absence of osteodystrophy in this and other reported cases who had chronic renal failure.

The earlier radiographs were loaned by Dr. H. R. Gamsu and Dr. J. Laws, King's College Hospital. Pathological studies were performed by Dr. R. A. Risdon, and we are grateful to Professor H. A. Sissons, Institute of Orthopaedics, for his opinion on the bone histology, to Dr. T. M. Barratt for permission to report the case, and to Professor C. E. Dent, University College Hospital, for advice.

REFERENCES

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‘Juvenile’ myasthenia gravis in early infancy

An infant aged 6 months is described with clinical features resembling the ‘juvenile’ form of myasthenia gravis. As far as we are aware this has not previously been described at this young age.

Case report

A female, aged 6 months was admitted to the Royal Children's Hospital on 8 October 1974, with a short history of lethargy and hypotonia. She had been a normal and active infant until 2 weeks before admission when she became drowsy and lethargic, and appeared
to have difficulty in clearing secretions from her pharynx. Her local doctor treated her with antibiotics and she apparently improved.

One week later she again became floppy, lethargic, and drowsy, and again developed noisy gurgling respirations. She was unable to feed properly, and her eyelids were drooping. She was admitted to a local hospital where pneumonia was diagnosed, and was treated with parenteral antibiotics. She required frequent aspiration of secretions and was fed through a nasogastric tube. However, her condition did not improve and after a brief respiratory arrest due to retained secretions she was transferred to this hospital.

Examination showed a floppy but alert infant with bilateral ptosis (Fig. 1). There was little spontaneous movement and she was hypotonic. However, muscle bulk and deep tendon reflexes were normal; there was no fasciculation of her tongue; pain sensation was normal; cough reflex was diminished; and auscultation of her chest showed evidence of retained secretions.

The combination of weakness, ptosis, and hypotonia, together with normal muscle bulk and deep tendon reflexes, suggested the diagnosis of myasthenia gravis and this was subsequently confirmed by her response to a neostigmine test. She was given 0·1 mg atropine, and 0·25 mg neostigmine 30 minutes later intramuscularly.

Fig. 1.—Before administration of neostigmine showing the expressionless face with bilateral ptosis.
Within several minutes she made spontaneous and vigorous movements for the first time since admission. There was a marked increase in muscle tone and strength and decrease in degree of ptosis (Fig. 2). No auto-antibodies to thyroid cytoplasm, striated muscle, and thymic myoid cells were found, and a chest radiograph showed a normal thymic shadow.

She was started on oral pyridostigmine 10 mg every 8 hours, with intramuscular neostigmine as necessary. Over the next few days the pyridostigmine was gradually increased but good control was established with a dose of only 50 mg every 4 hours. She was also started on oral supplements of potassium and calcium.

At the time of discharge from hospital muscle tone and power were considered to be normal, and cough reflex was good. A slight degree of ptosis was still present but acceptable in view of her generally satisfactory condition. She remained well for 6 weeks but was readmitted because of an episode of respiratory distress. During this short admission it was noted that she tired with feeds so the dose of pyridostigmine was increased to 80 mg 5 times a day. Over the next few weeks it was increased gradually to 100 mg 5 times a day because of persisting reduced muscular activity and ptosis.

With this higher dose she remained very well for several months until 12 March 1975. Shortly after being fed she was found by her mother lying limply in her cot. When picked up she vomited, stopped breathing, and became cyanosed. She was given intramuscular atropine and neostigmine, but died shortly afterwards in the local hospital. The cause of death was probably anoxia due to inhalation of vomitus. Necropsy examination was not performed.

**Discussion**

Myasthenia gravis is rarely seen in infancy. It has been described as occurring either as a transient phenomenon in a newborn child of a myasthenic mother, or as a congenital form in a child of a nonmyasthenic mother (Dubowitz, 1969). Infants born to myasthenic mothers may develop weakness, respiratory distress, and feeding difficulty soon after birth. Some are normal and require only close observation, while others need urgent treatment with anticholinesterase medication for respiratory insufficiency. The condition is self limiting and complete recovery takes place within several weeks.

The congenital form is characterized by involvement mainly of facial and extraocular muscles with relatively mild involvement of other muscle groups. It is often familial (Rothbart, 1937; Namba et al., 1971), an autosomal recessive inheritance having been postulated (Bundey, 1972). Clinical features are apparent within a few weeks of birth and persist throughout life. The course tends to be benign and nonprogressive (Namba et al., 1971).

