CASE REPORT / OLGU SUNUMU

Partial Molar Pregnancy with Diploid Chromosomal Structure

Sabri Kurtay[1,*], Cuma Taşın[2]

[1] Mersin City Training and Research Hospital, Gynecology and Obstetrics Clinic, Mersin, Turkey
[2] Mersin City Training and Research Hospital, Perinatology Clinic, Mersin, Turkey

***Correspondence:** Sabri Kurtay (MD) Korukent Mah. 96015 Sok. Mersin Entegre Sağlık Kampüsü, 33240 Toroslar/Mersin E-mail: sabrikurtay@gmail.com Tel: +90 (0324) 225 10 00
Orcid ID: 0000-0003-0867-6972

ABSTRACT

Molar pregnancy is characterized by abnormal trophoblast proliferation and gestational trophoblastic neoplasms are included in the differential diagnosis. There are two types; complete and partial moles. Chromosome structure is generally diploid in the complete mole and triploid in the partial mole. Molar pregnancy recurs around 1%. Genetic mutations that can cause the disease have been identified. This report presents the management of a partial mole who had two previous molar pregnancies and whose current pregnancy was complicated by HELLP syndrome. The diploid fetal chromosome structure of the case with clinically partial mole features made the case more interesting.

Keywords: Diploid Chromosome, HELLP syndrome, Partial Molar Pregnancy, Triploid Chromosome.

ÖZET

Molar gebelik anormal trofoblast proliferasyonu ile karakterizedir ve ayırıcı tanıda gestasyonel trofoblastik neoplazmlar yer alır. Komplet ve parsiyel moller olmak üzere iki tipi vardır. Kromozom yapısı komplet molde genellikle diploid, parsiyel molde ise triploiddir. Molar gebelik %1 civarında tekrarlar. Hastalığa neden olabilecek genetik mutasyonlar tanımlanmıştır. Bu raporda, daha önce iki molar gebeliği olan ve mevcut gebeliği HELLP sendromu ile komplike olan bir parsiyel mol hastasının yönetimi sunulmaktadır. Klinik olarak parsiyel mol özellikleri gösteren olgunun diploid fetal kromozom yapısı olguyu daha ilginç hale getirmiştir.

Anahtar Kelimeler: Diploid Kromozom, HELLP Sendromu, Parsiyel Molar Gebelik, Triploid Kromozom.

1. INTRODUCTION

Molar pregnancy (mole, hydatidiform mole) (HM) is a disease characterized by abnormal proliferation of trophoblastic tissue. There are two types, complete and partial moles. Although they have different genetic, clinical and histopathological features, their common point is that they are benign. However, there is a risk of progression to gestational

trophoblastic malignancy. Therefore, invasive mole, choriocarcinoma, placental area trophoblastic tumor and epithelioid trophoblastic tumor are included in the differential diagnosis (Ning et al., 2019). Its frequency is 60-120/100,000 in North America and Europe and more in other parts of the world. Being over 35 years old (5-10 times increased risk) and under 20 years old, history of molar pregnancy (1-2 % risk increase for next pregnancy), history of spontaneous abortion and infertility, dietary factors such as carotene deficiency, animal fat deficiency, and smoking are known risk factors (Ghassemzadeh et al., 2022). The recurrence rate is about 1% (Sebire, 2021). A complete mole develops as a result of fertilization with two sperms or chromosome duplication of one sperm after the loss of the genetic material of the oocyte. Therefore, the complete mole consists only of paternal DNA and is most commonly of the 46, XX diploid karyotype (46, XY also occurs). Partial moles result from fertilization of an oocyte by two sperms and result in triploid genetic content in paternal/maternal DNA content in 2/1 ratio (Savage et al., 2017). The most common presenting complaint is vaginal bleeding that occurs in the first trimester. The most prominent ultrasound findings are heterogeneous mass appearance with multiple anechoic areas in the uterine cavity in complete mole, and enlarged placenta with cystic areas with the presence of fetus in partial mole (Bruce & Sorosky, 2022). Treatment includes surgical removal of molar pregnancy material and serial beta-hCG monitoring. Follow-up is necessary to detect possible conversion to gestational trophoblastic neoplasia. Although GTN is the invasive or malignant form of GTD, the response to chemotherapy is quite satisfactory (Berkowitz & Goldstein, 2013; Ngan et al., 2021). Intrauterine death or miscarriage rate was observed between 11% and 57% in a review of different studies (Lin et al., 2017).

