Jacobsen syndrome detected by noninvasive prenatal testing

Abstract

Background: Noninvasive prenatal testing has a high detection rate of common fetal chromosomal aneuploidies. However, detection of additional chromosome abnormalities has not been well described or validated.

Case: We report a case of Jacobsen syndrome, a congenital disorder involving deletion of chromosome 11q, detected by noninvasive prenatal testing at 14 weeks of gestation and confirmed on neonatal testing with array chromosomal genomic hybridization.

Conclusion: Noninvasive prenatal testing should be considered when multiple fetal anomalies are present and invasive testing is declined. As the clinical application of noninvasive prenatal testing continues to evolve, additional submicroscopic chromosomal information may be clinically helpful and should be confirmed with diagnostic testing until larger studies help further define the screening characteristics of noninvasive prenatal testing.

Lo JO, Feist CD, Hashima J, Shaffer BL. Jacobsen syndrome detected by noninvasive prenatal testing. Obstet Gynecol. 2015 Feb;125(2):387-389. doi: 10.1097/AOG.0000000000000528. PMID: 25569015.