

PREVALENCE OF DUODENAL ATRESIA IN PATIENTS WITH DOWN SYNDROME AT THE NATIONAL INSTITUTE OF PEDIATRICS, RELATED COMORBIDITIES, AND MATERNAL AGE.



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INTRODUCTION

Down syndrome is associated with various medical anomalies. Duodenal atresia is the most common gastrointestinal malformation in these patients. Despite being historically described, its precise etiology remains partially unknown.

METHODS

Retrospective, cross-sectional study of patients with Down syndrome evaluated during the period 2003–2023. A **representative sample of 518** was selected from 3,834 patients using Epidat (4% margin of error). Patients aged **0–17 years** with a history of **surgical repair** were included; those without a surgical report, or lacking maternal age data were excluded. **Cases of duodenal atresia** were further evaluated for demographic and clinical variables.

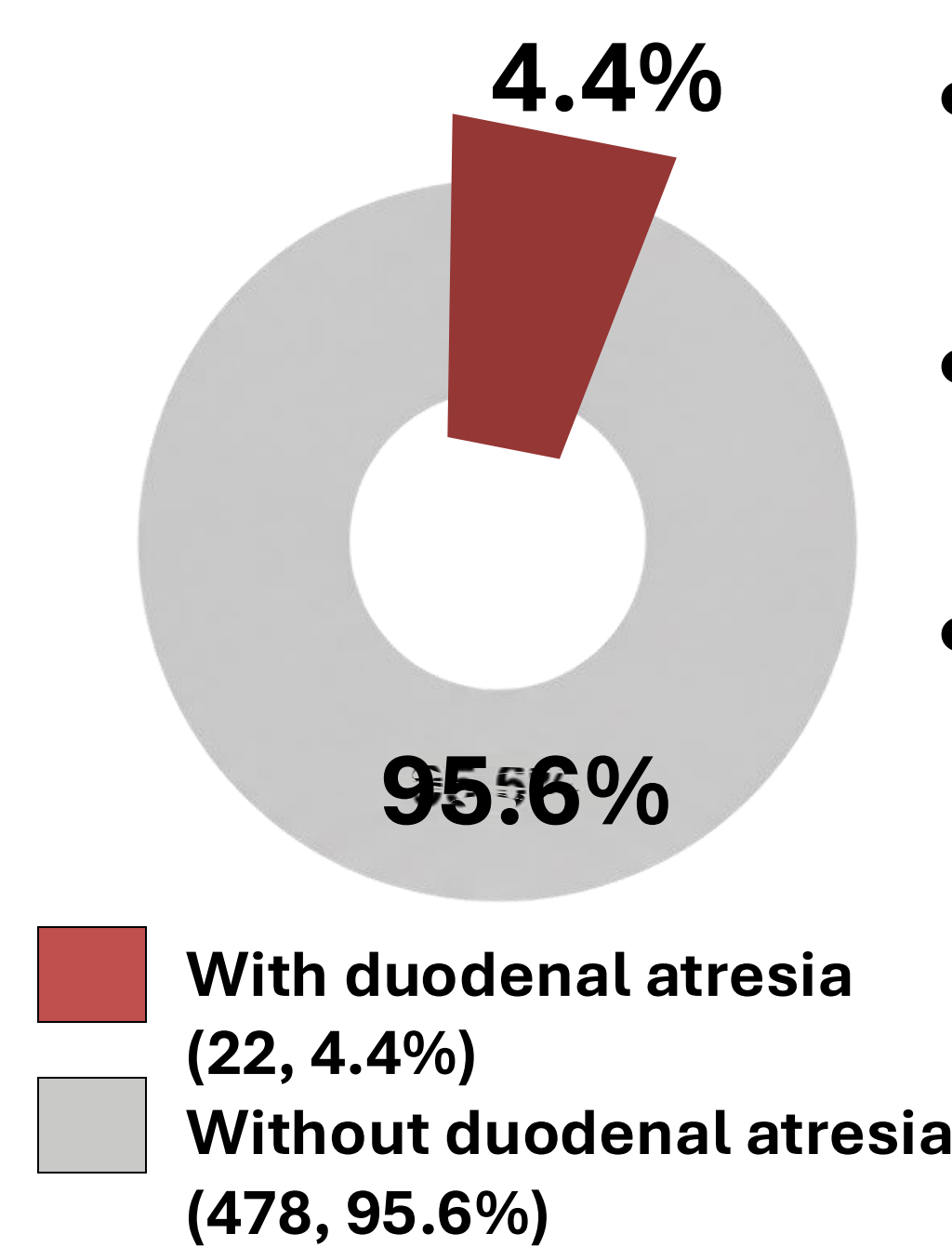


Figure 1. Prevalence of duodenal atresia.

Most cases were diagnosed at birth (21/22, 95%), with **only one prenatal diagnosis**.

