

Gonadic Function and Pubertal Development in Female Patients With Classic Galactosemia

NCT07461519

Status RECRUITING
Sponsor IRCCS Azienda Ospedaliero-Universitaria di Bologna
Enrollment 60 participants

Key Eligibility Criteria

Inclusion (2)

- Female patients followed at participating Pediatric Endocrinology and Inherited Metabolic Disease Centers, born between January 1st, 1980 and December 31st, 2024, with a diagnosis of classic galactosemia and confirmed by genetic testing.
- Obtaining informed consent.

Exclusion (2)

- Patients with known chromosomal abnormalities (e.g., trisomy 21, Turner syndrome, Fragile X syndrome, Kabuki syndrome, or other genetic conditions associated with pubertal disorders);
- Patients with primary ovarian insufficiency due to causes other than galactosemia.

Locations (19 total)

SSD Nutrizione Clinica e Metabolismo - IRCCS Azienda Ospedaliero-Universitaria di Bologna, Bologna, Italy, Italy
UOC Pediatria - IRCCS Azienda Ospedaliero-Universitaria di Bologna, Bologna, Italy, Italy
Ospedale Pediatrico "Giovanni XXIII", Bari, Italy
... and 16 more locations