2. CASE PRESENTATION

The 27-year-old patient had a history of two abortions and two molar pregnancies. Gestational age according to the last menstrual period was 10 weeks + 0 days. Ultrasonography revealed cystic areas in the placenta and bilateral theca lutein cysts with a single live pregnancy, consistent with intrauterine CRL 8 weeks + 6 days (Figure 1, 2 and 3).

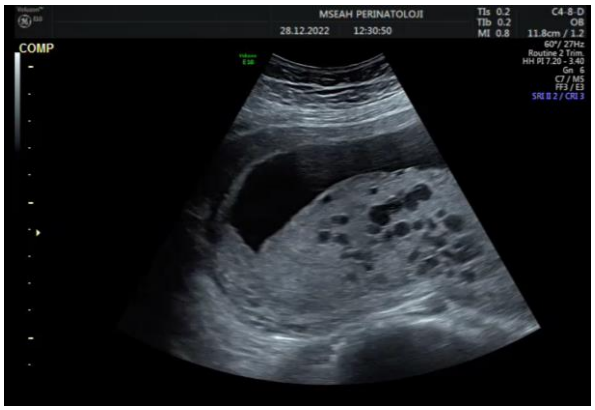

Figure 1. Cystic areas in the placenta.

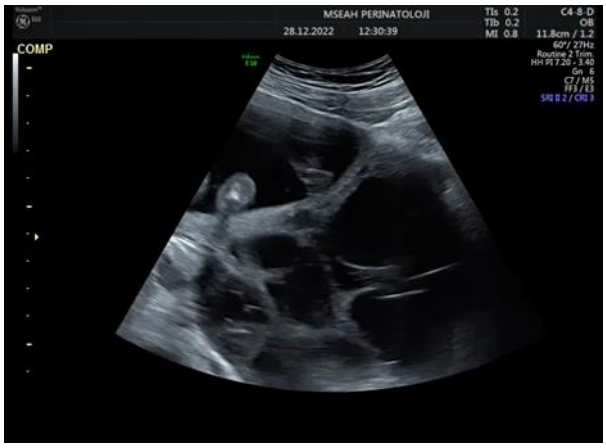

Figure 2. Theca lutein cysts.

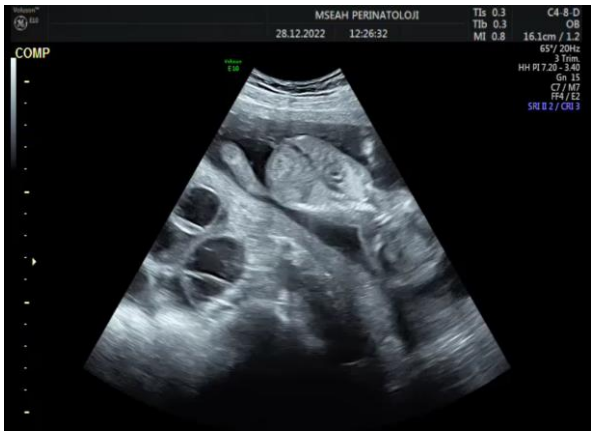

Figure 3. Fetus with intrauterine location.

Pregnancy termination was recommended to the patient because of the history of recurrent molar pregnancy. The patient demanded the continuation of the pregnancy. Amniocentesis was performed for genetic analysis. After 10 days of amniocentesis, the patient applied with the complaint of amniotic fluid leakage. As the patient also showed signs of HELLP syndrome at 10 weeks + 2 days of gestational age, pregnancy termination was performed by hysterotomy. Consent for amniocentesis, pregnancy termination, and hysterotomy were obtained at the appropriate time. There were molar pregnancy findings in the macroscopic view of the placenta (Figure 4). As a result of the analysis obtained by amniocentesis, the fetal chromosome number was 46, -- (Figure 5).

