The significance of elevated CSF lactate

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The final diagnosis of 158 patients who had a cerebrospinal fluid (CSF) lactate concentration greater than 2 mmol/l was ascertained. The conditions included seizures, inflammatory changes, and proven metabolic disorders. For the diagnosis of congenital lactic acidoses, CSF lactate should ideally be measured in a seizure free patient after any acute illness.

As blood lactate concentrations are influenced by several factors, cerebrospinal fluid (CSF) lactate concentrations are widely used in the diagnosis of congenital lactic acidoses. However, CSF lactate may also be raised in association with many other illnesses including seizures, meningitis, encephalitis, and cerebral ischaemia. In order to determine the value of measuring CSF lactate, we have reviewed the records of 158 children who attended a tertiary referral hospital with concentrations greater than or equal to 2 mmol/l.

METHODS

In this study, CSF lactate measurements were reviewed for the 10 year period of 1992–2002. The medical records of those patients with values equal to or greater than 2 mmol/l were retrieved. For patients in whom more than one CSF lactate measurement was taken, the highest value was used. Additional details were also recorded, including the patient’s condition at the time of the lumbar puncture, the presence of seizures, haemodynamic status, other metabolic investigations, and the final diagnosis. The project was registered with the Research and Development office and the patient details anonymised.

RESULTS

During the study period, CSF lactate was measured on 2268 occasions. Of these measurements, 414 (18%) values from 267 patients were greater than or equal to 2 mmol/l. We were able to review the records of 158 (59%) patients. CSF lactate concentrations for patients grouped according to diagnosis are shown in fig 1.

Metabolic disorders

Forty five patients (28%) had a metabolic disorder (37 respiratory chain disorder, five pyruvate dehydrogenase deficiency, two Krabbe’s leukodystrophy, and one biotinidase deficiency).

Seizures

In 24 patients (15%) the raised CSF lactate value was attributed solely to seizure activity occurring within 3 days of the lumbar puncture.

Intra-cranial inflammation

Twenty nine patients (18%) had encephalitis, meningitis, or cerebral inflammation. Nine (6%) had cerebral ischaemia.

Malignancy

Six (4%) of the patients had malignancy. In five patients the malignancy was cerebral or meningeal in origin. The other patient had an adrenal neuroblastoma with cerebral metastases.

Other causes

Several patients had more than one cause for raised CSF lactate concentrations. Of the 36 patients with high CSF lactate values within 3 days of active seizures, 12 patients had other diagnoses which are likely to have contributed to the result as follows: three had a respiratory chain defect, six an inflammatory process (two meningitis, two encephalitis, one SLE, and one post-streptococcal acute disseminated encephalomyelitis), one cerebral ischaemia, and two other metabolic disorders (one Gaucher’s disease and one Menke’s syndrome). One patient with a posterior fossa medulloblastoma developed postoperative meningitis and another with tuberculous meningitis developed a middle cerebral artery infarct with both of these factors contributing to the raised CSF lactate values of 5.6 and 6.4 mmol/l, respectively.

In the remaining 45 (28%) patients, no obvious cause was found for the increased CSF lactate measurement. One 19 day old patient was acutely unwell with multi-organ failure; an infection screen, urine organic acids, plasma amino acids, muscle histology, and respiratory chain enzymes failed to identify the cause of a raised CSF lactate value of 21.9 mmol/l. Twenty nine of the 45 patients had normal metabolic investigations including muscle histology and respiratory chain enzyme assay. Eight of these had an illness consistent with congenital lactic acidosis and, although the investigations failed to confirm this, a metabolic disorder was likely.
Muscle fibre type grouping in high resolution ultrasound images in paediatrics

A 5 year old boy with motor retardation presented with proximal weakness maximal in the lower limbs and generalised muscular atrophy. The deep tendon reflexes were absent. Tremor or fasciculations were not seen. The muscles of the boy were examined by a high resolution ultrasound system and real time compound imaging. The deep muscles consisted of atrophic fibres (bright areas in fig 1) with groups of hypertrophic fibres (black areas in fig 2). The analysis of the survival motor neurone (SMN) 1 gene resulted in a homozygous deletion of exon 7 and 8 confirming the diagnosis of spinal muscular atrophy.

Muscle fibres of the denervated motor unit atrophy. Reinnervation by collateral sprouting results in grouping of hypertrophic fibres with the same histochemical type, commonly called "type grouping".

Using this new ultrasound technique, ultrasound morphological findings can be compared to histological findings.

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REFERENCE

Muscle fibre type grouping in high resolution ultrasound

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