Teased preparations of the sciatic nerve showed segmental demyelination.

Family History

The mother is 18 years old and unmarried. A 2-year-old sibling from a different father is normal. There is no family history of either ichthyosis or central nervous system disorder.

Discussion

Slow nerve conduction velocities and pathological changes have only recently been described in Krabbe's disease (Sourander and Olsson, 1968; Dunn et al., 1969). A similar demyelinating peripheral neuropathy with slow nerve conduction velocities also occurs in metachromatic leucodystrophy (Fullerton, 1964) and Cockayne's syndrome (Moosa and Dubowitz, 1970). Nerve conduction velocity measurement is therefore useful in separating some of the leucodystrophies from other progressive degenerative diseases of the nervous system, in which demyelinating peripheral neuropathy does not occur.

The association of the ichthyosis with Krabbe's disease in this patient may possibly be fortuitous, but there is some evidence to suggest that the two may be associated. Krabbe's disease is an inborn error of lipid metabolism in which deficiency of the enzyme cerebroside sulphatransferase results in an accumulation of cerebroside (Bachhawat, Austin, and Armstrong, 1967). Esterly (1968) has suggested that ichthyosis may be the result of a disturbance of lipid metabolism in the skin. Ichthyosis and peripheral neuropathy occur in Refsum's syndrome, in which the fatty acid phytanic acid accumulates in the body. Ichthyosis also occurs in two other central nervous system disorders, namely Sjögren-Larsson and Rud's syndromes. The ichthyosis and enteropathy in a case of Sjögren-Larsson syndrome was cured by substituting medium chain triglycerides for normal lipids in the diet (Hooft, Kriekemans, and Devos, 1967). A polyneuritis was also present in Rud's original case (Rud, 1927).

Motor nerve conduction studies may therefore be worth doing in patients presenting with ichthyosis and associated nervous system disorders.

Summary

A case of Krabbe's leucodystrophy with ichthyosis and peripheral neuropathy is described. This association has not previously been reported.

I wish to thank Dr. J. A. Black for referring the patient, Dr. V. Dubowitz for helpful advice and criticism, and Dr. A. D. Dayan for the cerebral histology.

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Birthweight in Haemolytic Disease of Newborn

Information about the birthweights of infants with haemolytic disease of the newborn (HDN) is surprisingly scant. Miller, Johnson, and Durlacher (1944) thought that these infants were heavier than normal, while Lind and Hyttten (1969) stated the reverse, though neither author reported figures. Javert (1942) found that birthweights of 22 non-hydropic infants with HDN did not differ from normal, while the mean birthweight of 16 infants with hydrops was 1 kg greater than those of normal infants of equivalent gestation. Naeye (1967) found that the necropsy weights of 23 infants with HDN (12 of whom had hydrops) did not differ from those of normal infants of the same gestational age. Karnicki (1968) found that birthweights of 88 infants with severe HDN were greater than normal. We have analysed birthweights of non-hydropic infants with HDN to determine whether they differed from the normal birthweight distribution of infants born at the Winnipeg General Hospital.

Material

The case records of all Rh negative mothers who had given birth to Rh positive liveborn, singleton infants between January 1966 and December 1968 were reviewed. 687 charts provided the following information about the infant: birthweight, gestational age
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Mean (±SE) Birthweights (g) by Gestational Age of Infants with Positive Coombs Test and of Normal Infant Population

<table>
<thead>
<tr>
<th>Gestational Age (wk)</th>
<th>34*</th>
<th>35</th>
<th>36</th>
<th>37</th>
<th>38</th>
<th>39</th>
<th>40</th>
<th>41</th>
<th>42</th>
<th>43</th>
</tr>
</thead>
<tbody>
<tr>
<td>Coombs positive group</td>
<td>2390±85</td>
<td>2520±101</td>
<td>2720±135</td>
<td>3030±68</td>
<td>3140±59</td>
<td>3380±84</td>
<td>3320±107</td>
<td>3740±288</td>
<td>3690±288</td>
<td>3030±593</td>
</tr>
<tr>
<td>Normal population</td>
<td>2630±79</td>
<td>2690±72</td>
<td>2880±43</td>
<td>3040±28</td>
<td>3230±20</td>
<td>3370±14</td>
<td>3450±13</td>
<td>3530±20</td>
<td>3570±27</td>
<td>3490±57</td>
</tr>
</tbody>
</table>

* (t = 1.904, p < 0.05).
† Figures in parentheses indicate number of patients.

(calculated in completed weeks from the first day of the mother's last menstrual period), the result of the direct Coombs test in the cord blood. Cord Hb levels were also available in some of the infants. 190 infants had a positive direct Coombs test and were diagnosed as having HDN. 26 of these had received fetal transfusions. None had hydrops.

Mean birthweights of the HDN infants were calculated for each week of gestation and compared with the mean weights of infants born at Winnipeg General Hospital (Pusey and Haworth, 1969).

