

Efficacy and clinical utility of syndrome-oriented interpretation of abnormal fetal ultrasound findings: ScanOFe – An AI- assisted decision support tool

Prashant Acharya, Foram Prashant Acharya

Objectives To evaluate the clinical utility of an artificial intelligence (AI)–assisted decision support tool “ScanOFe” (An AI- assisted decision support tool) designed to correlate abnormal fetal ultrasound findings with congenital malformations, genetic syndromes, and associated genes in routine fetal medicine practice. ScanOFe is designed for fetal ultrasound phenotyping, incorporating bidirectional logic that allows both phenotype-to-syndrome and syndrome-to-phenotype analysis

Method: A total of 12,545 ultrasound examinations, between January 2020 and September 2025, were done with 640 fetal ultrasound abnormal findings were identified. Cohort was comprising 7,860 evaluated in the second trimester and 4,685 evaluated in the first trimester.

Results The correct syndromic diagnosis was identified by the AI system within the top 1 suggestion in 71% of cases and within the top 5 in 86%. Gene-level concordance was observed in from 56% of cases. Diagnostic performance was higher in fetuses with multiple anomalies compared with isolated anomalies. The system frequently suggested additional targeted ultrasound findings and optimized genetic testing strategies and higher additional investigations.

Conclusion: An AI-assisted bidirectional decision support tool ScanOFe DST demonstrates meaningful clinical utility in improving syndromic recognition and guiding genetic evaluation in fetuses with abnormal ultrasound findings. Such tools may augment clinical decision-making and support precision prenatal diagnosis in daily fetal medicine practice.

