

Screening of newborn infants for hypothyroidism should be done twice

The background of the study. Infants with congenital hypothyroidism usually appear normal at birth, but have mental retardation if the hypothyroidism is not recognized very soon after birth. Screening of newborn infants for hypothyroidism has proven highly successful, allowing early treatment and therefore preventing retardation, but some affected infants are missed by screening. This study evaluated the results of second screening.

How the study was done. The first and second screening-test results for all infants born in Colorado from 1996 to 2004 were reviewed. The first blood sample was collected within three days after birth. Thyroxine (T4) was measured, and if the value was low thyrotropin (TSH) was measured. TSH values <20 mU/L were considered normal. If the TSH value was borderline (20 to 39 mU/L), follow-up for second screening was initiated. If the TSH value was presumed positive (>39 mU/L), the infant was recalled immediately for serum testing. The second screening test consisted of the same measurements with the same cut-off values, except that any TSH value >20 mU/L was considered positive, and the infant was recalled for serum testing.

The results of the study. During the study period, 494,324 infants were screened soon after birth (usually on day 2), and 471,877 (95.4 percent) were screened a second time (during days 8 to 14). The first test identified 185 infants with hypothyroidism (1 in 2672) and the second 42 infants, giving a total of 227 infants (1 in 2178); the infants identified by the second test constituted 18.5 percent of the total.

The results of the first test were borderline in 3083 infants. Among them, 2938 (95.3 percent) had a normal second test and 26 (0.8 percent) had hypothyroidism. The results of the first test were presumed positive in 463 infants. Among them, 300 (64.8 percent) had normal serum tests and 159 (34.3 percent) had hypothyroidism. The results of the second test were borderline in 70 infants and presumed positive in 34 infants, of whom 22 (31.4 percent) and 20 (58.8 percent), respectively, had hypothyroidism.

The conclusions of the study. Screening of newborn infants for hypothyroidism a second time at age 7 to 14 days identifies additional cases.