

Spectrum, Associations and Genetic Correlations of Fetal Urinary System Anomalies: A Tertiary Referral Center Study

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Introduction

The incidence of Urinary system anomalies is 0.5–1%, which accounts for 12–14% 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.

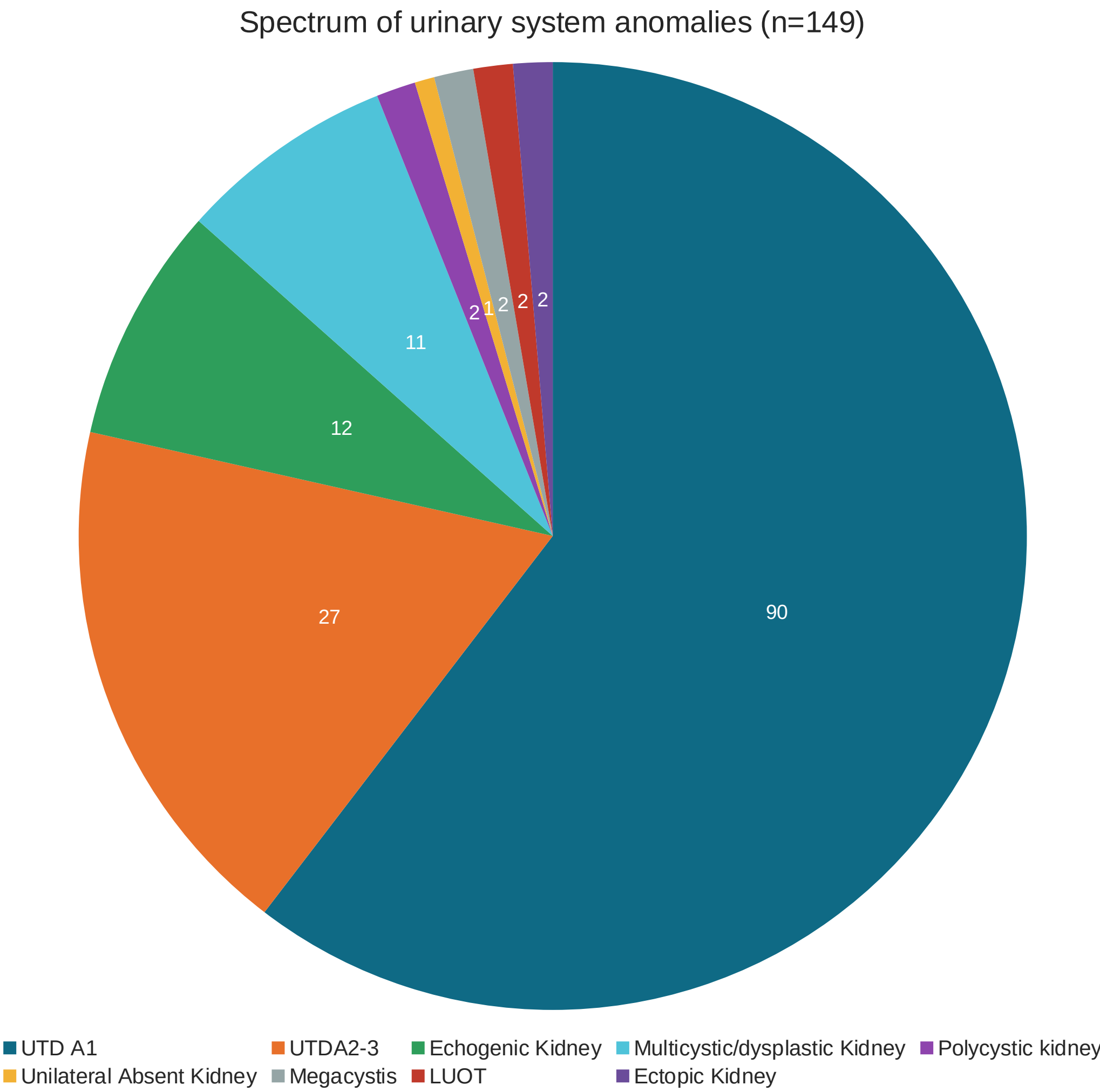
Objective

Spectrum, associations, genetic correlations and outcome of fetal Urinary system anomalies.

