Paediatric Research Society

19th Meeting, Royal Hospital for Sick Children, Glasgow
1 and 2 October 1971

Clicks, honks, and whoops. D. Pickering (*Radcliffe Infirmary, Oxford*). Three cases are described which were originally diagnosed as having ventricular septal defect, pulmonary stenosis, and innocent praecordial honk. These have been reinvestigated using pixie accelerometer, phonocardiographic techniques (Bew *et al.*, 1971). All three have been shown to have clicks which move with posture, Valsalvas, and other manoeuvres altering the left ventricular end-diastolic volume. These features enable a diagnosis to be made of prolapsing posterior cusp of the mitral valve.

REFERENCE

- Bew, F. E., Pickering, D., Sleight, P., and Stott, F. D. (1971). 'Pixie' cardiography. Accelerometer applications to phonocardiography and displacement cardiography in childhood. *British Heart Journal*, **33**, 702.

Measurement of total respiratory resistance using oscillation technique. Jeremy Cogswell (*introduced by David Hull*) (*The Hospital for Sick Children, Great Ormond Street, London*). A method for measuring total respiratory resistance by a forced oscillation technique has been described in adults. The technique requires minimal co-operation and causes no discomfort, and therefore can be used in children of all ages.

The details of the technique with modification required for children were described. Values for total respiratory resistance in 170 healthy schoolchildren aged 6-12 years were similar to values of airways resistance using a total body plethysmograph. Reproducible results were obtained in children between the ages of 2 and 5 years.

REFERENCE

- Fisher, A. B., DuBois, A. B., and Hyde, R. W. (1968). Evaluation of the forced oscillation technique for the determination of resistance to breathing. *Journal of Clinical Investigation*, **47**, 2045.

Paediatric electrodiagnosis. Allie Moosa (*Department of Child Health, University of Sheffield*). The measurement of the conduction velocity of the peripheral nerve and the application of electromyography (EMG) has proved invaluable in the diagnosis of peripheral neuropathy and in the differentiation of the various forms of muscle weakness in infancy and childhood.

Slow nerve conduction velocities have been found in patients with some of the leucodystrophies, e.g. Krabbe's and metachromatic leucodystrophies, and this has

provided a useful screening test for the leucodystrophies in patients with progressive degenerative diseases of the nervous system. Hypothyroid infants have also been found to have slow conduction velocities probably due to retarded myelination. The diagnosis of a peripheral neuropathy due to other causes, e.g. peroneal muscular atrophy, infections, polyneuritis, can also be confirmed by this test.

Needle EMG provides information about the state of the lower motor neurone. Characteristic changes are seen in lesions affecting the anterior horn cell, peripheral nerve, and muscle fibres. The EMG is therefore useful in the differential diagnosis of the floppy infant syndrome and of the child with proximal muscle weakness. It is particularly useful in the diagnosis of dystrophia myotonica and in assessing the prognosis of peripheral nerve lesions, e.g. facial palsy.

The quantitation of the EMG signal has led to its use in detection of carriers of Duchenne dystrophy.

Diagnostic advances in neuromuscular disorders.

Victor Dubowitz (*Department of Child Health, University of Sheffield*). The application of modern histochemical techniques to the assessment of muscle biopsies has led to the recognition of a number of new conditions, such as central core disease and nemaline myopathy, which may masquerade clinically as a dystrophy, and readily be missed with routine histological stains.

'All that waddles is not dystrophy'.

The identification of histochemical fibre types within muscle has also made possible the recognition of selective involvement of fibre types in certain disease situations. Examples include type I atrophy in dystrophia myotonica, type II atrophy in some cases of polymyositis, and fibre type grouping as may be seen in neurogenic atrophy, even in the absence of change in fibre size, thus making possible the diagnosis of these conditions where routine stains show no obvious abnormality.

Cases presented to illustrate these conditions include two floppy infants and their apparently normal related mothers (aunt and niece) with dystrophia myotonica; a mother with central core disease affecting 99% of the muscle fibres in her biopsy and her minimally affected son with eccentric central cores in 3% of his type I fibres; a series of children with spinal muscular atrophy of varying severity; two adults with polymyositis and selective type II atrophy and a 4-year-old girl with a