Maternal weight – impact on noninvasive prenatal testing (NIPT)

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INTRODUCTION

As the use of circulating cell-free DNA (cfDNA) in noninvasive prenatal testing (NIPT) has become a standard of care in the detection of fetal aneuploidy, it is increasingly important to understand factors that impact the ability to obtain a clinical result. Maternal weight has an inverse relationship on fetal fraction, potentially reducing the likelihood of obtaining an NIPT result. The American College of Obstetricians and Gynecologists (ACOG) states that more than one half of pregnant women are overweight or obese and 8% of reproductive-aged women are extremely obese, making maternal weight an important consideration in any prenatal testing. This study reviews the success rate of obtaining an NIPT result as a function of maternal weight.

METHODS

A retrospective analysis was performed on 172,131 maternal blood samples that were submitted to Sequenom Laboratories for MaterniT® 21PLUS laboratory developed testing between August 2014 and October 2015. Samples were subjected to DNA extraction, library preparation, and whole genome massively parallel sequencing as described by Jensen et al.[1] Sequencing data were analyzed using a novel algorithm to detect trisomies and select microdeletions.[2]

RESULTS

In this clinical cohort samples were stratified by maternal weight into 25 lb increments. Maternal weight was used instead of maternal BMI due to the fact that most providers and patients use this metric. The percent of NIPT samples that yielded a not reportable test result due to insufficient fetal fraction was evaluated. As expected the likelihood of receiving a successful result decreases as maternal weight increases with the lowest success rate in the >300 lbs population at 92.7%. For women >200 lbs, 14.8% of the population, the MaterniT® 21PLUS success rate is 96.6%.

CONCLUSIONS

Obese pregnant patients present a clinical challenge for prenatal procedures. Ultrasound is more complex and detection of structural abnormalities suggestive of genetic conditions can be difficult. Furthermore, in obese patients, it is technically challenging to perform invasive testing, and the risks of miscarriage are markedly increased[3]. In this study we show that despite slightly reduced success rates at extreme maternal weights, cell free DNA testing still delivers results for more than 92% of the patients even in the highest maternal weight category. Consequently, NIPT can be considered a viable option for aneuploidy screening in obese patients.

RESULTS

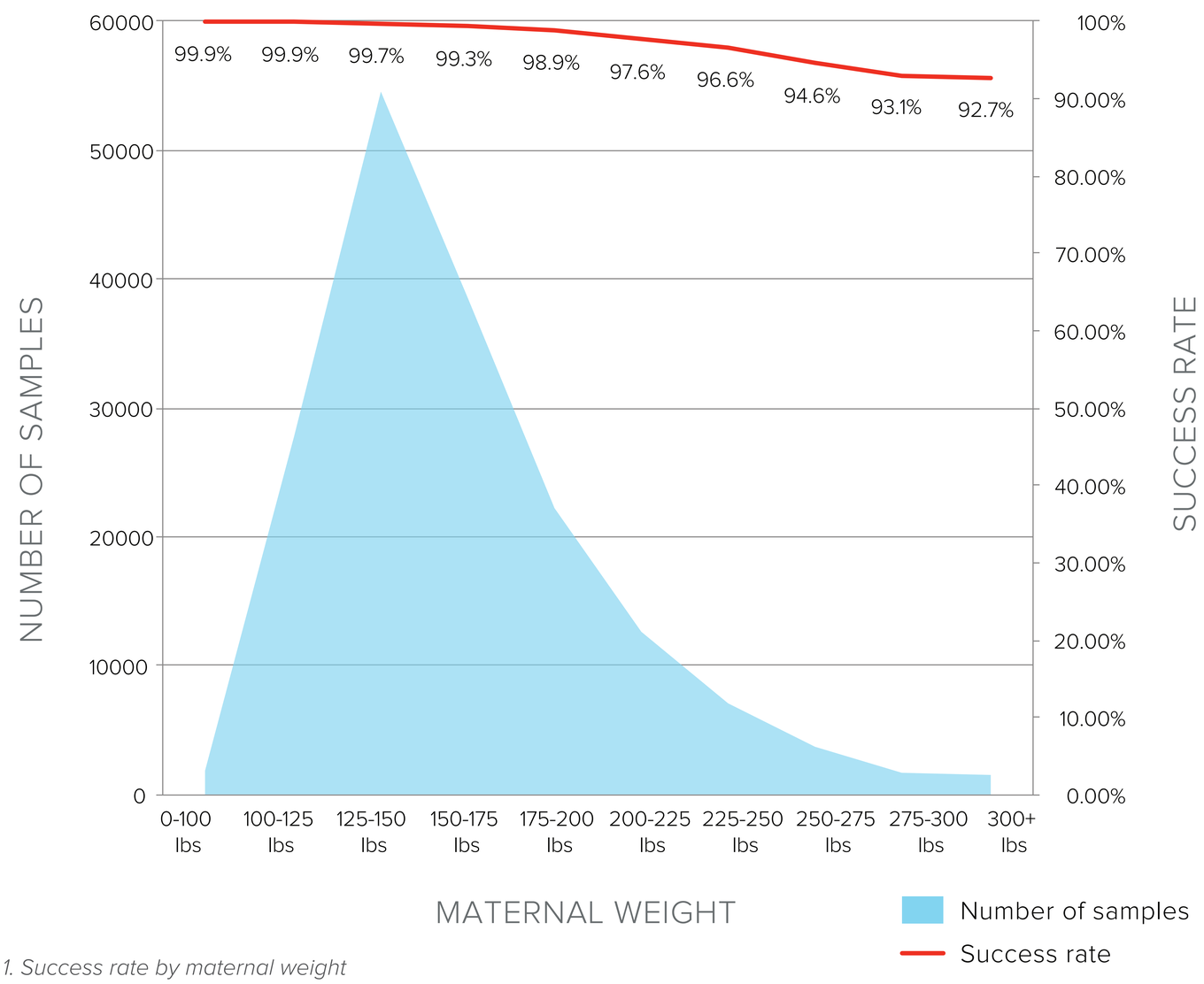

Figure 1. Success rate by maternal weight

NUMBER OF SAMPLES	SUCCESS RATE	WEIGHT
1960	99.9%	0-100 lbs
27787	99.9%	100-125 lbs
54651	99.7%	125-150 lbs
38546	99.3%	150-175 lbs
22310	98.9%	175-200 lbs
12635	97.6%	200-225 lbs
7100	96.6%	225-250 lbs
3727	94.6%	250-275 lbs
1794	93.1%	275-300 lbs
1621	92.7%	300 lbs+
Overall (172131)	99.0%	

Table 1. Success rate by maternal weight

REFERENCES

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