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.

Abbreviation: CSF, cerebrospinal fluid
still likely. Sixteen patients had metabolic investigations that did not include a muscle biopsy. In three of these patients, definitive diagnoses were made of illnesses not known to cause raised CSF lactate values (vanishing white matter disease, juvenile idiopathic arthritis with papillitis, and hereditary sensory motor neuropathy type VI). However, the values in these patients were only marginally raised (2–2.07).

**DISCUSSION**

Great Ormond Street Hospital is a tertiary referral children’s hospital and the specialist nature of the hospital will have influenced the diagnoses and frequency of raised CSF lactate values. Furthermore, only 60% of the notes were reviewed. Nevertheless, the results show that in the patients investigated a CSF lactate value greater than 2 mmol/l is not uncommon as 18% of all values were above this concentration.

Our study confirms that CSF lactate concentrations can be raised in association with seizures, meningitis or encephalitis, cerebral ischaemia, malignancy, and other metabolic disorders and is not specific for congenital lactic acidosis.

Of patients who were acutely unwell at the time of the lumbar puncture, only 9% were found to have congenital lactic acidosis, compared to 34% of patients who were in the chronic phase of their illness. CSF lactate concentration is less specific for congenital lactic acidoses in patients who are acutely unwell.

Measurement of blood lactate is often used as a first line assessment of those with presumed congenital lactic acidosis. However, it is recognised that blood lactate levels are notoriously variable, being affected by many factors including muscle movement. Furthermore, the CSF lactate level may be increased when the plasma or blood lactate concentration is normal in children with electron transport chain defects. If CSF lactate measurements are to be used to identify patients requiring further investigation for congenital lactic acidoses, the test should ideally be performed in a seizure free patient during the chronic phase of the illness, but even so it should be remembered that it may be normal in those with a proven mitochondrial disorder.

**REFERENCES**


**IMAGES IN PAEDIATRICS**

Muscle fibre type grouping in high resolution ultrasound

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 neuron (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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Competing interests: none declared