

# A BRCA1+ Patient with Twin Pregnancy of Complete Mole with Coexisting Fetus

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## BACKGROUND

### What is a molar pregnancy?

In a complete molar pregnancy, an egg without maternal DNA is fertilized by either one sperm whose genetic material is later duplicated, or two separate sperm, to form a diploid zygote.

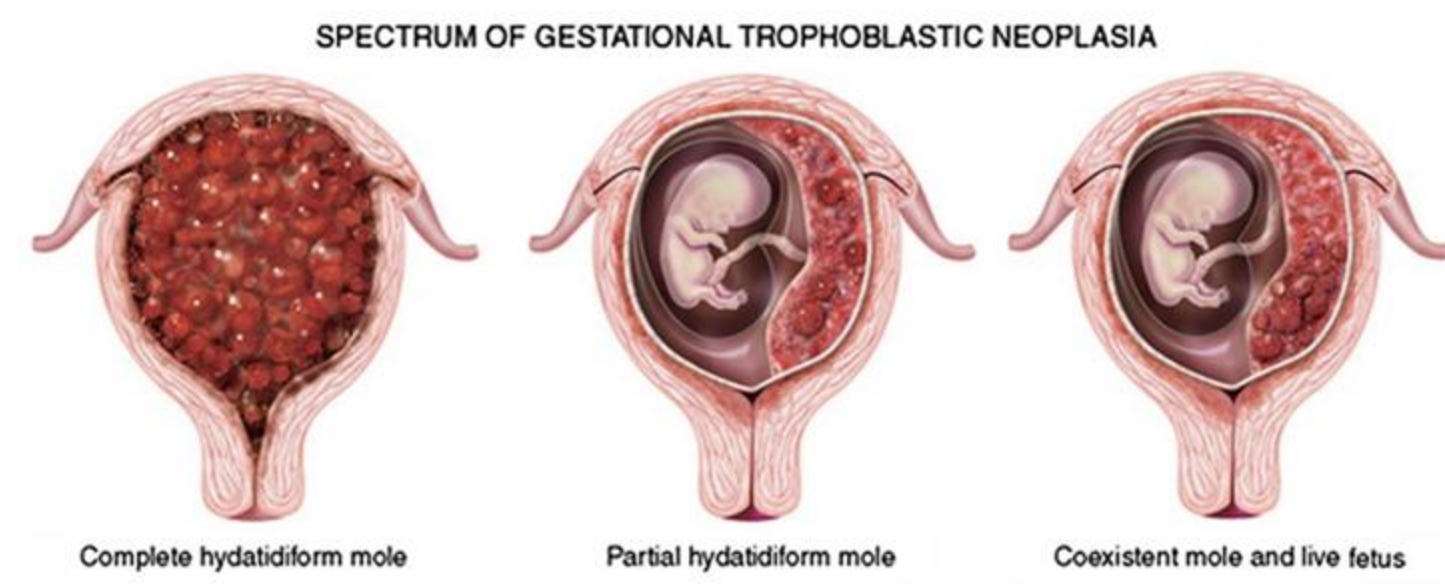


Figure 1. Schematic of complete mole, partial mole, and mole with viable fetus<sup>1</sup>

### What is a complete mole with a coexisting fetus (CMCF)?

A CMCF is a twin pregnancy where one "twin" is a complete mole and the other twin is a viable fetus. Twin pregnancy of a complete mole with coexisting fetus (CMCF) is a very rare occurrence with an incidence of 1/22,000 to 1/100,000 pregnancies.<sup>2</sup>

### What are the risks of a CMCF pregnancy?<sup>3</sup>

- Maternal
  - pre-eclampsia
  - hemorrhage
  - uterine rupture
  - gestational trophoblastic disease, a form of cancer
  - hyperthyroidism
- Fetal
  - intrauterine death
  - intrauterine growth restriction

### What is the significance of having BRCA1/2 mutation?

- BRCA1 mutation carries a ~60% lifetime risk of developing breast cancer and ~40% lifetime risk of ovarian cancer.<sup>4</sup>
- The incidence of BRCA1 mutation is 1/30,000<sup>4</sup>

