



Prenatal Screening after Successful Mosaic Embryo Transfer: A Case Utilizing pcfDNA Analysis to Aid in Patient-decision Making

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PURPOSE & OBJECTIVES

- Over time, transfer of low-level mosaic embryos has increased (especially when no euploid embryos are available) and studies have demonstrated positive reproductive outcomes following the transfer of mosaic embryos screened via preimplantation genetic testing for aneuploidy (PGT-A). However, it has also been shown that the type and extent of mosaicism can impact outcomes.
- Post mosaic embryo implantation, diagnostic testing via amniocentesis is recommended over other prenatal aneuploidy screening or testing (i.e., prenatal cell-free DNA [pcfDNA]) for several reasons including: both pcfDNA and PGT-A screen placental DNA vs fetal DNA, and pcfDNA is not universally available for additional aneuploidy analysis outside of chromosomes 21, 18, 13, X, and Y.
- Patients with pregnancies achieved via *in vitro* fertilization often decline amniocentesis due to risk of complications and miscarriage.
- Selected laboratories offer aneuploidy analysis covering all chromosomes; pcfDNA may therefore be an option for patients post mosaic embryo transfer.
- This poster presents a compelling case in which pcfDNA screening and ultrasound were utilized for patient decision making regarding diagnostic testing post transfer of a known mosaic embryo.

CASE DESCRIPTION

- Patient was referred to genetic counseling post successful transfer of an embryo showing mosaic monosomy 3 on PGT-A.
- After counseling on screening vs diagnostic testing, patient elected non-invasive pcfDNA screening (Prequel® Prenatal Screen, Myriad Genetics), which includes analysis of rare autosomal aneuploidies.
- Sample for pcfDNA was collected at 12 weeks 0 days gestation.
- Notation of a known mosaic monosomy 3 embryo transfer on screen order triggered manual review of raw data specifically focusing on chromosome 3 (**Figure 1**).

RESULTS

- Laboratory directors observed:
 - no evidence of aneuploidy (monosomy or otherwise) involving chromosome 3, including no evidence of mosaicism
 - no evidence of possible partial involvement of chromosome 3
- Results returned screen-negative for all aneuploidies, including chromosome 3

Figure 1: Chromosome 3 Scatterplot

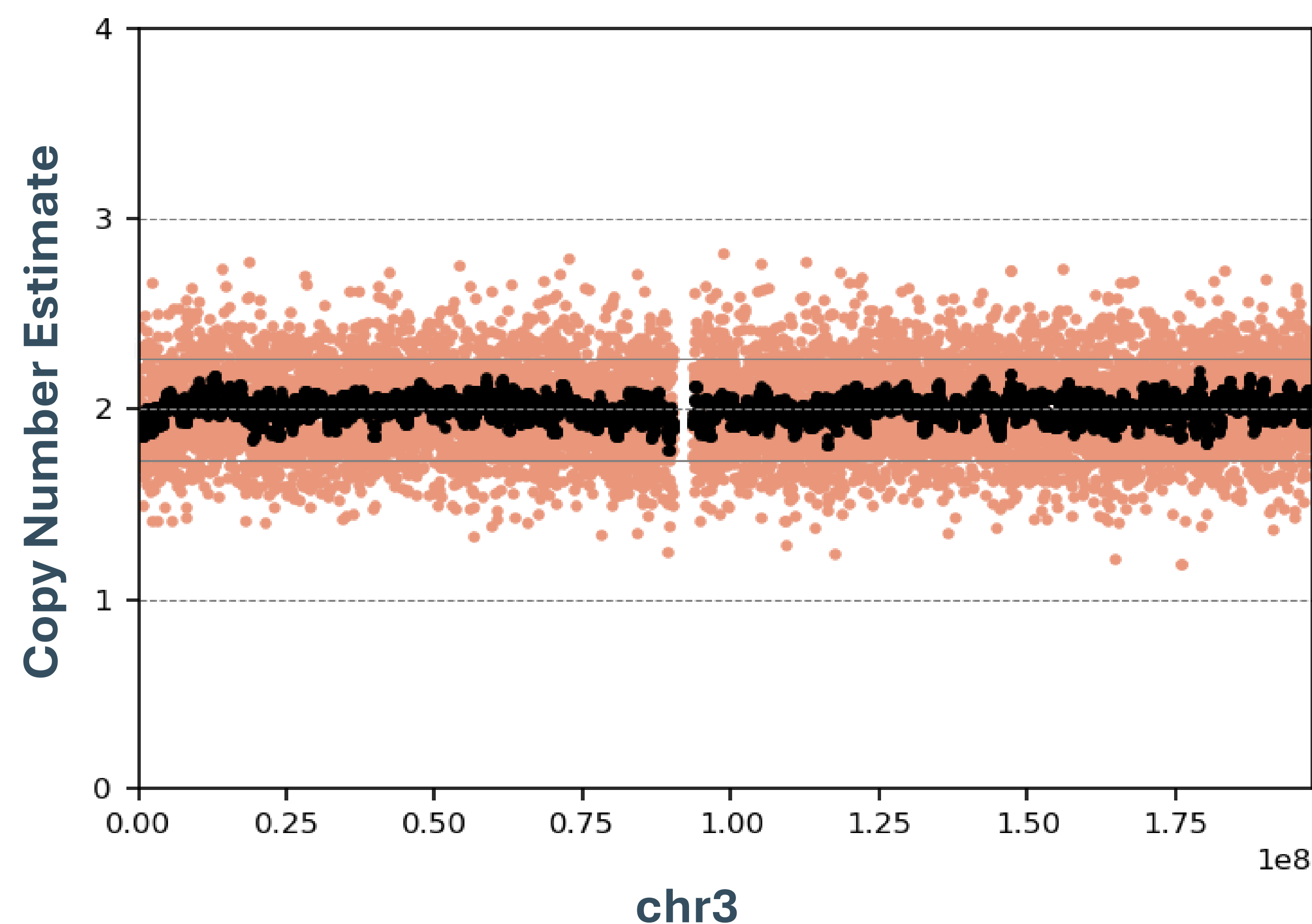


Figure 1: pcfDNA scatterplot specific for chromosome 3 was clean, unremarkable, and consistent with the low-risk result reported.

IMPACT STATEMENT

This case report demonstrates an opportunity for patients and providers to consider pcfDNA with expanded analysis in instances of mosaic embryo transfers.

MANAGEMENT OUTCOME

- Anatomy ultrasound was completed at 21 weeks 0 days and showed no evidence of fetal anomalies or placental complications.
- Due to negative pcfDNA results and an unremarkable ultrasound, patient declined amniocentesis. Neonatal karyotype was offered after delivery but is not planned at this time.

CONCLUSIONS

- Literature has shown that the vast majority of pregnancies following positive mosaic PGT-A findings will have normal diagnostic results.
- While diagnostic testing via amniocentesis is still the gold standard in instances of mosaic embryo transfers, due to the availability of expanded aneuploidy analysis, pcfDNA may be a viable option for patients who desire additional genetic insights but decline invasive diagnostic testing in pregnancy.

ACKNOWLEDGEMENTS

Disclosures: A.P., L.E., and M.M.M. were employees of Myriad Genetics, Inc. at the time of this study and received salaries and stock as compensation.

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