



## BACKGROUND

### Placental Masses

- Placental teratomas thought to arise from germ cells migrating into extraplacental membranes<sup>2</sup>
- Historically, placental teratomas were differentiated from acardiac amorphous twins by presence of multiple tissue types, but lack of axial organization and umbilical cord<sup>1</sup>
- New evidence - previously documented placental teratomas may instead represent acardiac amorphous twin given similar pattern of short tandem repeat (STR) polymorphic loci between placental masses (thought to be teratoma) and adjacent normal placental tissue<sup>1</sup>

### Trisomy 21 (T21)

- Most commonly due to spontaneous meiotic non-disjunction<sup>4</sup>
- 1-2% of cases from mitotic errors, causing mosaicism

## OBJECTIVES

Describe the genetic profile of a placental mass, initially characterized as a placental teratoma, occurring in association with a newborn with mosaic T21.

## CASE

### Pregnancy Course:

**Mother:** 21 yo G1PO, unremarkable medical and surgical history

### Prenatal Genetic Testing:

- 2<sup>nd</sup> Trimester Serum Marker Screen (1/92 risk of T21)
- Noninvasive prenatal screening (68.21% positive predictive value for T21)

**Fetal Anatomy Ultrasound:** bilateral ventriculomegaly, short humeri, VSD\*

**Delivery:** male infant, 35 wks 1 day gestational age (GA), cesarean section after failed induction of labor

### Infant

**Weight:** 1850 g

**APGAR:** 6 & 9

**Dysmorphology exam:** plagiocephaly, fine eyebrows, thin upper lip, smooth philtrum, deep vertical plantar creases, mild 5<sup>th</sup> finger clinodactyly, overriding 4<sup>th</sup> toe, grade II/VI murmur, hypospadias with chordee

