

The New Generation Non Invasive Prenatal Test

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There is a great need for the development of new generation safe, dynamic, highly accurate, cost effective technologies, which can facilitate the widespread adoption of Non-Invasive Prenatal Testing (NIPT). We hereby present a novel, safe and cost effective assay, called VERACITY, of diagnostic level accuracy for the non-invasive prenatal detection of genetic disorders which overcomes the limitations of current technologies. VERACITY enables the targeted analysis of selected genomic regions at very high sequencing depth, allows extremely high accurate fetal fraction determination and ensures extremely accurate detection of different types of genetic abnormalities located anywhere in the human genome¹. The analytical performance of the VERACITY assay was evaluated in two blind validation studies exhibiting 100% sensitivity and 100% specificity and correctly classified all trisomy 13, 18, 21 as well as aneuploidies of X and Y chromosomes. The performance of VERACITY was also assessed in a third blind validation study which consisted of 100 twin pregnancies of at least 10 weeks of gestation, including two trisomy 21 cases, one trisomy 18 case and one trisomy 13 case. Using a special fetal fraction estimation algorithm for dichorionic twins and an optimized dichorionic-twin specific aneuploidy detection algorithm we classified correctly all cases with 100% sensitivity and specificity. Further results from our studies also illustrate the feasibility of our novel targeted technology for the detection of common and rare genetic disorders in cell free DNA from maternal plasma such as deletions, duplications and point mutations. The clinical impact of cell free fetal DNA testing has been significant as indicated by its quick adoption in prenatal care. VERACITY overcomes limitations of current technologies and enables safe, diagnostic level accuracy and cost-effective non-invasive fetal aneuploidy detection of genetic disorders, which is critical for wide-spread adoption of NIPT.