Results

The Table shows the birthweights (mean±SE) of the HDN infant and also of 4547 unselected infants born at this centre, between 32 and 43 weeks of gestation. With the exception of 34 weeks' gestation, the mean weights in the two groups did not differ significantly. Cord Hb levels were recorded in 165 infants with HDN and ranged from 4.2 to 18.8 g/100 ml. In 62 infants Hb was less than 12 g/100 ml. No correlation between cord Hb and birthweight was found at any gestational age. The birthweights of infants with cord Hb <12 g/100 ml and of infants who received fetal transfusions were normal for their gestational age as shown in the Fig.

Discussion

During the preparation of this report, Kitchen (1970) published data on birthweights of liveborn HDN and normal infants. He found no difference between the two groups, but infants who had undergone fetal transfusion were not included. In the present series, infants receiving fetal transfusion had a normal birthweight distribution for their gestational age, as was also found by Lucey (1966).

When all HDN infants were considered together, birthweight was significantly less than the control population only at 34 weeks' gestation. This finding may be spurious and it is difficult to explain.

The present findings indicate that HDN does not interfere with fetal growth as measured by birthweight.

Summary

Birthweights of 190 infants with Rh haemolytic disease of the newborn (HDN) were compared with those of normal newborns. From 35 weeks' gestation on, no significant differences were noted between the two groups. Infants who received fetal transfusion had normal birthweights for their gestational ages and did not differ from other infants with HDN. It is concluded that HDN
does not interfere with fetal growth as measured by birthweight.

I wish to thank Dr. J. M. Bowman, Director, Rh Laboratory, Winnipeg, Manitoba, for permission to review the records of the Laboratory, and to Dr. J. C. Haworth and Dr. V. Chernick for guidance.

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Growth Retardation and Thyroxine-binding Globulin Deficiency

The combination of growth retardation and thyroxine-binding globulin (TBG) deficiency* had not been reported until recently when Nikolai (1969) presented an 11-year-old Caucasian boy with delayed bone age and zero TBG levels.

The present paper reports a second case of TBG deficiency associated with delayed bone age and growth retardation. The case differs in that the TBG levels were low but not zero.

Methods

Thyroid function studies. (See Table for normal values.) Blood for thyroid function studies was collected iodine-free and the serum separated.

Thyroid hormone levels. Total thyroxine iodine (total T₄ iodine) was determined by the method of Murphy and Pattee (1964). Values obtained are similar to those found using the older protein bound iodine (PBI) estimation, but the former test gives more reliable results in the presence of extraneous iodide contamination.

Free binding sites. Binding sites on serum thyroxin-binding proteins, not occupied by thyroid hormone, were estimated by the tri-iodothyronine resin uptake method of Woldring, Bakker, and Doorenbos (1961), and expressed as a percentage of a normal control serum (T₃ uptake % N).

Free thyroxine index. The free thyroxine index (FTI) is roughly proportional to the freely circulating thyroid hormone.

\[ \text{FTI} = \frac{(\text{Total } T₄ \text{ iodine}) \times (\text{T₃ uptake } \% \text{ N})}{100} \]

A normal FTI suggests euthyroidism and a low FTI hypothyroidism. It gives a more reliable estimate of the patient's thyroid status than when either the total T₄ or the T₃ uptake is considered singly.

Thyroid binding proteins. Thyroxine-binding globulin (TBG) and thyroxine-binding prealbumin (TBPA) were measured by a modified technique of Elzinga, Carr, and Beierwaltes (1961). Individual specimens of sera were enriched with ¹²⁵I labelled thyroxine (T₄) in chemical concentrations ranging from 30–500 μg/100 ml. The serum proteins were separated on paper (Schleicher and Schuell 2043A) by reverse flow electrophoresis and the T₄ binding fractions located by radioautography. The radioactivity in each fraction was counted, from which the T₄ binding capacities of TBG and TBPA were calculated.

Case Report

The patient was a 9-year-old boy who had been referred by his school medical officer for investigation of short stature and loss of visual acuity in his right eye.

On examination he was an alert boy 114 cm tall (5 cm less than the 3rd centile) and 21.8 kg in weight (1 kg greater than the 3rd centile). There was no clinical evidence of thyroid dysfunction. His pulse rate was 84/minute and blood pressure 100/70 mm Hg. Fundal examination and visual fields were normal. Lenses partially corrected a loss of visual acuity in his right eye from 6/18 to 6/9.

Radiological examination of his wrists showed a bone age of between 6 and 6½ years (Greulich and Pyle). A skull x-ray was normal.

The following were found to be within normal limits; the serum levels of albumin, globulin, uric acid, cholesterol, calcium, magnesium, phosphorus, aspartate transaminase, alkaline phosphatase, and creatinine; blood urea nitrogen and the urinary hydroxyproline excretion; cystic activity in stools and xylose tolerance test.

Endocrine function studies included a normal 24-hour urinary excretion of 11-hydroxycorticosteroids and...