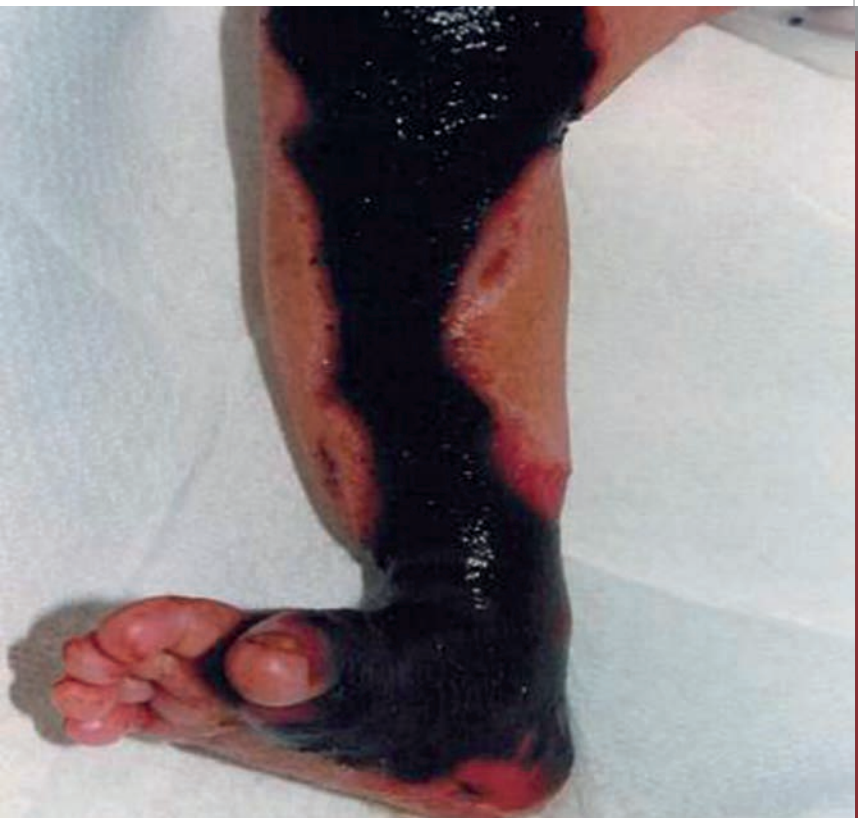
SWISS SOCIETY OF NEONATOLOGY

Bart's syndrome with severe newborn encephalopathy: a delayed diagnosis



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After an uneventful pregnancy, a 20-year-old G1/P1 delivered a female infant at 41 0/7 weeks by emergency Cesarean section for failure to progress and meconium stained liquor. The infant weighted 3440 g and had Apgar scores of 4 and 9 recorded at one and five minutes, respectively. She was transferred to the nursery for what was thought to be gangrene of an extensive area of her right lower leg apparent at birth (Figure).

Generalized tonic-clonic seizures with apnoea and cyanosis began six hours after birth and recurred frequently for the next few hours. Despite commencing phenobarbitone and phenytoin, seizures persisted and she required mechanical ventilation and sedation.

The infant was transferred to the Children's Hospital at Westmead at the age of 15 hours. An EEG at 17 hours of age showed runs of spike and spike-wave discharges associated with localised 1-2 Hz slowing consistent with status epilepticus. Clonazepam was started. A cerebral ultrasound demonstrated cerebral edema and a left-sided infarction. The CT scan showed edema and diffuse low densitiy changes through the cortex with sparing of the basal ganglia and posterior fossa. These findings were consistent with a perinatal hypoxic event. An MRI at 10 days of age showed diffuse hyperintensity throughout the cerebral hemispheres consistent with widespread cerebral infarction. Cultures of cerebrospinal fluid and blood were negative. Metabolic and coagulation studies were all normal.

Although initially she was thought to have a thrombotic disorder which had resulted in a cerebral infarct and gangrene of her lower limbs, a diagnosis of epidermolysis bullosa was later suspected when she developed several blisters on her left hand, lips and scalp where the EEG electrodes had been placed. A dermatological opinion was sought and with the deformity of nails and the lesion on her right leg the diagnosis of Bart's syndrome was made. Skin biopsies were submitted for electron microscopy and immunomapping and were consistent with the diagnosis of the dystrophic from of epidermolysis bullosa (EB). The results were consistent with the dominant as well as recessive forms of EB. No other family member is affected with any form of EB or localized absence of skin.

The infant was extubated on day three and all medications were ceased. Thought she had no further clinical seizures her neurological behavior remained abnormal. The area of her skin loss on her leg healed over the next few months. After 12 months, mucosal and skin blistering became minimal with normal appearance of her nails. She has since developed spastic quadriplegia with microcephaly and epilepsy.

