lymphoma as to be virtually diagnostic of Burkitt's tumour.

The girl received cyclophosphamide (40 mg/kg per day). A short and incomplete remission was obtained, but she died after 15 days with a complete intestinal obstruction. To our knowledge, this is the second case (Rufino and Pavlovsky, 1972) described in our country, and the first in the southern zone.

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REFERENCES

Hyperlysinaemia

Sir,

I would like to add to the article by Drs. van Gelderen and Teijema (Archives, 1973, 48, 892) by recording three important findings that support their thesis that hyperlysinaemia due to lysine-ketoglutarate reductase deficiency is a harmless inborn error of metabolism. 4 patients with this condition have been under biochemical and clinical scrutiny for periods up to 12 years. 3 are sibs: a girl (the propositus) now 12½ years, a boy 9½, and a girl 8. The fourth patient, a cousin, is now 18½ years old. Though many clinical and biochemical features of these familial hyperlysinaemics have been recorded (Woody, 1964; Woody, Hutzler, and Dancis, 1966; Woody, Ong, and Pupene, 1967; Dancis et al., 1969) three important findings have not been published.

First, 3 of these children have had normal growth and development. Growth and developmental delays were seen only in the index case and probably represent a sampling artifact. The other children manifest the biochemical findings of hyperlysinaemia to the same degree as the propositus and have normal growth and intelligence. They have also lacked the ligamentous and muscular ashenia found in the index patient. Only the boy, with bilateral ectopia lentis, might be considered to have a connective tissue abnormality somehow related to defective lysine metabolism. All 3 have had normal EEGs.

The second important observation is that 2 of these children, the boy and younger girl, did not manifest increased serum lysine levels until they were 6 months old. Since it must be assumed that the enzyme defect was present at birth, an alternate minor pathway via pipecolic acid has been postulated to function actively below the age of 1 year (Woody and Pupene, 1970, 1971). This serves to maintain normal lysine levels during early infancy when rapid growth—and its high requirement of lysine for protein synthesis—keeps lysine catabolism low and within the capacity of the pipecolic acid pathway to handle. As growth slows after 6 months, the balance between absorption and utilization of lysine shifts towards the catabolic side. Such a shift 'unmasks' the defect in lysine-ketoglutarate reductase activity with an accumulation of lysine and lysine metabolites.

The third finding is the presence in these children of normal levels of serum ammonia despite serum lysine and arginine levels far in excess of those reported for patients with congenital lysine intolerance (Table). The relation of lysine metabolism to ammonia production remains obscure.

TABLE

<table>
<thead>
<tr>
<th></th>
<th>Ammonia (μg/100 ml)</th>
<th>Lysine (μmol/l)</th>
<th>Arginine (μmol/l)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Familial hyperlysinaemia</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Sib 1</td>
<td>43-5</td>
<td>1354</td>
<td>110</td>
</tr>
<tr>
<td>Sib 2</td>
<td>46-2</td>
<td>1016</td>
<td>106</td>
</tr>
<tr>
<td>Sib 3</td>
<td>38-2</td>
<td>835</td>
<td>108</td>
</tr>
<tr>
<td>Congenital lysine intolerance*</td>
<td>270-370</td>
<td>47</td>
<td>36</td>
</tr>
<tr>
<td>Normal values</td>
<td>18-48†</td>
<td>71-151‡</td>
<td>25-86‡</td>
</tr>
</tbody>
</table>


These observations indicate (1) that familial hyperlysinaemia probably is clinically benign, neither interfering with growth nor causing mental retardation; (2) that it is not associated with hyperammonaemia; and (3) since it may not be biochemically manifest for several months after birth, it can be missed in neonatal screening programmes for aminoacidopathies.

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REFERENCES