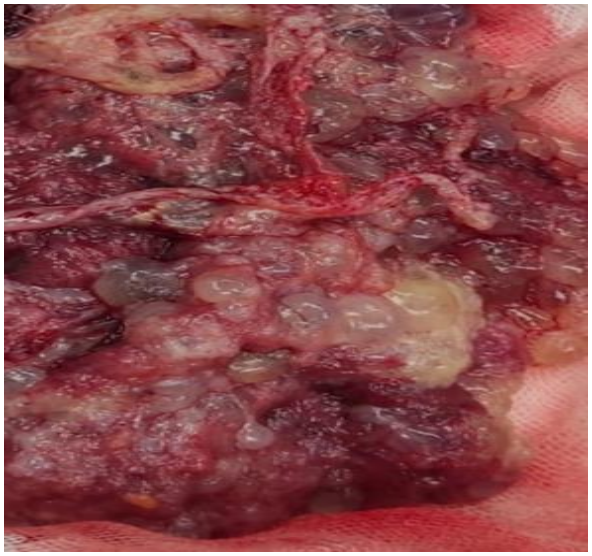

Figure 4. Macroscopic view of the placenta

3. DISCUSSION

The maximum number of recurrent consecutive molar pregnancies known in the literature has been reported as 9 (Garg & Dhingra, 2022). The patient's absence of a living child, having had 2 molar pregnancies before, and having a live fetus in the current pregnancy affected the patient's decision to demand the continuation of the pregnancy. These conditions complicate the management of molar pregnancy.

HELLP syndrome is a condition seen in 0.5-0.9% of all pregnancies, mostly secondary to preeclampsia and may cause maternal mortality (Kirkpatrick, 2010). The presence of HELLP syndrome findings along with the amniotic fluid leakage made pregnancy termination inevitable. After termination of pregnancy with hysterotomy, clinical findings improved significantly. Changes in the expression level of the NLRP7 gene have also been reported to have an effect in recurrent HM cases (Abi Nahed et., 2022). In a case report, a history of recurrent HM due to a mutation in the KHDC3L gene and the presence of familial HM were reported (Fatemi et al., 2022). These two genes account for 60% of recurrent HM cases. Mutations in the NLRP5 and PADI6 genes from the subcortical maternal complex family have also been reported to be responsible for recurrent HM cases (Rezaei et al., 2021). In our case, the quantitative fluorescent PCR result was reported as normal and the final chromosome analysis showed a 46, -- karyotype.

The genetic structure of partial molar pregnancy is known as triploid. Partial molar pregnancy cases with diploid chromosomal structure have been reported rarely in the literatüre (Rahamni & Parviz, 2016; Al Ghadeer). This partial mole, which is accompanied by HELLP syndrome and has a diploid chromosome, is a very rare condition.

4. CONCLUSIONS

Maternal and paternal genetic evaluation will help detect possible mutations in this pregnant woman with a history of recurrent molar pregnancy. The HELLP syndrome that emerged during pregnancy and the diploid chromosome structure of the fetus made the case rare and unique. The difficulty and complexity in the diagnosis and treatment process of this case were overcome with a

meticulous approach in the management of molar pregnancy. Preimplantation genetic diagnosis and oocyte donation options should be considered for the next pregnancy planning of the patient.

