Blood Enzymes in the de Lange Syndrome

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The de Lange syndrome is a clinical entity in which retardation of mental and physical development occurs in association with a number of other characteristic features, involving particularly the face and limbs (Berg *et al*, 1970). Dahlqvist, Hall, and Källén (1969) reported that galactose-1-phosphate uridyl transferase activity in red blood cells of patients with de Lange syndrome was elevated. More recently, Daniel and Higgins (1971) found increased serum α -ketoglutarate and serum glutamate levels in patients with the characteristic features of this syndrome. Since the aetiology and the pathophysiology of the de Lange syndrome has not been established it was decided that a careful look at the red and white blood cell enzymes was indicated in this syndrome.

Material and Methods

Ten to 20 ml of heparinized blood was obtained from patients and matched controls. The blood was sedimented in the cold for 45–90 minutes with 6% dextran in normal saline. These were mixed in a ratio of 1 ml of dextran per 5 ml of blood. The white cell rich supernatant was removed to within 0.2 cm of the red blood cells. For leucocyte glucose-6-phosphate dehydrogenase the blood was sedimented with 3% fibrinogen solution in Tris buffer pH 7.0 containing 50 mM β -mercaptoethanol.

Leucocyte and erythrocyte galactose-1-phosphate

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It is with great regret that I have to report the death of Professor Hsia on 27 January 1972—Ed.

uridyl transferase were assayed according to the method of Mellman and Tedesco (1965).

The tests for glucose-6-phosphate dehydrogenase, acid phosphatase, and alkaline phosphatase were done with white cells prepared by washing with 0.2% saline to lyse the red cells. The white cells for acid phosphates and alkaline phosphates were lysed with 100 mg% solution of saponin in water. For the glucose-6-phosphate dehydrogenase, the white cells were lysed in Tris buffer 0.19 M pH 8 containing 10 mg% saponin and 0.4 mM TPN.

Erythrocyte glucose-6-phosphate dehydrogenase was assayed by the method of Zinkham (1959). Leucocyte glucose-6-phosphate dehydrogenase was assayed by the method of Shih *et al* (1965). Acid and alkaline phosphatase were assayed using p-nitrophenol phosphate as substrate (Bessey *et al*, 1946).

Results

The results of the enzyme studies are seen in Table I. While there were minor differences between the results of the de Lange and control groups, none is statistically significant.

In general, the values for the de Lange patients were more variable than those for the control group suggesting greater heterogeneity. However, there was no evidence of any consistent blood cell enzyme changes as has been described with certain other chromosomal disorders (Hsia *et al*, 1971).

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TABLE I

	Controls		De Lange	
	No.	Mean $\pm$ SD	No.	Mean $\pm$ SD
<i>White blood cells</i>				
Galactose-1-phosphate uridyl transferase (μ M/hr/ 10^6 WBC)	12	14.8 $\pm$ 3.1	11	18.5 $\pm$ 5.5
Glucose-6-phosphate dehydrogenase (μ M/hr/ 10^6 WBC)	5	1.08 $\pm$ 0.20	5	0.92 $\pm$ 0.22
Acid phosphatase (mg P/hr/mg protein)	11	147.0 $\pm$ 37.0	7	164.0 $\pm$ 35.0
Alkaline phosphatase (mg P/hr/mg protein)	11	8.4 $\pm$ 1.6	6	8.8 $\pm$ 3.5
<i>Red blood cells</i>				
Galactose-1-phosphate uridyl transferase (μ M/hr/ml RBC)	12	5.7 $\pm$ 1.8	13	6.0 $\pm$ 2.0
Glucose-6-phosphate dehydrogenase (μ M/hr/ml RBC)	8	1.69 $\pm$ 0.3	8	1.87 $\pm$ 0.49
6-Phosphogluconate dehydrogenase (μ M/hr/ml RBC)	6	1.63 $\pm$ 0.31	6	1.92 $\pm$ 0.45

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