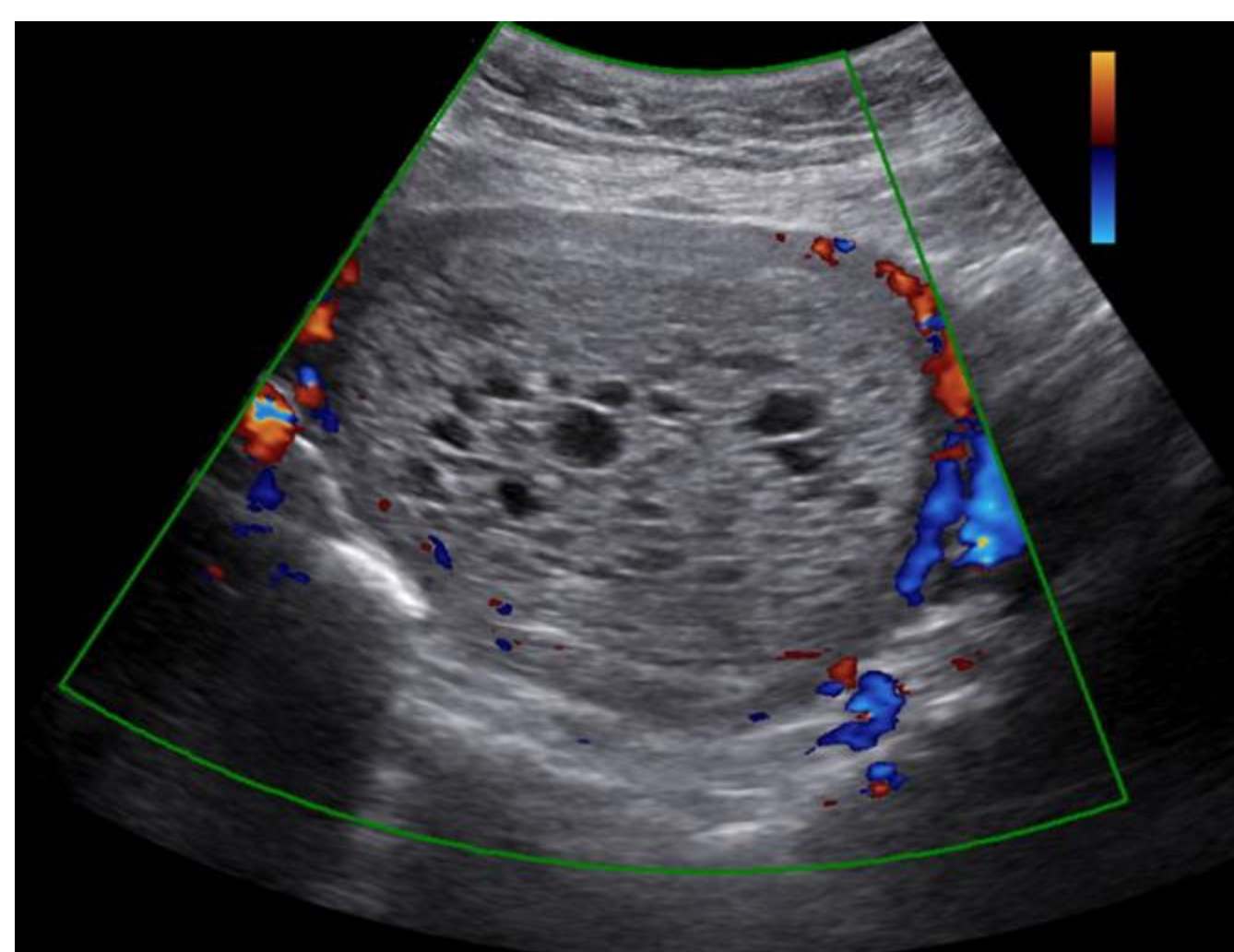


Figure 2. Ultrasound depicting the classic "snowstorm" or "cluster of grapes" appearance of molar pregnancy and decreased blood flow with color flow Doppler sonography.<sup>5</sup>

## CASE

### Diagnosis

- 35 yo G3P2 female presented with a 20-week OB scan suspicious for both a viable fetus and molar pregnancy
  - Initial 10-week US indeterminate on whether the mole was partial or complete
- PMHx significant only for BRCA1 mutation
  - Mother and sister diagnosed with ovarian cancer at young ages

### Treatment Options

- Pregnancy Termination
  - Patient desired this pregnancy but did not intend to pursue future pregnancy
  - Selective reduction via D&C would endanger the viable fetus
- Continuation of Pregnancy
  - Posed significant risks, including development of gestational trophoblastic neoplasia
- Ovarian Cancer Prophylaxis due to BRCA1 mutation
  - A prophylactic hysterectomy with bilateral oophorectomy increased the risk of surgical complications in an already high-risk patient

### Delivery

- 34 weeks: Planned Cesarean section and hysterectomy with bilateral oophorectomy
- After delivery, the patient experienced elevated blood pressures but did not develop pre-eclampsia

### Outcome

- Discharged post-op day 2 without maternal or fetal complications
- Serial  $\beta$ -HCG levels were monitored until undetectable
- Genetic analysis revealed the removal of a complete mole

