High Risk of Congenital Hypothyroidism in Multiple Pregnancies

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Context: In Italy, the surveillance of congenital hypothyroidism (CH) is performed by the Italian National Registry of Infants with CH (INRICH). Up to now, about 3600 infants with CH are recorded in the INRICH, and a high number of twins are included.

Objective: Our objective was to estimate the risk of CH in multiple and single deliveries and to compare neonatal features of CH twins with twins from the general population.

Design: The Italian population of CH infants recorded in the INRICH from 1989–2000 was investigated.

Results: A more than 3-fold higher frequency of twins was found in the CH population than in the general population, and for the first time, it was possible to estimate the CH incidence in multiple (10.1 in 10,000) and single deliveries (3.2 in 10,000 live births). Significantly higher frequencies of in situ gland as well as lower TSH mean level at screening were found in twin than in singleton CH babies. The concordance rate for permanent CH was very low (4.3%) and due to only three concordant couples. However, a high recurrence risk for CH was estimated in siblings of affected babies recorded in the INRICH, including twins considered as siblings.

Conclusions: The high CH incidence observed in twins is worthy of interest for the high number of induced pregnancies in Italy as well as in other Western countries. Moreover, the low concordance rate for CH among twins together with a high recurrence risk for the disease among siblings indicates that environmental risk factors may act as a trigger on a susceptible genetic background in the etiology of the disease.