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Evolving trend in patient decision-making on non-invasive screening versus invasive testing following a prenatal diagnosis of ultrasound anomaly

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I. Introduction and Purpose

The advent of non-invasive cell-free DNA screening (cfDNA) has impacted prenatal patient care since its debut in 2011. This study includes patients referred by physicians for genetic counseling at Integrated Genetics due to an ultrasound anomaly and evaluated their subsequent choice regarding cfDNA and invasive prenatal diagnostic testing. We evaluated 2108 patients over the course of six years, from January 1, 2011 to December 31, 2016. Patients in this study were less than 24 weeks gestation with a noted ultrasound anomaly and included only those offered both cfDNA and invasive testing options as directed by physician request. No distinction was made between those patients with soft markers versus hard findings, or those with one ultrasound anomaly versus multiple anomalies.

II. Findings

Overall, 20.5% of patients chose cfDNA, 46.7% of patients elected invasive testing, and 9.3% of patients declined any follow up testing. 23.6% of patients had both cfDNA and invasive testing. In 2011, only 21 patients were available for analysis indicating that cfDNA was not a readily available option for patients at that time. These 21 patients were excluded from statistical analysis for this reason; however, their pattern of test election displays the same trend seen in other years of the study. The difference in the rates of acceptance for both cfDNA and invasive testing were statistically significant when comparing the earliest data, 2012, to the most recent data, 2016. Comparing choice trends through each year of the study as in Table 1 and Figure 1, there was a statistically significant difference in patient decision-making toward cfDNA testing and away from invasive procedures ($p<0.0001$). The percentage of patients declining all testing decreased markedly from 2012-2016 ($p<0.0001$).

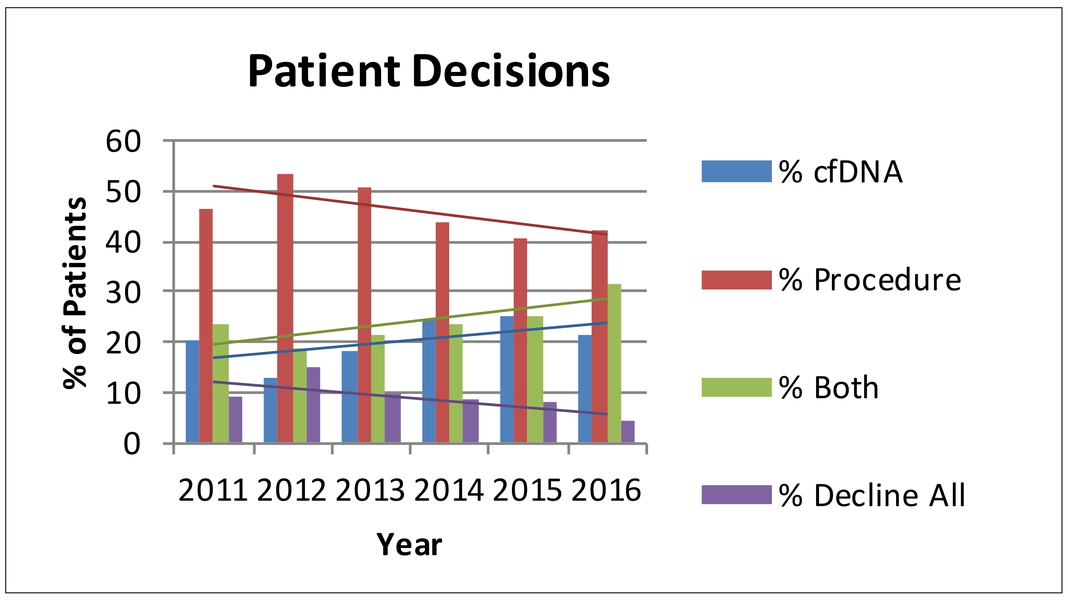

Figure 1. Comparison of the percentages and trendlines of patient testing decisions over the years 2011-2016

	Patients (n)	% cfDNA	% Procedure	% Both	% Decline All
2011	21	20.49	46.68	23.58	9.25
2012	366	13.11	53.28	18.58	15.03
2013	554	18.41	50.72	21.30	9.57
2014	498	24.50	43.57	23.49	8.43
2015	358	25.42	40.78	25.42	8.38
2016	311	21.54	41.12	31.83	4.50

Table 1. Distribution of patient testing decisions over the years 2011-2016.

III. Discussion

Over the years, the data show both an increase in the percentage of patients choosing cfDNA versus invasive testing and a marked decrease in the percentage of patients declining all testing (p-values <0.0001). These trends may be due to the greater availability and understanding of cfDNA testing as a non-invasive option for those patients interested in a certain level of information but not wanting to assume any additional risk in their pregnancies with invasive procedures such as CVS and amniocentesis.

Of note, 2016 data indicates a decrease in patient decisions for cfDNA accompanied by a mild increase in patient decisions for invasive procedures. The percentage of patients electing to do both testing methodologies is also the highest of all the years. One explanation may be ACOG's Committee Opinion 162 on fetal aneuploidy screening in May 2016 stating that some women "may prefer to have cell-free DNA screening rather than undergo definitive testing" but that "this approach may delay definitive diagnosis and management and may fail to identify some fetuses with aneuploidy". In addition, cfDNA will not provide essential information such as the distinction between a nondisjunction trisomy and a translocation trisomy.

A significant proportion of patients elected both testing methodologies, either on the same date of service or in subsequent visits. Despite our study not specifically addressing the motivations of patients who elected both testing options, we propose a variety of reasons for this: the patient's cfDNA was positive; the patient's anxiety remained high even after a negative cfDNA result; additional ultrasound findings were noted later in pregnancy.

IV. Conclusion

The trend away from invasive, diagnostic procedures for ultrasound anomalies is evident in the patient population analyzed in this study. When presented with the testing options of cfDNA and invasive prenatal diagnostic testing, cfDNA may be seen by patients as a preferable option in the genetic evaluation of an ultrasound anomaly. Care must be taken to ensure that patients are aware of the benefits and limitations of cfDNA when compared to invasive testing.

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