**Postnatal Cytogenetics:** 3/22 (14%) cells T21

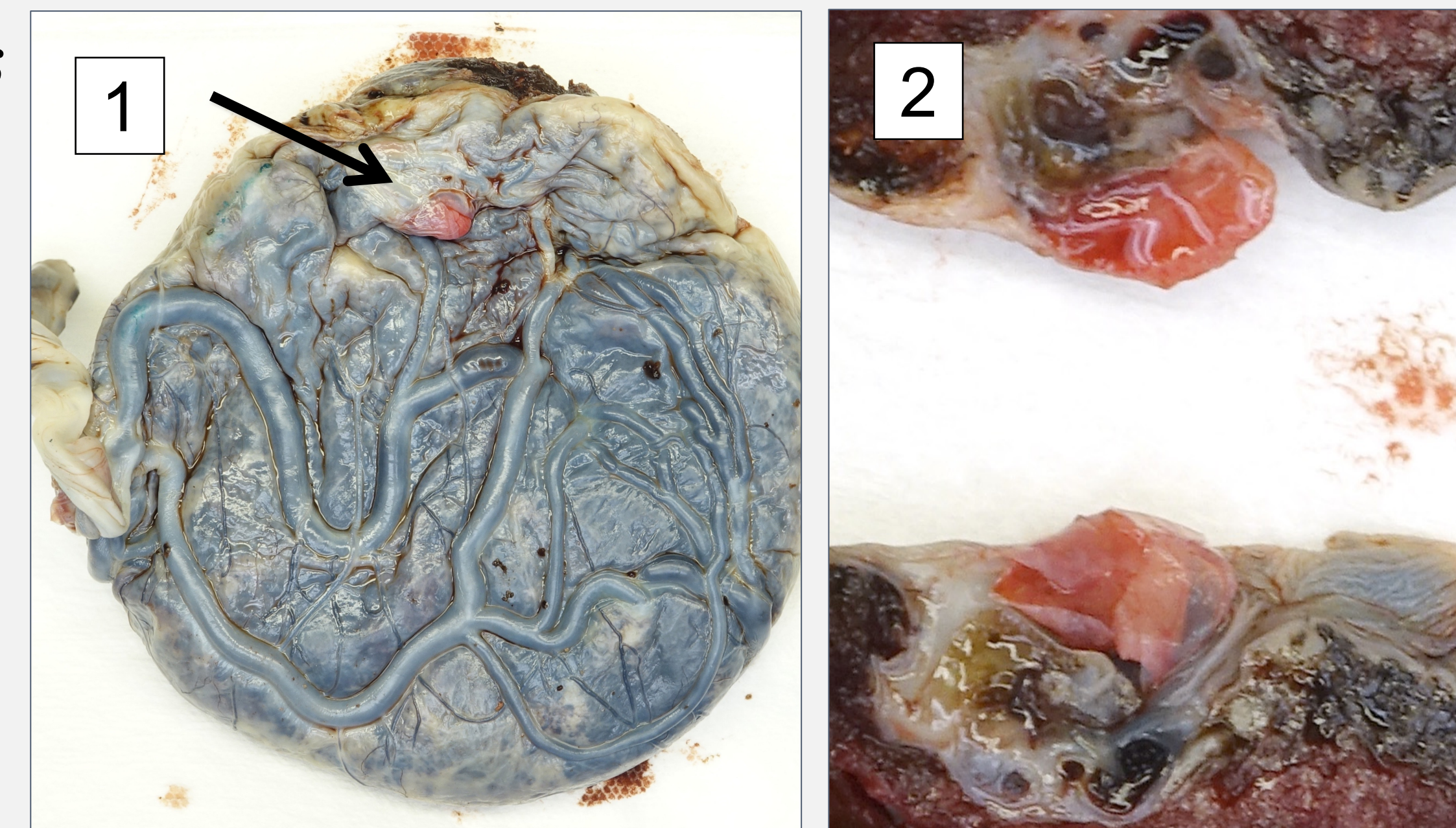
\*ventricular septal defect

## PLACENTA

### Placenta

- 286 g (<10<sup>th</sup> percentile for GA)
- **Mass:** 0.8 x 0.6 x 0.4 cm, between the amnion and chorion on the fetal surface (Image 1), initial diagnosis benign, non-trophoblastic mature teratoma

**Image 1:** Gross Appearance of Placental Mass, intact (1) and transected (2)



### STR (GenePrint 24) Analysis

- Placental mass had extra chromosome 19 allele compared to adjacent placental tissue
- Neither mass nor adjacent tissue had extra chromosome 21 alleles

**Table 1:** Whole Exome Sequencing of placental mass vs adjacent placental tissue

	Mass vs reference genome	Mass vs Adjacent Placental Tissue
All variants*	45758	159
Exonic variants*	15913	48

\*>20x coverage

## CONCLUSIONS

- Minimal difference in exome variants between placental mass and adjacent tissue suggests mass likely acardiac amorphous twin (Table 1)
- Explanations for discordant STR findings between placental mass and infant include:
  - Prezygotic meiosis II nondisjunction error caused T21 in the zygote. After twinning, amorphous twin regained normal chromosomal constitution following trisomy rescue.
  - Triplicate chromosome 19 in amorphous twin may represent a parental chromosome 19 duplication not detected on karyotyping or that corrected in infant.
- Our findings support conclusion of McHenry et al<sup>1</sup>

## REFERENCES

1. McHenry A, Morotti R, Hui P. Placenta Teratoma or Acardiac Fetus Amorphous: A Case Study by DNA Genotyping. *Int J Gynecol Pathol.* 2022;41(1):51-58. doi:10.1097/PGP.0000000000000769
2. Khormoji NN, Soleimani V, Sheikhhasani S. Mature Placental Teratoma: A Case Report. *Biomedical Research and Therapy.* 2022;9(1):4818-4821. doi:10.15419/bmrat.v9i1.718
3. Bull MJ. Down Syndrome. *N Engl J Med.* 2020 Jun 11;382(24):2344-2352. doi: 10.1056/NEJMr1706537. PMID: 32521135.