

Comment on “The price of abandoning diagnostics testing for cell-free fetal DNA screening”

We read with great interest the commentary by Evans et al,¹ which offers economic modeling for varying prenatal screening and diagnostic tests. The authors use a managed care cost of \$1 million for each child with Down syndrome, an estimation they say that “has been well vetted.” They cite a study by Angelis et al²; however, this manuscript offers no calculations for people with Down syndrome.

In our recent publication, we estimated the out-of-pocket medical costs and third-party healthcare costs for children with Down syndrome³ using data from the OptumHealth database, which contains medical claims for over 18 million individuals, including primary subscribers and their covered beneficiaries. Cost difference or “incremental costs” were defined as the healthcare costs for people with Down syndrome above and beyond that of matched controls; whereas “total costs” included these incremental costs plus those of the matched controls. Inpatient, outpatient, emergency room, home health agency, and pharmacy costs were included. Third-party payers incurred *incremental* costs of \$230,043 for each person with Down syndrome over the first 18 years; their *total* costs were \$264,489 per person. The *incremental* out-of-pocket costs incurred by caregivers were \$18,248 over the first 18 years, or about an additional \$84 per month when amortized over that time. Caregivers' *total* out-of-pocket expenses were \$23,624 per person for the first 18 years. When combined, the *total* healthcare costs for children with Down syndrome over the first 18 years would be \$288,113, at most, by our estimations.

We respectfully recommend that Evans et al consider revising their economic modeling to incorporate these more current numbers for people with Down syndrome. The authors further make an “educated guess” that the medical costs for children with microdeletions and microduplication syndromes would be 50% smaller than that of children with Down syndrome. If this were to be true, these estimations would benefit from concomitant revision. The authors' central claim is that universal offering of amniocentesis/chorionic villus sampling with a microarray would result in substantial healthcare savings, compared to universal offering of cfDNA, if a sufficient number of expectant couples elected to terminate fetuses with genetic conditions. While the comparative conclusion would remain unchanged, the quantity of savings would be sizably smaller if our estimations were incorporated.

CONFLICT OF INTEREST

B.G.S. serves in a non-paid capacity on the Medical and Science Advisory Board for the Massachusetts Down Syndrome Congress and the Board of Directors for Band of Angels Foundation, both non-profit

organizations. He further serves on the Medical Advisory Board for the non-profit National Center for Prenatal and Postnatal Down Syndrome Diagnoses Resources. B.G.S. occasionally gets remunerated from Down syndrome non-profit organizations for speaking engagements about Down syndrome. He has a sister with Down syndrome. He receives research support for clinical drug trials involving patients with Down syndrome from Hoffmann-La Roche, Inc. He also receives annual royalties from Woodbine House, Inc., for the publication of his book, *Fasten Your Seatbelt: A Crash Course on Down Syndrome for Brothers and Sisters*. J.C. is a member of the Massachusetts Down Syndrome Congress and has a son with Down syndrome. A.K., D.S., M.H., and P.L. report no conflicts of interest. The authors have no financial conflict of interests related to the content of this letter.

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