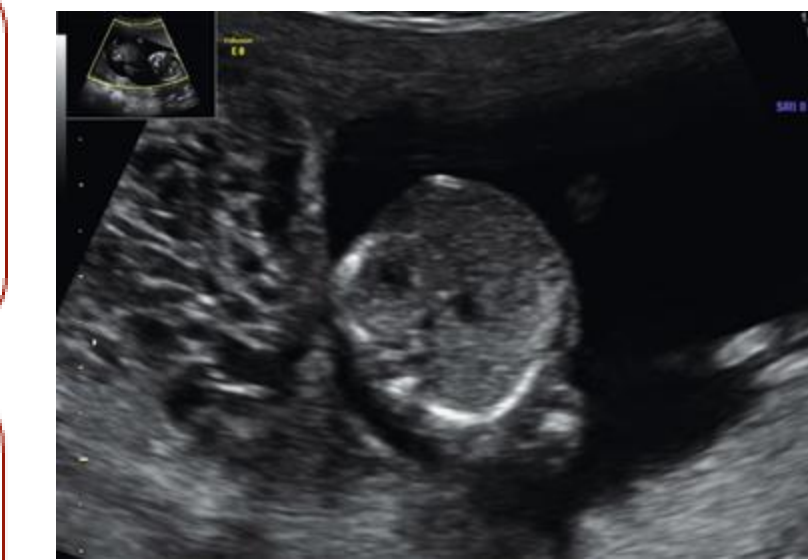


Figure 3. Ultrasound depicting a CMCF pregnancy. Molar tissue is seen on the left and the fetal abdomen is seen on the right. This image was taken from a previous case report of a CMCF pregnancy in a 33 year-old at 14 weeks gestation.<sup>2</sup>