Much more common than the above types is the 'juvenile' form of myasthenia gravis in which features become apparent after the first year of life, and usually at a much later age (Millichap and Dodge, 1960). There is no family history and girls are affected more frequently than boys. Ptsis and muscular weakness are the usual presenting features and respiratory difficulty is a frequent complication. Our patient's clinical presentation was of this type, as she was perfectly normal for the first 4 months of life. We believe this patient to be the youngest yet reported with the juvenile form of myasthenia gravis.

**Summary**

A previously well infant developed severe muscle weakness and hypotonia at 6 months of age. This was reversed by anticholinesterase medication. However, she had subsequent further weakness and died at 10 months after an acute respiratory arrest. The clinical pattern was that of the 'juvenile' form of myasthenia gravis rather than the 'congenital' forms which have previously been described in early infancy.
However, a similar picture may be encountered in some patients who have a congenital defect of testosterone synthesis (Givens et al., 1974) and in whom adrenal insufficiency may cause severe illness. Serious illness in a child wrongly diagnosed as a case of testicular feminization led to discovery of 17-α-hydroxylase deficiency.

**Methods**

Pregnanediol and pregnanetriol were estimated in urine by gas-liquid chromatography after enzymatic hydrolysis (Moolenaar and Van Seters, 1971). 11-Hydroxycortico-steroids in serum were estimated according to Mattingly (1962), cortisol and corticosterone by a competitive protein-binding technique (Dr. H. J. Degenhart, Sophia Children's Hospital Rotterdam), and testosterone, progesterone, and follicle stimulating hormone by radioimmunoassay.

**Case report**

A girl of 3 years was admitted to this hospital in a semicomatose condition. 6 months earlier she had been admitted to another hospital with inguinal hernia. A testicle was then found in the hernial sac, and karyotyping showed a 44,XY pattern, a diagnosis of testicular feminization being made. The external genitals were of normal female appearance; the vagina was present; neither uterus nor ovaries could be felt.

When admitted to our hospital the child had been ill for 24 hours and was found to have otitis media.

**TABLE**

Concentrations of main steroids in blood and urine

<table>
<thead>
<tr>
<th></th>
<th>Untreated</th>
<th>Treated with dexamethasone</th>
<th>Normal values in children</th>
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<tbody>
<tr>
<td></td>
<td>1 mg/d for 2 days</td>
<td>0·25 mg/d for 70 days</td>
<td></td>
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</table>

<table>
<thead>
<tr>
<th><strong>Urine</strong></th>
<th></th>
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<tbody>
<tr>
<td>17-oxosteroids mg/24 h (μmol/24 h)</td>
<td>0·61±0·2 (2·1±4·2)</td>
</tr>
<tr>
<td>17-oxogenic steroids mg/24 h (μmol/24 h)</td>
<td>2·4±8·3 (11·1±13·5)</td>
</tr>
<tr>
<td>Pregnanediol mg/24 h (μmol/24 h)</td>
<td>0·99 (0·31)</td>
</tr>
<tr>
<td>Pregnanetriol mg/24 h (μmol/24 h)</td>
<td>0·04 (0·12)</td>
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</table>

<table>
<thead>
<tr>
<th><strong>Serum</strong></th>
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</tr>
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<tbody>
<tr>
<td>11-OH steroids μg/100 ml</td>
<td>45±57</td>
</tr>
<tr>
<td>Cortisol μg/100 ml (nmol/l)</td>
<td>&lt;0·3 (8·28)</td>
</tr>
<tr>
<td>Corticosterone μg/100 ml</td>
<td>47</td>
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<tr>
<td>Testosterone ng/100 ml (nmol/l)</td>
<td>&lt;5 (0·17)</td>
</tr>
<tr>
<td>Testosterone after Pregnyl ng/100 ml (nmol/l)</td>
<td>&lt;5 (0·17)</td>
</tr>
<tr>
<td>Progesterone ng/ml (nmol/l)</td>
<td>4·3 (13·67)</td>
</tr>
<tr>
<td>Follicle stimulating hormone mIU/ml</td>
<td>17·0</td>
</tr>
</tbody>
</table>

**Blood pressure (mmHg)**: 145/100, 100/70, 100/70.

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*REFERENCES*


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**Female phenotype in a male child due to 17-α-hydroxylase deficiency**

The finding of testicles in a normal looking girl usually leads to a diagnosis of testicular feminization, i.e. congenital insensitivity to testosterone (due to lack of conversion to dihydrotestosterone).