Early intervention: Sequencing fetal DNA could identify genetic disorders

A new test allows doctors to diagnose genetic disorders in fetuses early in pregnancy by sequencing small amounts of fetal DNA in the mother's blood.

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[This test] looks at 30 genes associated with dominant genetic diseases, which occur when someone has just one copy of a faulty gene. These usually occur because of a mutation in the sperm, egg or embryo, and are more common when one of the parents is relatively old.
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If it gives a positive result for a genetic disorder, women can be offered a more invasive test to confirm the diagnosis. If that test is positive, couples would ideally be offered genetic counselling so they can make informed decisions about how to manage the pregnancy.

Jinglan Zhang at Baylor College of Medicine, Texas, and colleagues tested blood samples from 422 pregnant women, from nine weeks of gestation onwards, sent to them from clinics in the US, Europe and Asia. Some of them had abnormal results on ultrasound scans suggesting a skeletal disorder in the fetus. Thirty-two tested positive for mutations in one of the 30 genes in the test.

Read full, original post: Blood test can diagnose fetal genetic disorders early in pregnancy