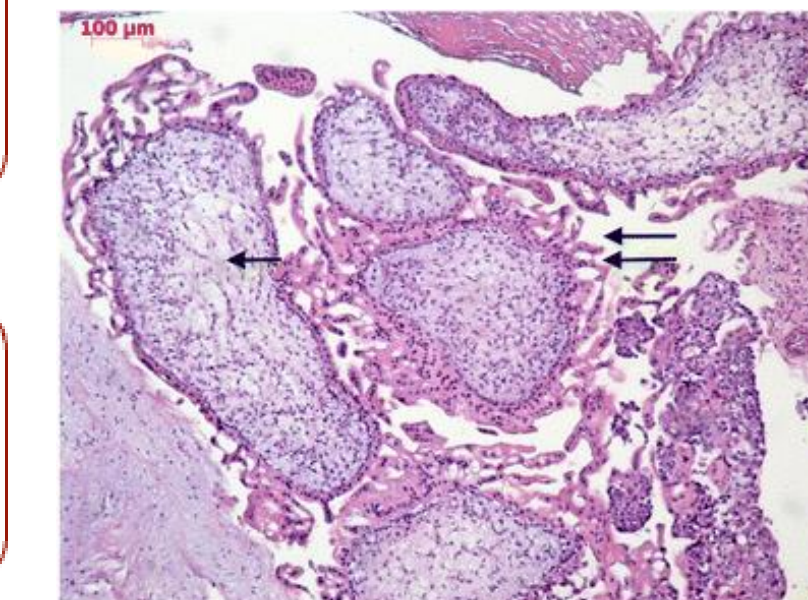


Figure 4. H & E Stain Showing Histology of Complete Mole<sup>6</sup>

## CLINICAL SIGNIFICANCE

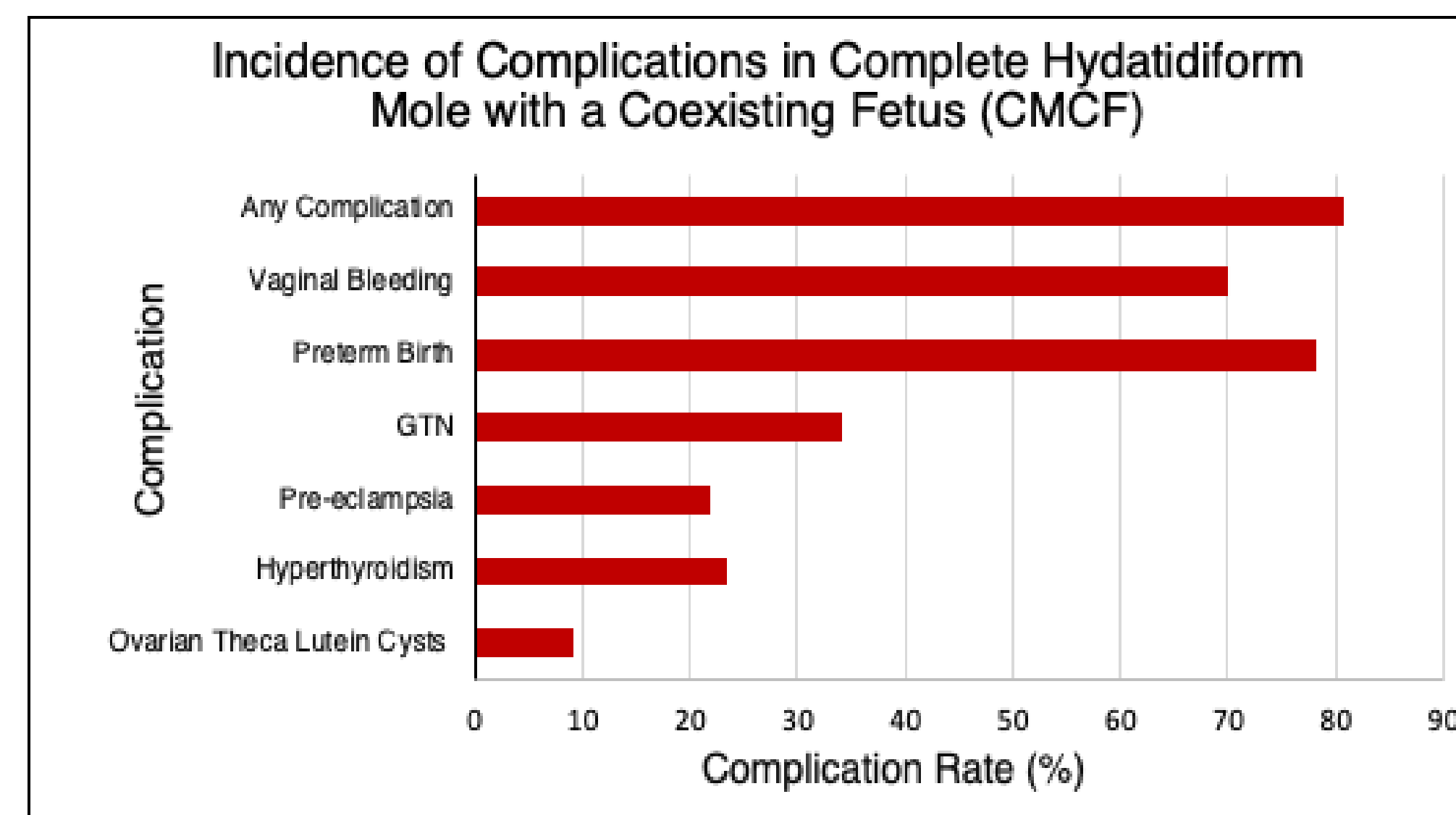


Figure 5. Incidence of Common Maternal Complications in Complete Hydatidiform Mole with a Coexisting Fetus.<sup>3</sup> This figure was adapted from a meta-analysis of 244 cases of CMCF pregnancy. The overall incidence of complications was 80.8%. Pregnancy complications included vaginal bleeding (70%), pre-eclampsia (21.9%), and hyperthyroidism (23.3%). Ovarian theca-lutein cysts were described in 9% of cases and gestational trophoblastic neoplasia (GTN) occurred in 34% of CMCF cases.

### Key Takeaways

1. Patients who choose to continue pregnancy should be thoroughly informed about the expansive and potentially severe complications
  - a. Overall incidence of complications is 80.8% in CMCF pregnancy<sup>3</sup>
1. Successful delivery without complication is, while rare, still possible
  - a. Chance of a live birth in CMCF pregnancy is below 50% in patients who do not undergo an elective abortion<sup>3</sup>
1. Patients with BRCA1 mutations require complex and ongoing care to mitigate the risks of developing ovarian/breast cancer
  - a. Although this patient's risk of developing ovarian cancer is now decreased after surgery, her BRCA1+ status still confers a significant risk of developing breast cancer in the future
  - b. Further management of her BRCA1 mutation may involve discussions of prophylactic mastectomy and/or chemotherapy

## CONCLUSION

- In BRCA1+ patients who choose to continue a CMCF pregnancy, extensive counseling is necessary with consideration made for risk-reducing surgical management at the time of delivery.
- Uncomplicated delivery of a fetus is rare in CMCF pregnancy, but it is still possible with close surveillance and follow-up.
- Hysterectomy with bilateral oophorectomy at the time of delivery of a CMCF pregnancy in a BRCA1/2+ patient decreases the risk of developing GTN, ovarian, and breast cancer.

## FUTURE OUTLOOK

Areas for further research:

- Methods to more accurately diagnose CMCF pregnancy
  - current detection rates with ultrasound are around 60%<sup>2</sup>
  - usually diagnosed around 12-14 weeks gestation
- Diagnosis of CMCF pregnancy at an earlier gestational age
- MRI another useful imaging tool
- Possible role for PET-CT

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