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Authors' response

We extend our gratitude to the authors for their opinion¹ after meticulously reading our paper.² We fully agree with the authors regarding the inherent limitations of the qPCR method in diagnosing Turner syndrome (TS) and have acknowledged the same in our article. Rather than prenatal screening, we believe a qPCR based diagnostic test would better suit as a neonatal screening test and as a screening test for TS among all girls with short stature where universal karyotyping, though recommended by guidelines, is not pragmatic. Prenatal screening for TS has its own set of ethical concerns given that girls with TS can have almost near-normal lifespan with good quality of life. QF-PCR as rightly mentioned by the author is a good molecular cytogenetic method to diagnose aneuploidies and currently in use for prenatal detection, but QF-PCR availability is limited to high-end genetic laboratories and dedicated fertility clinics. qPCR on the other hand requires much lower setting up cost and maintenance

charge and many govt hospitals are already equipped with one/multiple qPCR machines specially after the COVID-19 pandemic. QF-PCR would lead to inconclusive results in isochromosome TS and deletion of Xp. So, using a qPCR based screening test for TS may be more cost-effective than other methods but definitely acknowledging the inherent limitations of the method like failing to detect structural variants. Further research is required in this field.

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