Potential of cfDNA aneuploidy screening to reduce invasive prenatal testing

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Replacing conventional multi-marker Down syndrome screening protocols by universal cfDNA screening would increase the detection rate and substantially reduce the invasive prenatal test rate. For the public health perspective, the latter is more important than the former, for financial reasons. However, in practice the magnitude of this reduction is smaller than anticipated. Firstly, conventional screening includes all common autosomal trisomies, not just Down syndrome, and cfDNA screening now includes these disorders. Secondly, in the last decade there has been considerable 'scope creep' whereby the cfDNA test can now include sex chromosomal aneuploidies, microdeletion syndromes, rare autosomal trisomies and segmental aneuploidies. All of these together, could potentially the magnitude of the anticipated reduction by at least 5-fold.