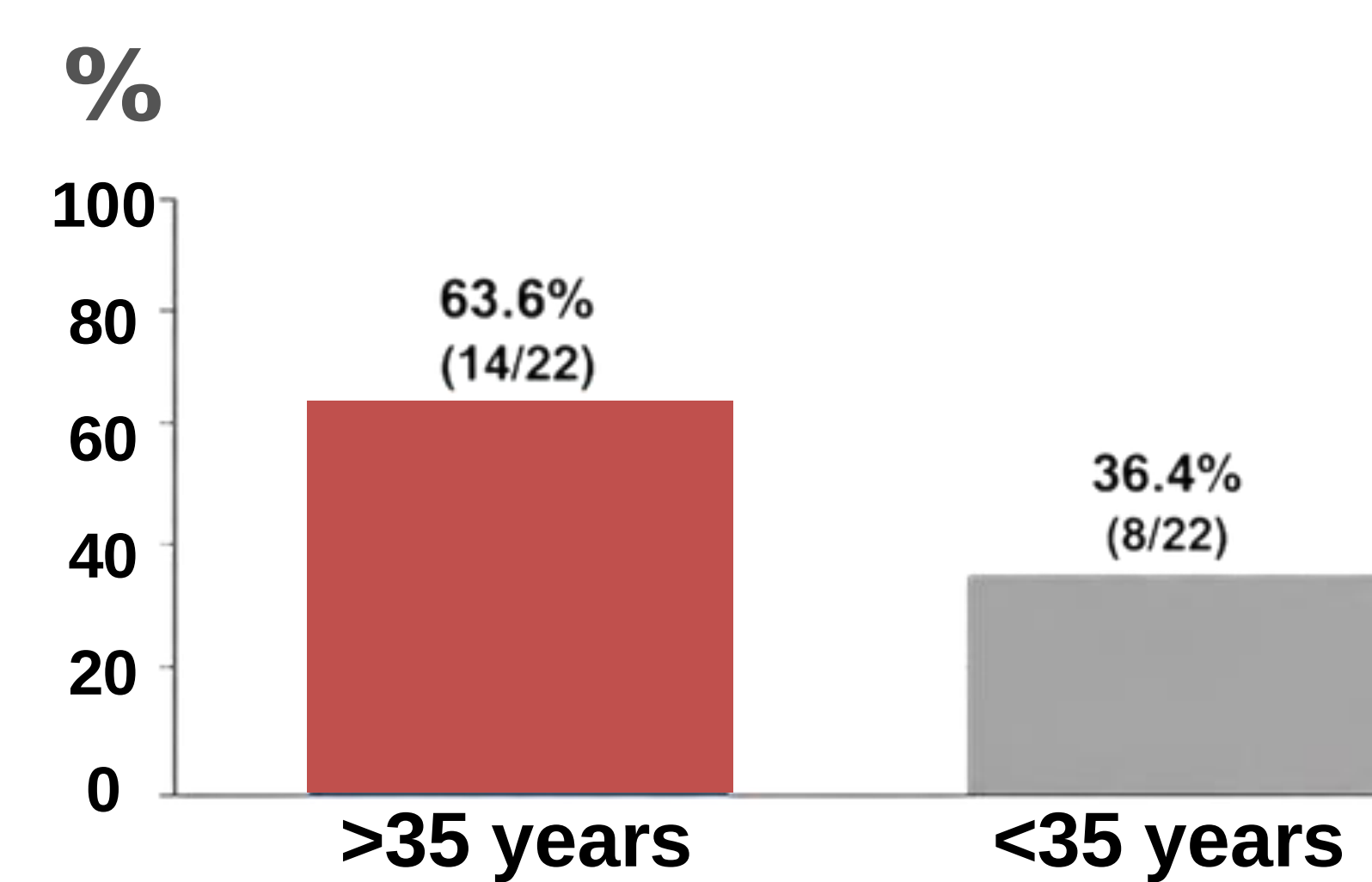


Figure 3. Maternal age in patients with duodenal atresia

- A **total of 500** patients with Down syndrome were included.
- **247 (49.4%) males** and **253 (50.6%) females**.
- **Duodenal atresia** was identified in **22/500** patients (prevalence of 4.4%).

