Arch Gynecol Obstet (2015) 292:715–716
DOI 10.1007/s00404-015-3794-y

REPLY

Reply to the letter: "Comments on non-invasive prenatal testing"

Gwendolin Manegold-Brauer[1,2] · **Annegret Geipel**[1]

Published online: 20 June 2015

Dear Editor, Dr. Kim and Dr. Ryu,

Thank you for reading our original research article on the implementation of non-invasive prenatal testing (NIPT) in a high-risk collective [1]. Our study clearly demonstrated that the clinical implementation of NIPT is highly dependent on the presence or absence of ultrasound findings and does not necessarily reduce the number of invasive procedures in every setting. Most women who opted for NIPT did not have a high risk after first-trimester screening, but were mainly of advanced maternal age and the number of invasive procedures was not reduced in our study most likely since our unit has a high prevalence of patients presenting with fetal malformations.

The letter by Ryu et al. [2], however, does not discuss the findings of our study but gives general comments on the technology of NIPT. The authors mainly discuss the need for an invasive procedure to confirm the diagnosis of a positive NIPT result and give recommendations for indications for NIPT. We agree with most of the statements for the indications for NIPT which roughly are in accordance with the recommendations given by the International Society of Ultrasound in Obstetrics and Gynecology (ISUOG) and the American Society of Obstetrics and Gynecology (ACOG) [3, 4]. We might want to discuss two points, which we do not agree with. The authors suggest maternal serum-AFP in the second trimester, most likely for detection of neural tube defects. Maternal serum-AFP has a much lower detection rate for anencephaly and open spina bifida than ultrasound and today open spina bifida can reliably be detected in almost 95 % of the cases [5]. Therefore, we would recommend maternal serum-AFP only when prenatal ultrasound is not available. Secondly, the authors state that NIPT cannot be used to confirm a diagnosis due to its high positive predictive value (PPV). The contrary, however, is the case. The PPV for NIPT for the detection of trisomy 21 varies widely depending on the disease prevalence in the investigated collective. While the PPV was high in the initial studies stemming from high-risk populations, the PPV for the detection of trisomy 21 has been reported between 45 and 81 % in low-risk populations [6, 7]. Due to the severe consequences that can be drawn from a positive NIPT result and due to the low PPV in certain populations, NIPT is an advanced screening test but requires confirmation by invasive testing.

Conflict of interest The authors declare that there is no conflict of interest.

References

1. Manegold-Brauer G, Berg C, Flöck A, Rüland A, Gembruch U, Geipel A (2015) Uptake of non-invasive prenatal testing (NIPT) and impact on invasive procedures in a tertiary referral center. Arch Gynecol Obstet. doi:10.1007/s00404-015-3674-5
2. Ryu A, Kim TH (2015) Comments on non-invasive prenatal testing (NIPT). Arch Gynecol Obstet. doi:10.1007/s00404-015-3793-z
3. Salomon LJ, Alfirevic Z, Audibert F, Kagan KO, Paladini D, Yeo G et al (2014) ISUOG consensus statement on the impact of non-invasive prenatal testing (NIPT) on prenatal ultrasound practice. Ultrasound Obstet Gynecol 44(1):122–123

This reply refers to the comment available at doi:10.1007/s00404-015-3793-z.

✉ Gwendolin Manegold-Brauer
gwendolin.manegold-brauer@usb.ch

[1] Department of Obstetrics and Prenatal Medicine, University of Bonn, Bonn, Germany

[2] Department of Prenatal Medicine and Gynecologic Ultrasound, University of Basel, Basel, Switzerland

4. American College of Obstetricians and Gynecologists (2012) Noninvasive prenatal testing for fetal aneuploidy. Committee Opinion No. 545. Obstet Gynecol 120(6):1532–1534
5. Fleurke-Rozema JH, Vogel TA, Voskamp BJ, Pajkrt E, van den Berg PP, Beekhuis JR et al (2014) Impact of introduction of mid-trimester scan on pregnancy outcome of open spina bifida in The Netherlands. Ultrasound Obstet Gynecol 43(5):553–556
6. Bianchi DW, Parker RL, Wentworth J, Madankumar R, Saffer C, Das AF et al (2014) DNA sequencing versus standard prenatal aneuploidy screening. N Engl J Med 370(9):799–808
7. Zhang H, Gao Y, Jiang F, Fu M, Yuan Y, Guo Y et al (2015) Non-invasive prenatal testing for trisomies 21, 18 and 13: clinical experience from 146 958 pregnancies. Ultrasound Obstet Gynecol 45(5):530–538