

From Diagnosis to Outcome: Genetic Insight into Fetal Gastrointestinal Tract Anomalies

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Introduction

The incidence of Gastrointestinal tract anomalies is 0.02–0.07%, which accounts for 7–9% of all congenital anomalies. Associations with other anomalies significantly impact the outcome of the baby. Accurate molecular diagnosis further helps in predicting risk of recurrence and providing prenatal diagnosis.

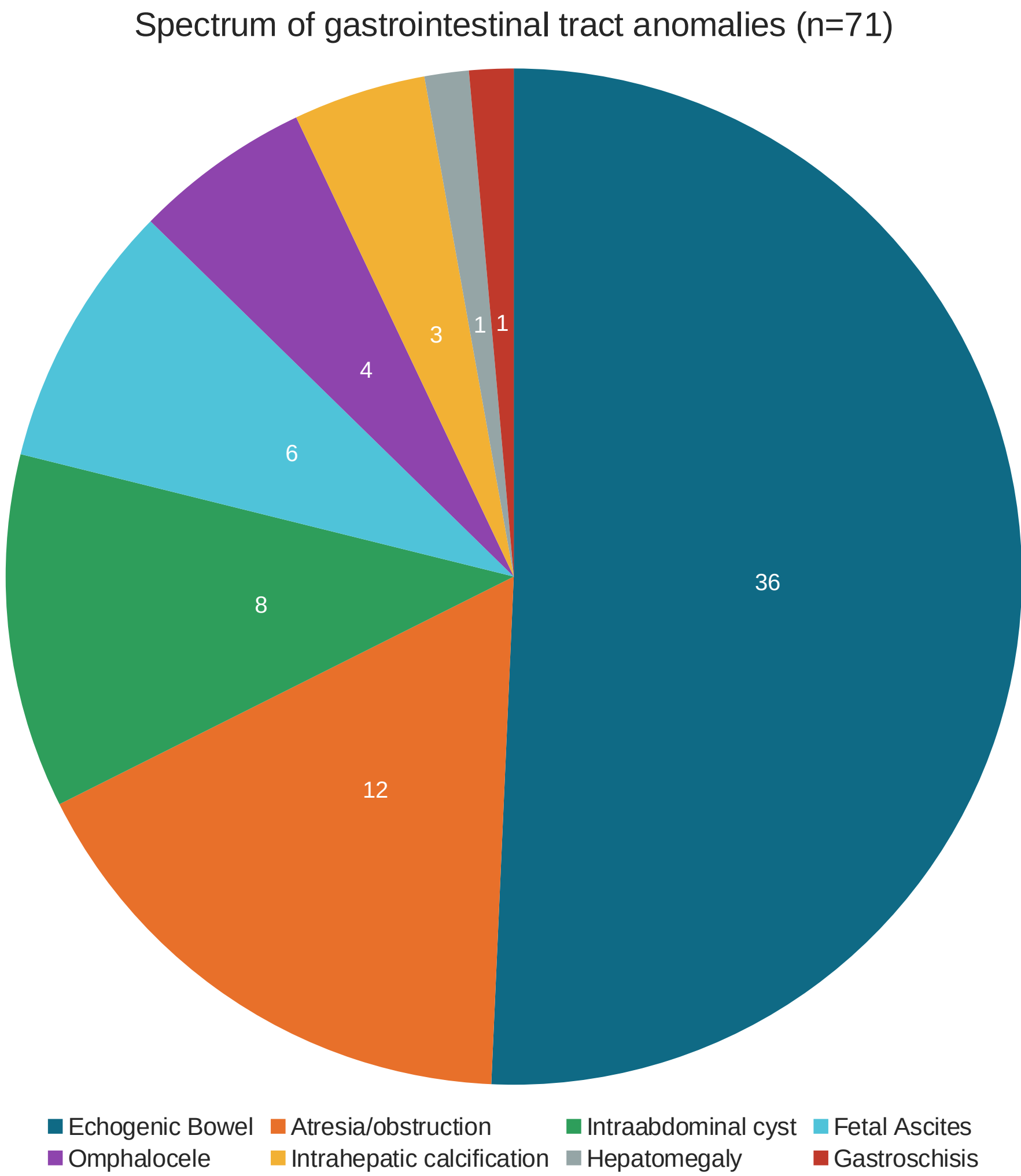
Objective

Spectrum, associations, genetic correlations and outcome of fetal Gastrointestinal tract anomalies.

Methods

A retrospective observational study conducted at a tertiary referral center between August 2023 and August 2025. All patients with Gastrointestinal tract anomalies were evaluated for association with other anomalies and genetics.

Results



Associations with other anomalies including genetics				
Anomaly	Screen Positive	Invasive test done	Invasive test positive	Association with other anomaly
Echogenic Bowel	2	8	2 (25%)*	18 (50%)
Atresia/obstruction	0	1	1 (100%)*	9 (75%)
Intraabdominal cyst	0	2	0	2 (25%)
Fetal Ascites	0	2	1 (50%)*	6 (100%)
Omphalocele	0	1	0	2 (50%)
Intrahepatic calcification	0	0	0	1 (33%)
Hepatomegaly	0	0	0	1 (100%)
Gastroschisis	0	0	0	0
Total	2 (2.81%)	14 (19.71%)	4 (28.5%)	39 (54.9%)

Genetic associations	
Findings	Genetic associations
Echogenic bowel, Fetal ascites, Cystic hygroma, ventriculomegaly	Trisomy 18
Echogenic bowel, EIF, VSD, Microcephaly	Pathogenic variant TYREX gene (Aicardi-Goutieres Syndrome 1), Homozygous, AR
Echogenic Bowel, Dilated bowel loops	CFTR, Cystic Fibrosis, Homozygous, AR
Double Bubble at 27 weeks	Trisomy 21

Outcome			
Anomaly	Live birth (%)	IUD + Neonatal death	Abortion
Echogenic Bowel	26 (72%)	1+1	7
Atresia/obstruction	6 (50%)	0	6
Intraabdominal cyst	8 (100%)	0	0
Fetal Ascites	2 (33.34%)	2	2
Omphalocele	2 (50%)	0	2
Intrahepatic calcification	1 (33%)	2	0
Hepatomegaly	1 (100%)	0	0
Gastroschisis	1 (0%)	0	1
Total	47 (66.19%)	6 (8.45%)	18 (25.35%)

Gastroschisis



Omphalocele



Meconium Pseudocyst



Double bubble sign



Conclusion

Risk stratification should be done not on the basis of organ system but on the basis of the anomaly, its association with other anomalies and with genetics. Invasive testing should be offered to all patients with Gastrointestinal system anomalies irrespective of first or second trimester biochemical screening result.