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CLINICAL VIGNETTE

A case of a patient at reproductive age with BRCA2 and CHEK2 mutations and multiple uterine fibroids

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BACKGROUND

Uterine fibroids (UFs) are the most common benign tumors of the female genital tract. UFs mostly affect women during their reproductive years, are remarkably rare prior to menarche, and usually regress following menopause [1]. In most cases, UFs are asymptomatic, however, the most common symptoms they cause are heavy, long and painful periods, pelvic pressure or pain, frequent urination, constipation, backache or leg pains and dyspareunia. Additionally uterine fibroids may also be associated with reproductive and obstetrical problems [2].

The reasons UFs occur and grow are still not well understood, but several UF risk factors have been identified including rising age, black race, obesity, vitamin D deficiency, endo- and exogenous hormonal factors [1]. Some UFs might develop as a result of inherited genetic mutations. However, not all of them develop from hereditary reasons [3].

Treatment tactics for most of UFs are size reduction, UF-related abnormal uterine bleeding control, or definitive removal of the fibroids. Present therapeutical methods include pharmacology, interventional radiology, and surgical procedures [4]. The choice of therapy

depends on the patient's personal treatment purpose. Nowadays, the patient should also take responsibility for her choices and actively participate in the decision-making process [5].

CASE REPORT

A now 29-year-old patient (in 2023 year) with UFs, cancer familial aggregation, familial renal suspicion and high risk of breast cancer. The patient is a carrier of CHEK2 and BRCA2 mutations. Transvaginal ultrasound confirmed UFs in 2015. In this year patient had enlarged uterus, 4 UFs were found, the largest of them had a diameter of 14 mm (Fig. 1A). The patient reports that she has always had heavy and painful periods. In the next years, the growth of UFs and the intensification of symptoms occurred. In 2019, 8 fibroids were found in transvaginal scan and the largest of which was 42 mm in diameter (Fig. 1B). Due to BRCA mutations and patients worries combined contraception was not prescribed. Some clinicians had already persuaded the patient to remove the uterus, but the patient still wanted to remain fertile. In order to reduce the symptoms, a 19.5 mg levonorgestrel-releasing intrauterine system (LNG-IUS) was inserted (Fig. 1C), which fell out after 10 months. Second insertion of the same type of system was done, but this system felt out only after 2 months. Patient and clinicians decided to change the therapy, intramuscular medroxyprogesterone was administered. Unfortunately, patient reported pain and irregular bleeding. In the next months, symptoms were increasing, patient had numerous consultations in the emergency ward. The next step was a two-stage (12.2020 and 02.2021) hysteroscopic resection of intracavitary UFs, preceded by the use of goserelin. Histopathological result revealed the typical UFs. Procedures reduced the menstrual bleeding and patient quality of like raised. In the next ultrasound scan about 12–13 fibroids were found. Due to significant reduction of visualization with ultrasound the magnetic resonance imaging examination was performed. The lesions were unsuspected and the largest UF had an average of 54 mm in diameter. Few months later a new 52 mg LNG-IUS was inserted. The use of this system gave the patient several months of very good functioning. Unfortunately, after some time, the patient came back for a check-up due to a palpable tumor over the pubic symphysis. A decision was made to enucleate them UFs surgically. Eleven UFs were enucleated in open surgery procedure (12.2022). In the histopathological examination, atypical fibroids were confirmed. One month later a diagnostic hysteroscopy was performed to exclude intrauterine adhesions. Ultimately, the patient decided to accelerate her procreation plans.

CONCLUSION

A young patient and her genetic burden make the therapy of UFs difficult. The patient's desire to leave the uterus and the possibility of becoming pregnant requires complex pharmacological and surgical therapy. Additionally, the choice of therapy depends on the patient's personal treatment purpose.

Article information and declarations

Conflict of interest

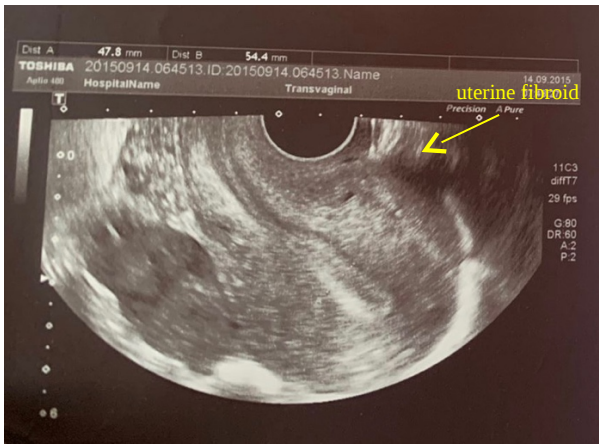
All authors declare no conflict of interest.

