In the homocystinuric patient, cystathionine synthetase is absent or abnormal, and this results in an increase in the concentration of the blood and urine homocystine and in an absence of cystine. As the reaction methionine \( \rightleftharpoons \) homocysteine is reversible, methionine levels are also found to be raised in the serum. It is these findings that are used to establish a diagnosis.

Because pyridoxine is an essential co-factor in the enzymatic reaction of homocysteine to cystathionine, two groups of workers have given this vitamin in high dosage, and have reported biochemical success in some of their patients.

The response of 10 patients with homocystinuria, treated with oral pyridoxine and a normal mixed diet, is presented. In 5, the biochemical changes were normalized, i.e. homocystine concentrations in the serum have been much reduced, and cystine has appeared. In those cases in which the serum methionine concentration was raised, it has reverted to normal. The biochemical and therapeutic implications of these results were discussed.

M. M. SEGAL introduced by DR. JUNE K. LLOYD (London). 'Treatment of Familial Hypercholesterolaemia in Childhood.' Familial hypercholesterolaemia is associated with a high incidence of ischaemic heart disease in adult life. Treatment has therefore been instituted in affected children in an attempt to reduce this risk. Our experience with 12 heterozygous patients (aged from birth to 16 years) and one homozygous patient (aged 10 years) are presented. The homozygous patient developed tuberous and tendon xanthomata at the age of 18 months; her pretreatment serum cholesterol level was 1050 mg./100 ml. The heterozygous patients did not have xanthomata, but 2 had corneal arcus; their pretreatment serum cholesterol levels were 284–510 mg./100 ml. (mean 375).

Treatment reduced the serum cholesterol levels in all the patients. In the heterozygous children this was achieved by dietary means alone. The diet consisted of restriction of saturated fat and the addition of corn oil to provide polyunsaturated fat. A significant negative correlation was found between the percentage reduction in serum cholesterol and the amount of saturated fat allowed in the diet \((r = -0.80; p < 0.01)\). In the homozygous child treatment had been previously unsuccessful, and cholestyramine and clofibrate were therefore used in addition to diet. A fall in serum cholesterol of about 400 mg./100 ml. was achieved, and this reduction has been maintained over a period of one year; though the levels remain high (589–675 mg./100 ml.), the xanthomata are regressing.