# Role of Next Generation Sequencing in the Etiological Diagnosis of Permanent Congenital Hypothyroidism With in Situ Thyroid

NCT06728735

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| **Status** | RECRUITING |
| **Sponsor** | IRCCS Azienda Ospedaliero-Universitaria di Bologna |
| **Enrollment** | 350 participants |

## Key Eligibility Criteria

### Inclusion (5)

- Patients born in Emilia-Romagna region, Italy, between January 2003 and December 2023;
- Patients screened at the Regional Neonatal Screening Centre for Endrocrine-Metabolic Diseases, IRCCS Azienda Ospedaliero-Universitaria of Bologna, Italy, and recalled for suspected congenital hypothyroidism;
- Confirmed diagnosis of congenital hypothyroidism and in situ thyroid;
- Hormonal and clinical follow-up of at least 36 months at Centre for Endrocrine-Metabolic Diseases, IRCCS Azienda Ospedaliero-Universitaria of Bologna, Italy;
- Obtaining informed consent from parents/legal guardians of paediatric patients.

### Exclusion (1)

- • Patients with hypothyroidism associated with chromosomal syndromes.

## Locations (1 total)

IRCCS Azienda Ospedaliero-Universitaria di Bologna, Bologna, Bologna, Italy