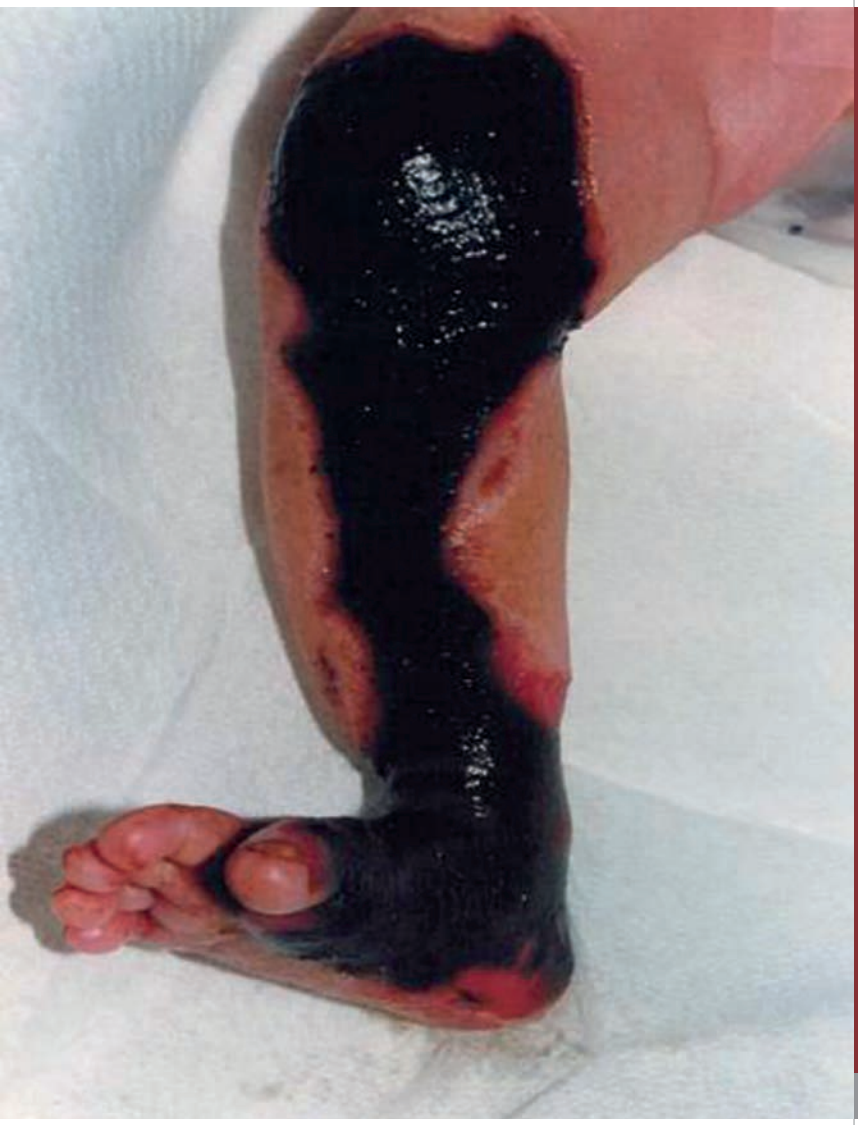

Fig. 1

Gangrenous lesion of the right lower leg.

DISCUSSION

We present this case as the infant was referred with what was thought to be gangrene of the leg. This presumptive diagnosis was initially supported by the abnormal neurological findings.

In 1966, Bart described a syndrome with congenital absent skin (CAS) of the lower extremities, epidermolysis bullosa (EB) and deformity of nails (1). Various types of EB are associated with Bart's syndrome and CAS is reported to be associated with pyloric atresia (3,4). Our patient had the dystrophic type of EB. Most cases of Bart's syndrome are reported with the dystrophic form but rarely also with the junctional and simplex form (5). We could not find a report of EB associated with newborn encephalopathy.

Neonatal encephalopathy is a clinically defined syndrome of disturbed function in earliest days of life in the term infant, manifested by difficulty with initiating and maintaining respiration, depression of tone and reflexes, subnormal level of consciousness, and often by seizures (2,6). It is a condition with diverse associations in the preconceptional, antepartum and intrapartum periods (2,6,7). It is likely in this case that the encephalopathy was not directly associated with EB although newborn encephalopathy is known to be associated with birth defects (2,6,7). These defects, which occur early in gestation, may be markers of other factors in early pregnancy which may also cause encephalopathy. It is also possible that the presence of

a birth defect may render the fetal brain susceptible to other damaging factors. Encephalopathy with birth defects is known to have a worse prognosis with a higher likelihood of the infant developing cerebral palsy (8) as in this case.

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