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 genetic condition.

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.

Table 1. Genetic Testing by Type of Anomaly

<table>
<thead>
<tr>
<th>Type of Anomaly</th>
<th>Genetic Counsel</th>
<th>Chromosomal</th>
<th>Multisystem</th>
<th>Neurologic</th>
<th>Cardiac</th>
<th>Skeletal</th>
<th>Genitourinary</th>
<th>Facial</th>
<th>Chest</th>
<th>Gastrointestinal</th>
</tr>
</thead>
<tbody>
<tr>
<td>Yes</td>
<td>116/150 (78%)</td>
<td>34/36 (94%)</td>
<td>7/13 (54%)</td>
<td>2/14 (14%)</td>
<td>2/11 (18%)</td>
<td>7/11 (64%)</td>
<td>4/8 (50%)</td>
<td>5/9 (56%)</td>
<td>4/4 (100%)</td>
<td></td>
</tr>
<tr>
<td>No</td>
<td>131/250 (53%)</td>
<td>26/41 (66%)</td>
<td>12/23 (52%)</td>
<td>3/14 (21%)</td>
<td>4/15 (27%)</td>
<td>8/15 (53%)</td>
<td>6/11 (55%)</td>
<td>2/6 (33%)</td>
<td>0/3 (0%)</td>
<td></td>
</tr>
</tbody>
</table>

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

<table>
<thead>
<tr>
<th>Genetic Counseling</th>
<th>Diagnostic testing performed on 327%</th>
<th>No diagnostic testing performed on 72%</th>
<th>OR (95% CI)</th>
<th>aOR (95% CI)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Yes</td>
<td>2.70 (1.06-5.36)</td>
<td>2.31 (1.05-5.03)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>No</td>
<td>1.00 (0.50-2.00)</td>
<td>1.00 (0.50-2.00)</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

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.