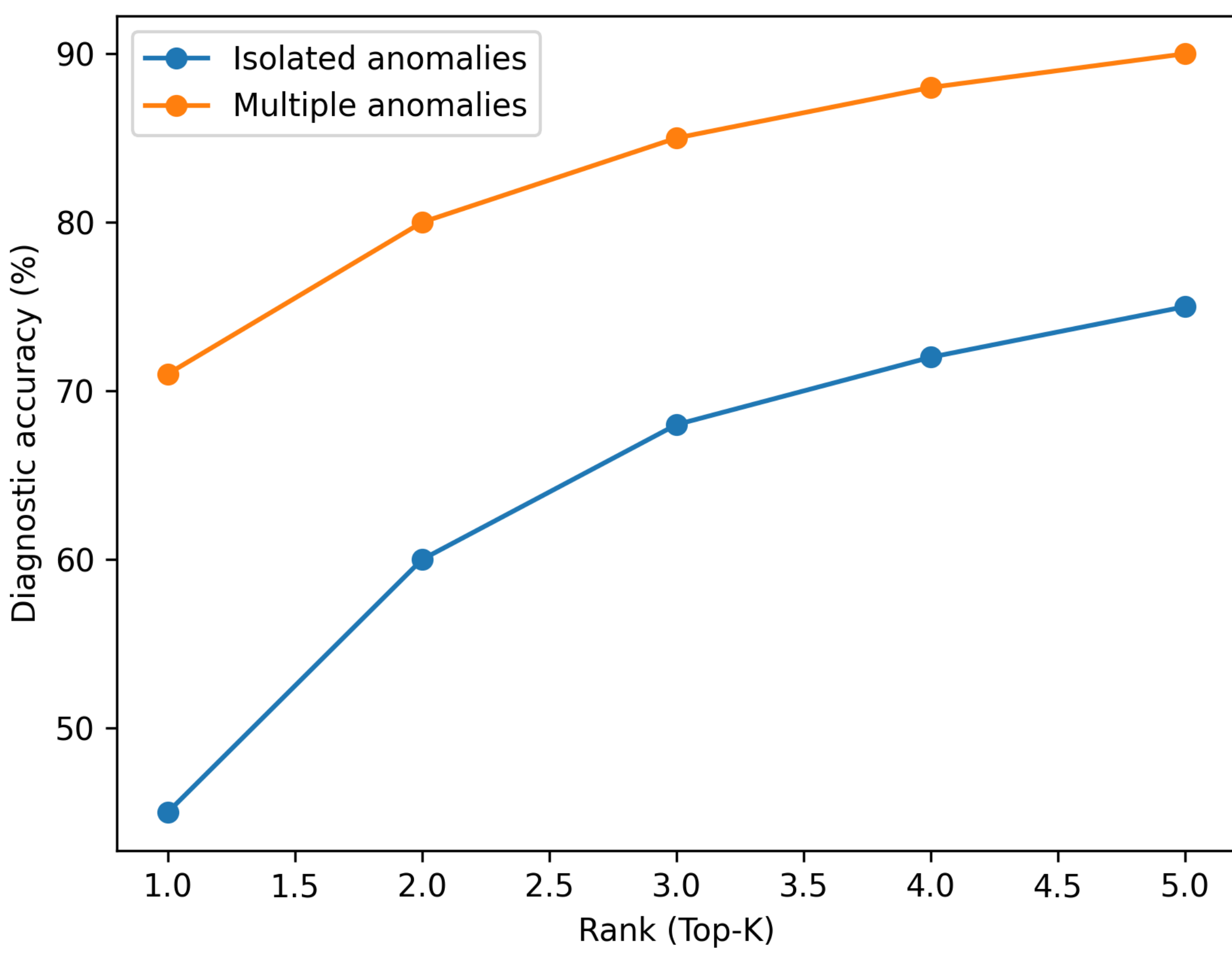


Fig 1 Top-K in isolated and multiple anomalies

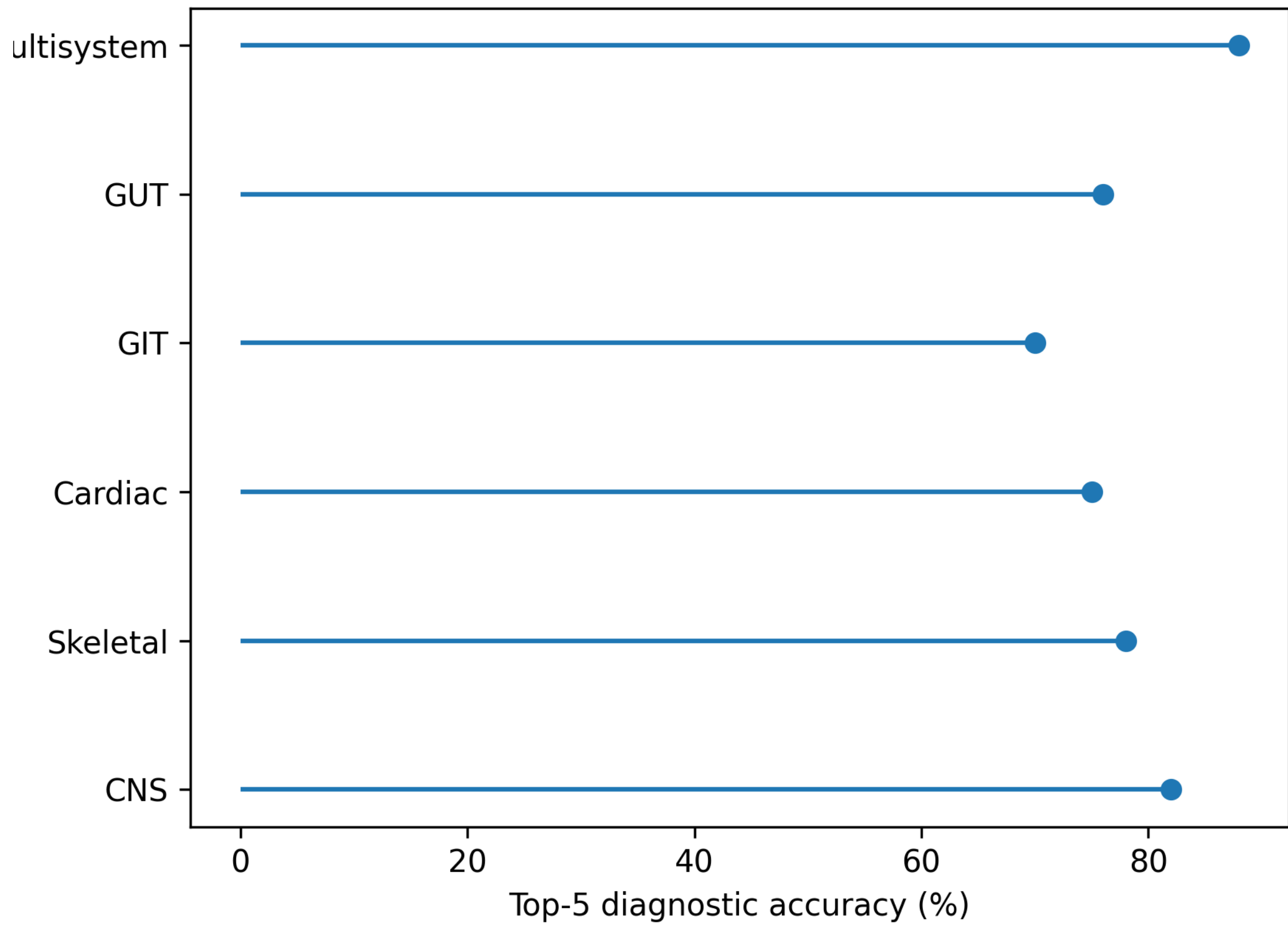


Fig 2: Forest graph for top5 diagnostic accuracy -system wise

Fetal findings	Fetal System involved	Appropriateness of suggested genetic testing strategies	additional USG subsequently confirmed	Performance differences between single and multiple anomaly cases in %	Syndromic association	autopsy /birth confirmation	Diagnostic accuracy	Gene-level concordance
Increased NT	Fetal Aneuploidy	100%	28%	70%	32%	85%	96%	88%
Absent Nasal bone	Fetal Aneuploidy	100%	32%	75%	38%	82%	94%	78%
Absent Corpus callosum	CNS anomalies	84%	38%	82%	26%	79%	79%	56%
ventriculomegaly	CNS anomalies	82%	42%	70%	32%	75%	84%	54%
Echogenic kidney	Skeletal anomalies	74%	46%	32%	20%	78%	89%	46%
Increased IT	CNS Anomalies	12%	12%	60%	2%	79%	94%	24%
Absent stomach	GIT anomalies	25%	18%	45%	18%	65%	93%	15%
Short femur	CNS anomalies	24%	38%	35%	10%	66%	93%	52%
Posterior fossa cyst	CNS anomalies	40%	45%	66%	25%	82%	90%	74%
Short humrus	Skeletal anomalies	25%	38%	33%	8%	77%	84%	52%
Cleft Lip and palate	GIT anomalies	56%	33%	45%	26%	81%	85%	66%
Absent Radius	Skeletal anomalies	32%	19%	21%	20%	78%	87%	32%
Club foot	Skeletal anomalies	39%	24%	10%	10%	62%	84%	54%
Ambiguous Genitalia	GUT anomalies	68%	32%	22%	25%	86%	94%	58%
Polyhydramnios	multifactorial	33%	53%	52%	15%	68%	20%	53&