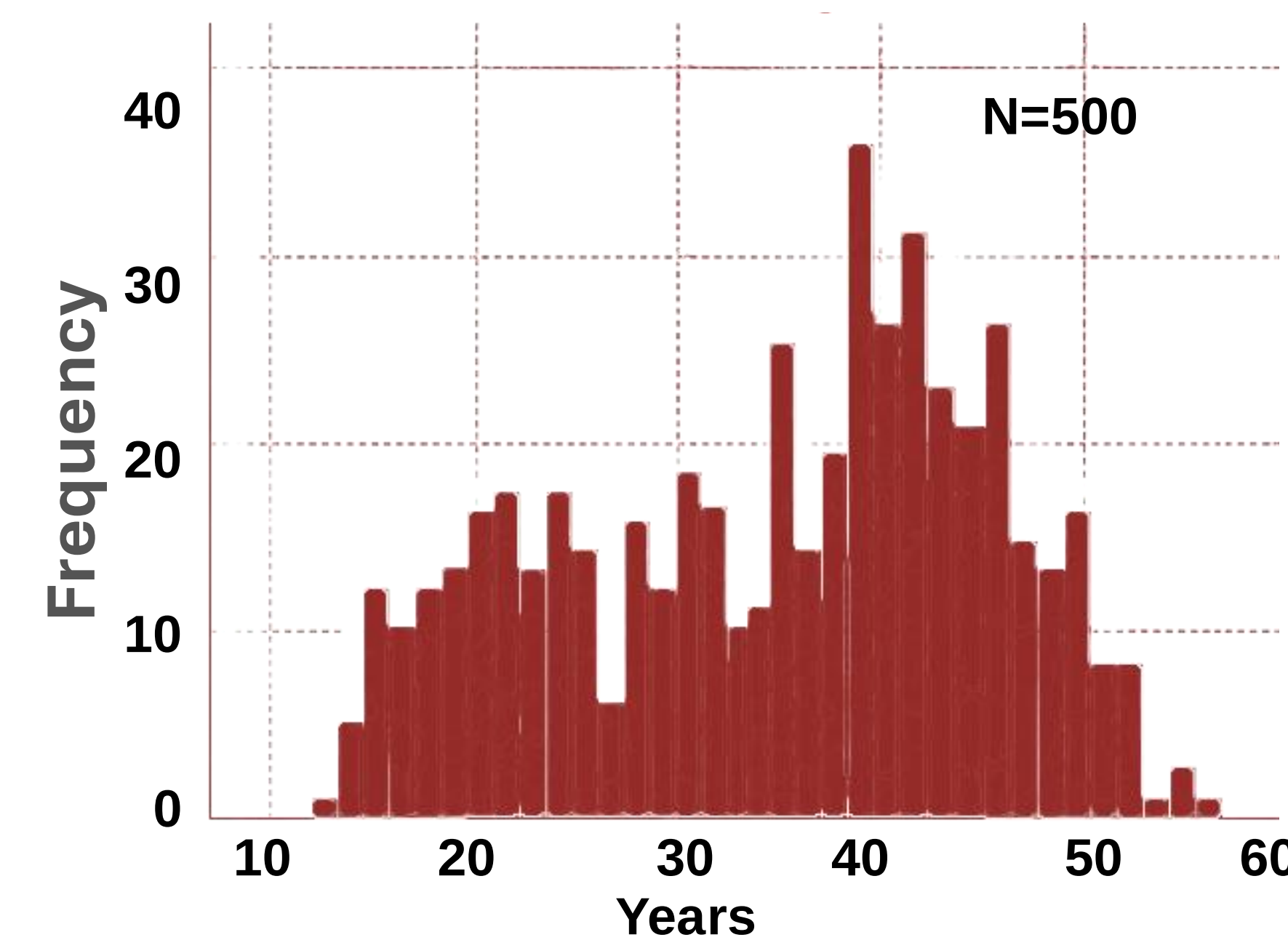


Figure 2. Maternal age at the time of conception

Advanced maternal age (>35 years) was observed in 225/500 (45.0%) cases in the overall cohort, compared with 14/22 (63.6%) cases among patients with duodenal atresia.

CONCLUSIONS

The findings are consistent with a **possible association between duodenal atresia, advanced maternal age, and embryologically related comorbidities**, such as annular pancreas and intestinal malrotation. Only 1/22 cases were diagnosed prenatally, **highlighting the need to improve prenatal detection**. Despite limitations related to its retrospective design and single-center setting, further studies are needed to identify factors and potential genotype–phenotype correlations.

RESULTS

COMORBIDITY	FREQUENCY IN COHORT (N=500)	ASSOCIATION WITH DUODENAL ATRESIA (Among patients with the comorbidity)	PREVALENCE IN PATIENTS WITH DUODENAL ATRESIA (N=22)
Congenital Heart Disease	328/500 (65.6%)	DA (+) 16 (4.9%) DA (-) 312 (95.1%)	16/22 (72.7%)
Umbilical Hernia	20/500 (4.0%)	DA (+) 0 (0%) DA (-) 20 (100%)	0/22 (0%)
Intestinal Malrotation	5/500 (1.0%)	DA (+) 3 (60%) DA (-) 2 (40%)	3/22 (13.6%)
Annular Pancreas	5/500 (1.0%)	DA (+) 4 (80%) DA (-) 1 (20%)	4/22 (18.2%)

Figure 4. Associated surgical comorbidities.

Comorbidities were evaluated by organ system.

- **Congenital heart disease was the most frequent comorbidity**, present in 16/22 of patients with DA.
- **Umbilical hernia** was the most common abdominal condition (20/500, 4%) but was **not associated with duodenal atresia**.
- **Intestinal malrotation** (3/5 cases) and **annular pancreas** (4/5 cases) were **more frequent** among patients with duodenal atresia, suggesting possible associations.
- No other specific associations were identified.

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