Methods

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

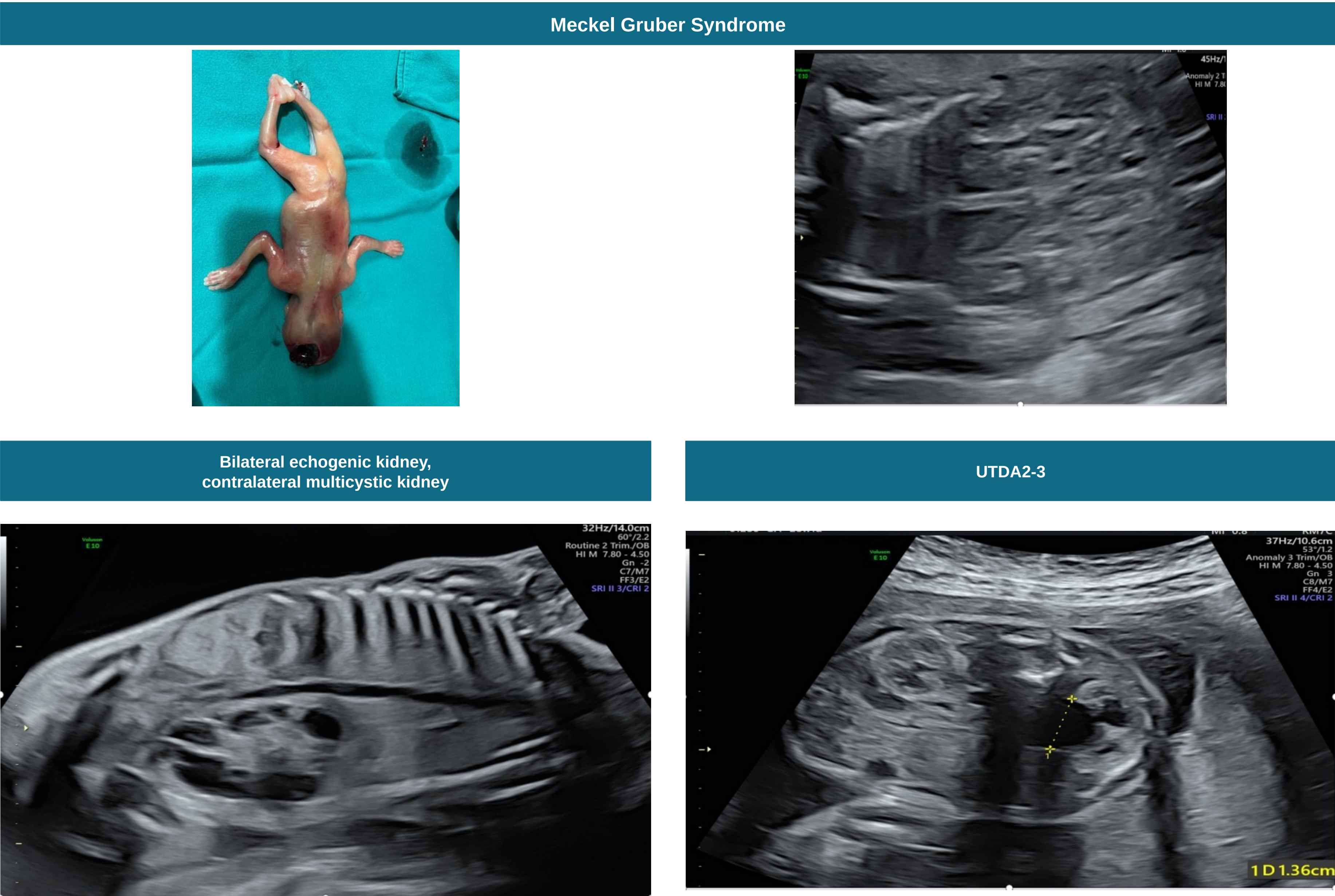
Results



Associations with other anomalies including genetics				
Anomaly	Screen Positive	Invasive testing done	Invasive test positive	Association with other anomaly
UTD A1	2	10	0	17 (18%)
UTDA2-3	2	4	2 (50%)	4 (14.81%)
Echogenic Kidney	1	7	2 (28%)*	7 (58%)
Multicystic/dysplastic Kidney	0	1	1 (9.09%)*	5 (45%)
Polycystic kidney	0	1	1 (50%)*	2 (100%)
Unilateral Absent Kidney	0	0	0	0
Megacystis	0	1	1 (50%)*	0
LUOT	0	0	0	1 (50%)
Ectopic Kidney	0	1	1 (50%)*	1 (50%)
Total	5 (3%)	25 (16.7%)	8 (32%)	37 (24.8%)

Genetic associations	
Findings	Genetic associations
Unilateral multicystic kidney, Bilateral echogenic kidney	17 q deletion
Early onset FGR, choroid plexus cyst, cerebellar hypoplasia, TR, echogenic kidney	Trisomy 18
UTDA2-3, echogenic kidneys	5-Oxoprolinase Deficiency, Heterozygous, AR
B/L Echogenic kidney, EIF	Gain in X Chromosome (Xq duplication)
Megacystis	POMT1, Muscular Dystrophy Dystroglycanopathy Heterozygous, AR
Polycystic kidneys, Occipital encephalocele, CTEV	MSK1 Exome 8, MSK1 Exome 5, Meckel Gruber syndrome
Ectopic kidney, Echogenic bowel, EIF, Cerebellum at 8th centile, increased NF thickness, right aortic arch, Redundant foramen ovale	Heterozygous BBS10 gene for Bardet-Biedl syndrome
Early FGR, EIF, B/L UTDA2-3	Silver-Russell syndrome, SRS3, Heterozygous AD

Outcome			
Anomaly	Live birth (%)	IUD + Neonatal death	Abortion
UTD A1	85 (94%)	1	4
UTDA2-3	23 (85%)	0	4
Echogenic Kidney	6 (50%)	1	4
Multicystic/dysplastic Kidney	1 (8.3%)	1	9
Polycystic kidney	0 (0%)	0	2
Unilateral Absent Kidney	1 (100%)	0	0
Megacystis	0 (0%)	0	2
LUOT	1 (100%)	0	1
Ectopic Kidney	1 (50%)	0	1
Total	117 (78.52%)	3 (2.01%)	27 (18.12%)



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 Urinary system anomalies irrespective of first or second trimester biochemical screening result.