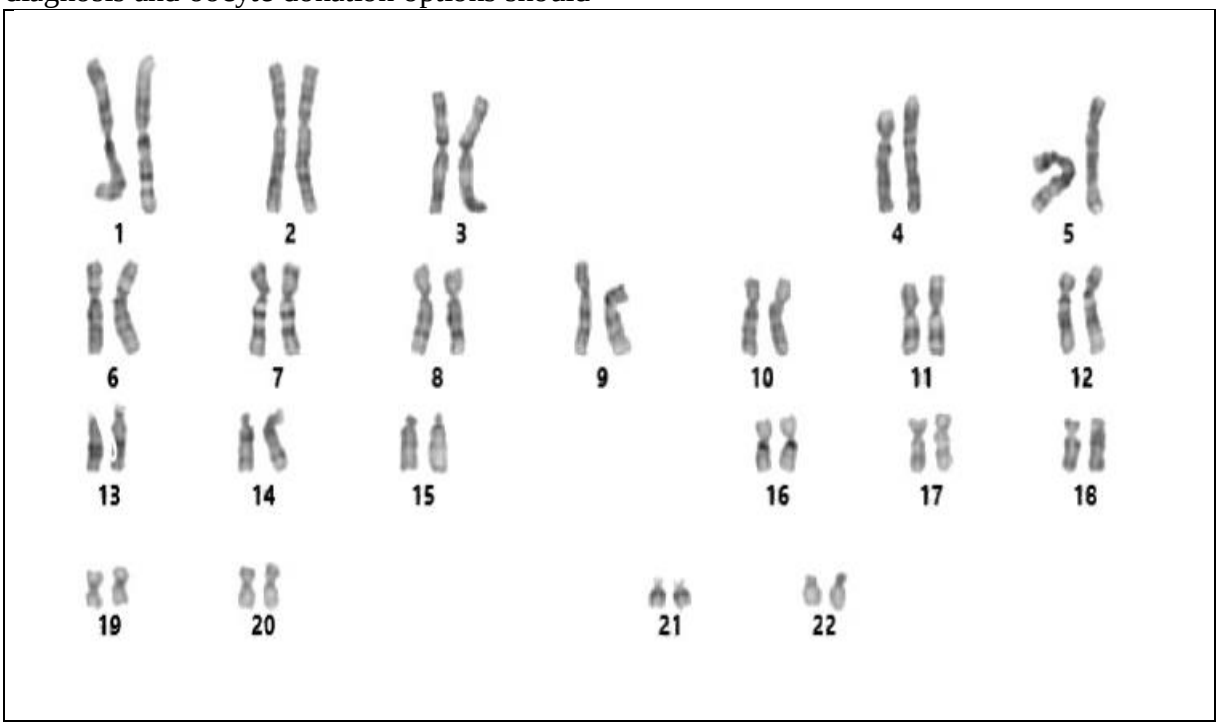

Figure 5. Fetal genetic examination result; 44 chromosomes (sex chromosomes are not shown).

Conflict of interest: None

REFERENCES

Abi Nahed R, Elkhoury Mikhael M, Reynaud D, et al. Role of NLRP7 in Normal and Malignant Trophoblast Cells. Biomedicines. 2022;10(2):252.

Al Ghadeer HA, Al Kishi N, Algurini KH, et al. Partial Molar Pregnancy With Normal Karyotype. Cureus. 2022;14(10):e30934.

Berkowitz RS, Goldstein DP. Current advances in the management of gestational trophoblastic disease. Gynecol Oncol. 2013;128(1):3-5.

Bruce S, Sorosky J. Gestational Trophoblastic Disease. StatPearls. Treasure Island (FL): StatPearls Publishing Copyright © 2022, StatPearls Publishing LLC.; 2022.

Fatemi N, Varkiani M, Ramazanali F, Almadani N, Totonchi M. A familial case of recurrent hydatidiform mole with p.Asp108Ilefs∗30 causing mutation in KHDC3L: A genetic and clinical report. Taiwan J Obstet Gynecol. 2022;61(2):395-398. doi: 10.1016/j.tjog.2022.02.038.

Garg P, Dhingra H. Recurrent hydatidiform mole in nine consecutive pregnancies: a rare case report from India. J Obstet Gynaecol. 2022;42(1):175-176.

Ghassemzadeh S, Farci F, Kang M. Hydatidiform Mole. StatPearls. Treasure Island (FL): StatPearls Publishing Copyright © 2022, StatPearls Publishing LLC.; 2022.

Kirkpatrick CA. The HELLP syndrome. Acta Clin Belg. 2010;65(2):91-97. doi: 10.1179/acb.2010.020.

Lin LH, Maestá I, Braga A, et al. Multiple pregnancies with complete mole and coexisting normal fetus in North and South America: A retrospective multicenter cohort and literature review. Gynecol Oncol. 2017;145(1):88-95.

Ngan HYS, Seckl MJ, Berkowitz RS, et al. Diagnosis and management of gestational trophoblastic disease: 2021 update. Int J Gynaecol Obstet. 2021;155(S1):86-93.

Ning F, Hou H, Morse AN, Lash GE. Understanding and management of gestational trophoblastic disease. F1000Res. 2019;8.

Rahamni M, Parviz S. A case report of partial molar pregnancy associated with a normal appearing dizygotic fetus. Asian Pac J Reprod. 2016;5(2):171-173.

Rezaei M, Suresh B, Bereke E, et al. Novel pathogenic variants in NLRP7, NLRP5, and PADI6 in patients with recurrent hydatidiform moles and reproductive failure. Clin Genet. 2021;99(6):823-828. doi: 10.1111/cge.13941.

Savage J, Adams E, Veras E, Murphy KM, Ronnett BM. Choriocarcinoma in Women: Analysis of a Case Series With Genotyping. Am J Surg Pathol. 2017;41(12):1593-1606. doi: 10.1097/pas.0000000000000937.

Sebire NJ. Recurrent GTD and GTD coexisting with normal twin pregnancy. Best Pract Res Clin Obstet Gynaecol. 2021;74:122-130. doi: 10.1016/j.bpobgyn.2020.12.002.