsi-
ness was noted. During physical examination of
the patient it was found that dermatomes C4–T4
and S1/S2 on the right side of the body showed
several palm-sized naevi telangiectatici, whereas
on the left side, at T6 and T11, only two naevi tel-
angiectatici were located, each one being the size
of a shilling. The right side of the body was hyper-
trophied, the difference in circumference of the
extremities varying from 1 to 2 cm. This was par-
ticularly obvious in the right hallux. Moderate
varicosis was present in the right foot and small
herniae in the fatty tissue of the heels. The skin,
predominantly on the right-hand side of the body,
was found to be excessively elastic and the joints
of the same side were slightly hyperextensible.

ADDITIONAL FINDINGS A chest radiograph was
normal. A film of the pelvic region showed
phleboliths located in the minor pelvis. The lowest
lumbar vertebra was partially united with the os
sacrum on the right side. Radiography of both
hands showed asymmetry due to macrosomia of
the right hand. An EEG was normal. Lumbar
puncture yielded CSF containing 11–3 cells per
mm³, albumin level was normal. Laboratory find-
ings were within normal limits.

ARTERIOGRAPHY Retrograde arteriography of the
left brachial artery revealed subnormal diameter
of the left vertebral artery which supplied exclu-
sively the left inferior cerebellar artery.

A percutaneous arteriogram of the left carotid
artery showed a wide-arched course of the
thalamostriate vein.

A retrograde arteriogram of the right brachial
artery (Fig. 1) showed aneurysmal dilatation of
the terminal part of the basilar artery, and ectasis
at the junction of the right vertebral artery (which
was of wide diameter) and the basilar artery.
Vascular abundance of the superior cerebellar
artery was noticeable.

FIG. 1. Retrograde arteriogram of the right brachial artery. (a) AP view. (b) Lateral view.
Ischaemic infarct of the brain stem

PROGRESS Some days after the attack the patient
was able to swallow again. His dexterity, which
at first had been most adversely affected on
the right side of the body, improved as well. After
one month there was still a noticeably atactic
gait, an obstinate nystagmus, and recurrent at-
tacks of headache. On discharge from hospital
the patient still showed ataxia on the right,
vertical spontaneous nystagmus, and nystagmus
on deviation of the eyes. Pupillary reaction on
convergence was sluggish. At the follow-up ex-
amination five months later the patient still com-
plained of clumsiness on the right side, and of
blurred vision when turning his head or when
looking sideways. His speech continued to return
to normal. One year after the attack there was
still evidence of spontaneous nystagmus with a
rotatory component, and of paralysis of the right
6th nerve, with the soft palate sagging on the
left, comparatively feeble gag reflex on the left,
development of the protruded tongue to the left,
sluggish pupillary reaction, and some remaining
signs of ataxia on the right side. Eighteen months
after the infarct the patient complains of sporadic
headaches limited to the occipital region and of
'blurred pictures' when walking. There is still
evidence of spontaneous nystagmus with a rota-
tory and a vertical component and of nystagmus
on deviation of the eyes caused by a slight
residual paralysis of the right 6th nerve. The
right side of the body shows increased tendon
reflexes, whereas the abdominal reflex is absent.
Co-ordination of the right side of the body is
still clearly impaired.

DISCUSSION

In 1954, Burke et al., in their discussion of
disseminated haemangiomatosis, stated that
the existence of such vascular deformities
should give rise to the consideration that they
might occur together with intracranial angio-
matous malformations. Cases of KTPWS coinci-
ding with such disturbances of the vascular
system supplying blood to the CNS have
relatively rarely been described in the litera-
ture. Den Hartog Jager (1949) reported on a
venous angioma in the spinal region, and
Krayenbühl et al. (1970) reported an atrio-
ventricular fistula located in the thoracic part
of the spine which led to compression of the
spinal cord. Brüning (1956) discovered cases
of angiomatosis in the diencephalon, in the
medulla oblongata, and in the spinal cord;
Thierbach and Hübner (1964) described angio-
matomatosis and regressive changes mainly in the
lower brain stem and in the cerebellum of a
20 year old patient. Haberland and Perou
(1966) reported on venous angiomatosis of the
skull vault. In 1968, Avasthy and Roy de-
scribed cases of arteriovenous deformities in the
cerebrum.

A considerable number of cases of KTPWS
are combined with phacomatoses, the latter
sometimes necessarily involving vascular
deformations in the area of the CNS: Sturge-
Weber syndrome (Laxenaire, 1961); Von
Hippel-Lindau syndrome (Den Hartog Jager,
1949); tuberous sclerosis (Laxenaire, 1961;
Troost et al., 1975); multiple neurofibromatosis
or Recklinghausen's disease (Laxenaire, 1961).
Schnyder et al. (1956) compiled the interrela-
tions between some of the syndromes (Fig. 2).
The case of KTPWS described by the author
in this paper is to be deemed bisymptomatic
because the varicosity component of the triad
(cutaneous symptoms, naevi, osteohyper-
trophy, and vascular symptoms) is rather
imperfectly developed. Arterial thromoses show
a marked inclination to develop in vascular
anomalies located in the cerebrobasilar circu-
latory system. This may evolve into a
secondary embolism and a consecutive
ischaemic infarct within this vascular region.
The protracted clinical course of the case de-
scribed here supports the assumption that this
infarct was caused by a thrombotic process.
Clinical examination yielded no evidence of
heart disease or of inflammatory disease. In
view of the age of the patient, however, the
thrombocyte aggregation test must be con-
sidered pathological. It is probable that the
brain stem infarct caused an extensive lesion
affecting the lower portion of the pons and the
medulla oblongata. This case also presents
an unusual feature by combining KTPWS with
intracranial vascular deformities and cutis
laxa. The only other case of a similarly diffuse
dystrophy of the elastic tissue was reported
by Thiers et al. (1964) in a 6 year old child. In
this case disturbance of tissue metabolism was
evidenced by increased diuresis of acid muco-
proteins.
Cutis laxa may be either of a general nature or segmentally restricted. A mesenchymal dysplasia of this kind is presented in its fully developed state by the Ehlers-Danlos syndrome, combining fragility and hyperelasticity of the skin (note: the skin was not fragile in our case), hyper-extension of the joints, and changes in the eyes and blood vessels. This syndrome may also combine with KTPWS (Lynch et al., 1965; Servelle, 1962), leading to the conclusion that any appreciable deficiency in the tissue metabolism may constitute an additional factor causing the development of certain symptoms such as vascular deformations and changes in the cutaneous structure. There are many primary defects which may lead to the above symptoms. The Ehlers-Danlos syndrome, for instance, is caused by a disturbance in the formation of the collagenous dystrophy, and osteogenesis imperfecta by an impaired maturation of collagen. The Pfaundler-Hurler syndrome involves a qualitatively and quantitatively abnormal formation of mucopolysaccharides or structural polysaccharides, and Marfan's syndrome consists of a complex deficiency phenomenon in the connective tissue (McKusick, 1959; Deckers, 1965; Laitinen et al., 1966; Alberti, 1970).

We may, therefore, conclude that hereditary factors disturbing the tissue metabolism may be of some relevance in symptom combinations similar to the case described in this paper. On the other hand, the presence of angiomatous and structural changes of the skin, especially in coincidence with neurological symptoms, may indicate that the structure of intracranial and spinal arteries and veins has been affected. The coincidence of KTPWS, cutis laxa, and intracranial vascular deformities is rare.

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Ischaemic infarct of the brain stem


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E Alberti

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