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SICKLE CELL ANEMIA IN ASSOCIATION WITH α-THALASSEMIA-2: BIOSYNTHETIC AND HEMATOLOGICAL STUDIES

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Patients with Sickle Cell Anemia (SS) associated with homozygous α-thalassemia-2 ($-\alpha/-\alpha;\beta^S/\beta^S$) are difficult to detect because the *in vitro* synthesis of hemoglobin chains may be balanced after prolonged incubation (>120 min). However, a distinct imbalance can be present at early incubation times. We studied chain synthesis on whole cell globin at 10, 30, and 120 min incubation in 38 SS and 4 S/β^0-thal patients and compared the data with hematological observations. There was good correlation between duplicate analyses of the $\alpha/(\beta+\gamma)$ (total CPM/ml in respective chromatographic zones) ratios. Most of the data at 120 min clustered around 1.0 except for 4 patients with S/β^0-thal and 4 of the patients with $-\alpha/-\alpha;\ \beta^S/\beta^S$. At 10 and 30 min incubations, however, four groups of patients could be distinguished. These had $\alpha/(\beta+\gamma)$ ratios suggesting discrimination between patients with the $-\alpha/-\alpha;\ \beta^S/\beta^S$ (n=11), $-\alpha/\alpha\alpha;\ \beta^S/\beta^S$ (n=16), $\alpha\alpha/\alpha\alpha;\ \beta^S/\beta^S$ (n=11), and S/β^0-thal (n=4) assumed genotypes. Limited data on some families were consistent with the assignments. Some patients with the $-\alpha/-\alpha;\ \beta^S/\beta^S$ genotype had MCV values of more than 80 fl, but not all patients with an MCV value below 80 fl had this genotype. The SS patients with an associated α-thalassemia-2 heterozygosity ($-\alpha/\alpha\alpha$) or homozygosity ($-\alpha/-\alpha$) tended to have less severe hemolysis than the SS patients with 4 active α chain genes ($\alpha\alpha/\alpha\alpha$).