References

1. Yang Q, Ciebiera M, Bariani MV, et al. Comprehensive Review of Uterine Fibroids: Developmental Origin, Pathogenesis, and Treatment. Endocr Rev. 2022; 43(4): 678–719, doi: 10.1210/endrev/bnab039, indexed in Pubmed: 34741454.
2. Stewart EA, Laughlin-Tommaso SK, Catherino WH, et al. Uterine fibroids. Nat Rev Dis Primers. 2016; 2: 16043, doi: 10.1038/nrdp.2016.43, indexed in Pubmed: 27335259.
3. Välimäki N, Kuisma H, Pasanen A, et al. Genetic predisposition to uterine leiomyoma is determined by loci for genitourinary development and genome stability. Elife. 2018; 7, doi: 10.7554/eLife.37110, indexed in Pubmed: 30226466.
4. American College of Obstetricians and Gynecologists (ACOG). Encyclopedia of Global Health. 2008, doi: 10.4135/9781412963855.n61.
5. Riggan KA, Stewart EA, Balls-Berry JE, et al. Patient Recommendations for Shared Decision-Making in Uterine Fibroid Treatment Decisions. J Patient Exp. 2021; 8: 23743735211049655, doi: 10.1177/23743735211049655, indexed in Pubmed: 34692992.

Figure 1. Transvaginal scans of a uterus

A — ultrasound of uterus with small UFs, the largest UF 14 mm in diameter; **B** — ultrasound of the enlarged uterus, the largest UF 41 mm in diameter; **C** – ultrasound of uterus with 19.5 LNG-IUS; **D** — ultrasound of uterus after the surgical intervention — proper healing of the uterine muscle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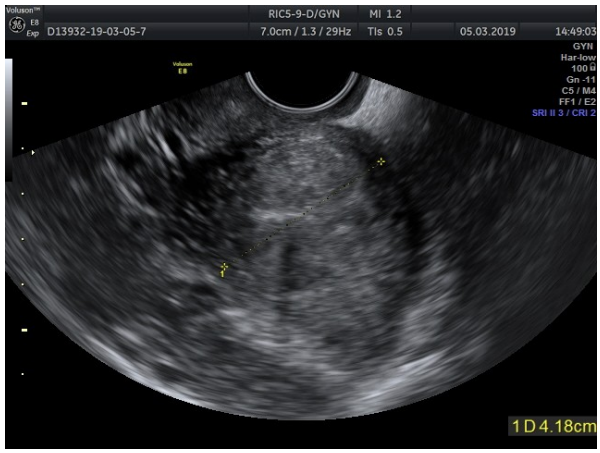
A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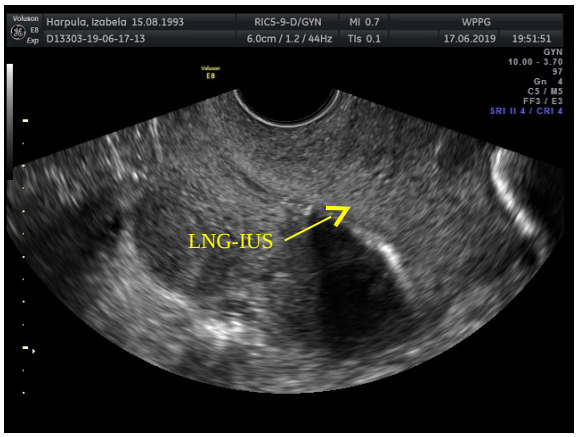
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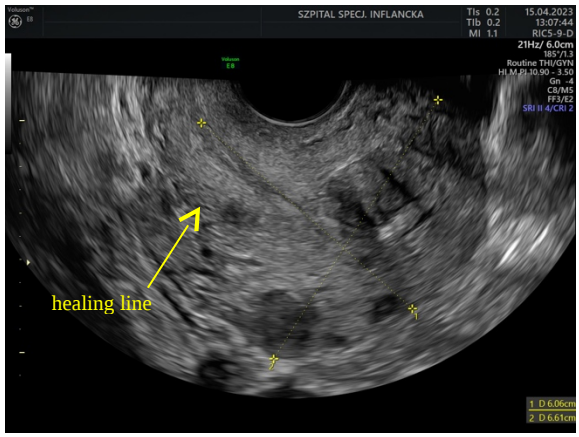
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