Role for pre-procedure genetic counseling in pregnancies interrupted for fetal anomalies

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Background

- Birth defects are identified in 3-5% of pregnancies.
- There has been an increase in access to complex genetic testing methods especially in the case of ultrasound or genetic screening abnormalities

Objective

- **Evaluate** the association between genetic counseling and diagnostic genetic testing
- **Identify** types of fetal anomalies most likely to receive genetic testing
- **Examine** yield of genetic testing per type of fetal anomaly

Study Design

- Retrospective cohort study
- Inclusion: Pregnancies referred for termination with suspected structural or genetic fetal anomalies over a 4 year period
- Variables collected: demographics, genetic screening results, diagnostic testing results, ultrasound findings
- Patients were identified as having a primary genetic abnormality (abnormal serum analytes or NIPT) or a primary structural abnormality (as identified on ultrasound)
- Analysis: chi squared, fischer exact, multivariate logistic regression

Results

From 2016-2020, 400 pregnancies identified (55% genetic, 45% isolated structural)

55% of all pregnancies with anomalies received genetic counseling

Patients who received genetic counseling were 2 times more likely to get diagnostic testing (aOR 2.21 [1.25-3.90] 88% vs. 74%, p< 0.001)

Pregnancies with primary genetic conditions were more likely to get diagnostic genetic testing compared to those with primary structural anomalies (92% vs. 82%, p= 0.016)

Isolated structural anomalies had low yield of karyotype (7%) and microarray (10%)

Genetic counseling prior to termination of pregnancy is associated with **higher rates** of **diagnostic testing** and **should be offered** to all pregnancies with fetal anomalies.

Questions?
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Table 1. Genetic Testing by Type of Anomaly

	n	Any diagnostic test performed	Abnormal diagnostic test	Abnormal karyotype	Abnormal microarray
Chromosomal	199	182/199 (92%)	168/182 (92%)	148/165 (90%)	31/33 (94%)
Multisystem	59	43/59 (73%)	5/43 (12%)	2/30 (7%)	3/24 (13%)
Neurologic	51	35/51 (69%)	3/35 (8%)	3/25 (12%)	0/12 (0%)
Cardiac	28	17/28 (61%)	2/17 (12%)	1/11 (9%)	1/9 (11%)
Skeletal	24	20/24 (83%)	1/20 (5%)	0/14 (0%)	1/11 (9%)
Genitourinary	22	17/22 (77%)	3/17 (18%)	1/4 (25%)	2/8 (25%)
Facial	6	6/6 (100%)	0/6 (0%)	0/6 (0%)	0/1 (0%)
Chest	5	3/5 (60%)	0/3 (0%)	0/2 (0%)	0/2 (0%)
Gastrointestinal	4	4/4 (100%)	0/4 (0%)	0/4 (0%)	0/3 (0%)

Table 2. Adjusted Odds Ratio for Receiving Diagnostic Testing with Karyotype and/or Microarray

	Diagnostic testing performed n=327 (%)	No diagnostic testing performed n=73 (%)	OR (95% CI)	aOR (95% CI)
Genetic counseling				
Yes	196 (60.0)	26 (35.6)		
No	131 (40.0)	47 (64.4)	**2.70 (1.60-4.58)**	**2.21 (1.25-3.90)**
Type of anomaly				
Chromosomal	182 (55.7)	17 (23.3)	Referent	Referent
Multisystem	43 (13.2)	16 (21.9)	**0.25 (0.12-0.54)**	**0.26 (0.11-0.59)**
Neurologic	35 (10.7)	16 (21.9)	**0.20 (0.09-0.44)**	**0.24 (0.10-0.57)**
Cardiac	17 (5.2)	11 (15.1)	**0.14 (0.06-0.36)**	**0.18 (0.07-0.48)**
Skeletal	20 (6.1)	4 (5.5)	0.47 (0.14-1.52)	0.52 (0.15-1.80)
Genitourinary	17 (5.2)	5 (6.9)	**0.32 (0.10-0.97)**	0.32 (0.09-1.03)
Facial	6 (1.8)	2 (2.7)	0.28 (0.05-1.50)	0.40 (0.07-2.29)
Chest	3 (0.9)	2 (2.7)	**0.14 (0.22-0.90)**	**0.10 (0.01-0.69)**
Gastrointestinal	4 (1.2)	0 (0)	---	---
Isolated structural abnormality				
Yes	267 (81.7)	68 (93.2)		
No	60 (18.4)	5 (6.8)	**0.32 (0.12-0.85)**	0.79 (0.27-2.35)

Bold identifies statistical significance

Conclusion

- Genetic counseling should be offered to all presenting for termination for anomalies
- Workup for isolated anomalies should move beyond karyotype and microarray due to low yield of abnormal results and panels for single-gene disorders or exome sequencing may be considered