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CLINICAL CASE

Dysgerminoma in an adolescent with Down syndrome: a case report

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Abstract

Introduction: Germ cell tumors are the most prevalent ovarian malignancy in adolescents and girls; they are generally detected in early stages. The association with Down syndrome, the reason for presenting this case, is unknown.

Clinical Case: We present the case of a 13-year-old girl with Down syndrome, referred by a painful su- prapubic mass with two months of evolution.

Diagnostic workshop: The extension studies detected a tumor at the pelvic level dependent on the left ovary, for which a lumpectomy was planned. The histopathological examination determined the presence of a germ cell tumor with a dysgerminoma and trophoblastic component.

Evolution: The patient was prescribed chemotherapy treatment, with favorable development at 16 months of follow-up.

Conclusion: The classic symptoms of germ cell tumors in Down syndrome are not very indicative; In most cases, it is about preserving fertility, even when girls are carriers of Down Syndrome. Follow-up, in this case, has been favorable for 16 months.

Keywords:
MESH: Dysgerminoma, Down syndrome, Adolescent, Ovarian neoplasms.

Introduction

Dysgerminomas are a group of germ cell tumors representing less than 1% of all ovarian germ cell tumors, and 2-5% are malignant [1]. It has an incidence of 0.5-2 x 100 thousand in children under 15 years of age [2]. There is no clear relationship between these tumors and Down syndrome; instead, an association has been seen in this type of patient with seminoma in men;

the familial component is present in almost all reported case reports [1]. Disaia et al. explain his classification in Figure 1 [3].

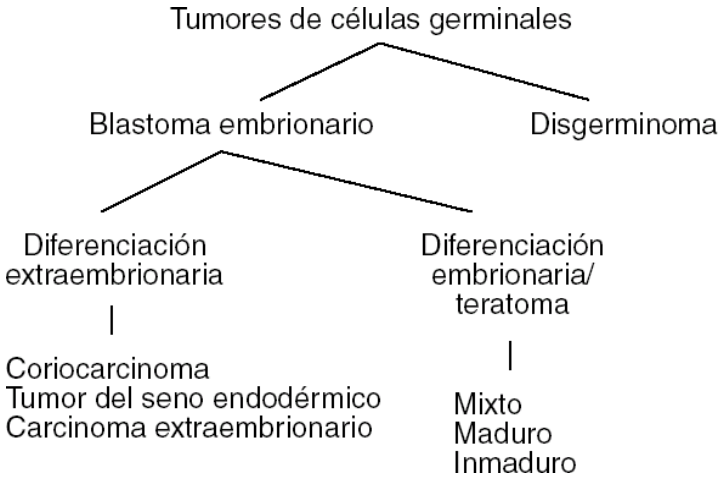

Figure 1. Classification of germ cell tumors

Clinical case

A 13-year-old female patient with a history of Down syndrome reported a painless increase in abdominal diameter two months prior, with no apparent cause, fever, and 3 kg weight loss; at the suprapubic level, she palpated an indurated, mobile, painless mass measuring 14 x 10 cm.

Diagnostic workshop

The tomography revealed a solid heterogeneous ovarian mass with lobulated edges, meas uring 14×12×8 cm, with arterial flow, hypodense areas, necrotic degeneration and calcified areas, and retroperitoneal, pelvic, and mesenteric adenomegalic conglomerate. CA-125: 155.6 U/ml, alpha-feto-protein: 1000.0 ng/ml, Ca-embryonic antigen: 0.89 ng/ml, βHCG: 83.00 mIU/ml, alkaline phosphatase: 266 mg/dL, and lactic dehydrogenase 6510 U/l.

It was classified as an ovarian tumor of uncertain behavior. Left salpingo-oophorectomy , omentectomy, excision of para-aortic lymph nodes, and appendectomy were performed. The findings were as follows (Figure 2): a multinodular cystic formation of 14 x 13 x 6 cm, with an entire external surface, furrowed by tortuous vessels, an edematous left uterine tube of 6 x 0.5 cm with virtual lumen, adipose tissue with hemorrhagic areas from which a nodular formation of 0.5 cm was isolated, without complications. Peritoneal lavage fluid cytology was positive for malignancy, dysgerminoma-type germinal ovarian tumor and left adnexal tumor were present, and the omentum was positive for malignancy. Microscopically (Figure 2), histology showed a germ cell neoplasm composed of large polygonal cells with clear cytoplasmic borders, a rounded nucleus, a prominent nucleolus, and eosinophilic cytoplasm. Mitotic activity: 20 mitoses in 10 fields. Focal syncytiotrophoblastic cell formation. Omentum with focal tumor cells. Regional lymph nodes: four of six positive for malignancy, cecal appendix free of tumor. Therefore, it was classified in the ovarian cancer stage (FIGO 2014-AJCC 2017): IIIA1; and stage IV pT3-N1-M1; no molecular studies were available.

Evolution

Eleven cycles of chemotherapy with bleomycin, cisplatin, and etoposide were started. At 16 months of follow-up, the patient has remained without clinical relapse or, in complementary tests, favorable evolution.

Discussion

The association between Down syndrome and dysgerminoma is not clear. Nevertheless, its study has been carried out in greater detail due to the increase in life expectancy of these patients. Joseph D mentions that there is a relationship between seminoma in male patients with trisomy 21 due to genetic overexpression, hormonal alterations, and greater vulnerability of trisomic cells to carcinogenic agents. These factors would explain their equivalence to dysgerminoma in women [4].

According to Satgé D, among the hypotheses that associate both entities are 1-Creation of an aberrant premeiotic 4n oogonial reserve where the trisomy chromosomes interfere with the pairing of homologous chromosomes. 2-Greater survival of abnormal cells due to overexpression of genes on chromosome 21. 3-Due to genomic instability, a single clonogenic precursor develops related to carcinoma in situ in the testis. 4-Puberty triggers the proliferation of abnormal cells when stimulated by active estrogens at this stage of development. 5-In these patients, a higher prevalence of dysgerminoma and seminoma in malignant germ cell tumors has been evidenced due to the overexpression of CKIT as a result of the trisomic state [5].

Dysgerminoma symptoms include abdominal pain and distension, pelvic mass, precocious puberty, metrorrhagia, and acute abdomen (10%), caused by rupture, hemorrhage, or torsion of the tumor [1]. The tomography in our patient showed a solid and heterogeneous ovarian mass with lobulated edges and arterial flow. A case report indicates that multiple tumor nests and central blood vessels can guide suspicion of this diagnosis [6].

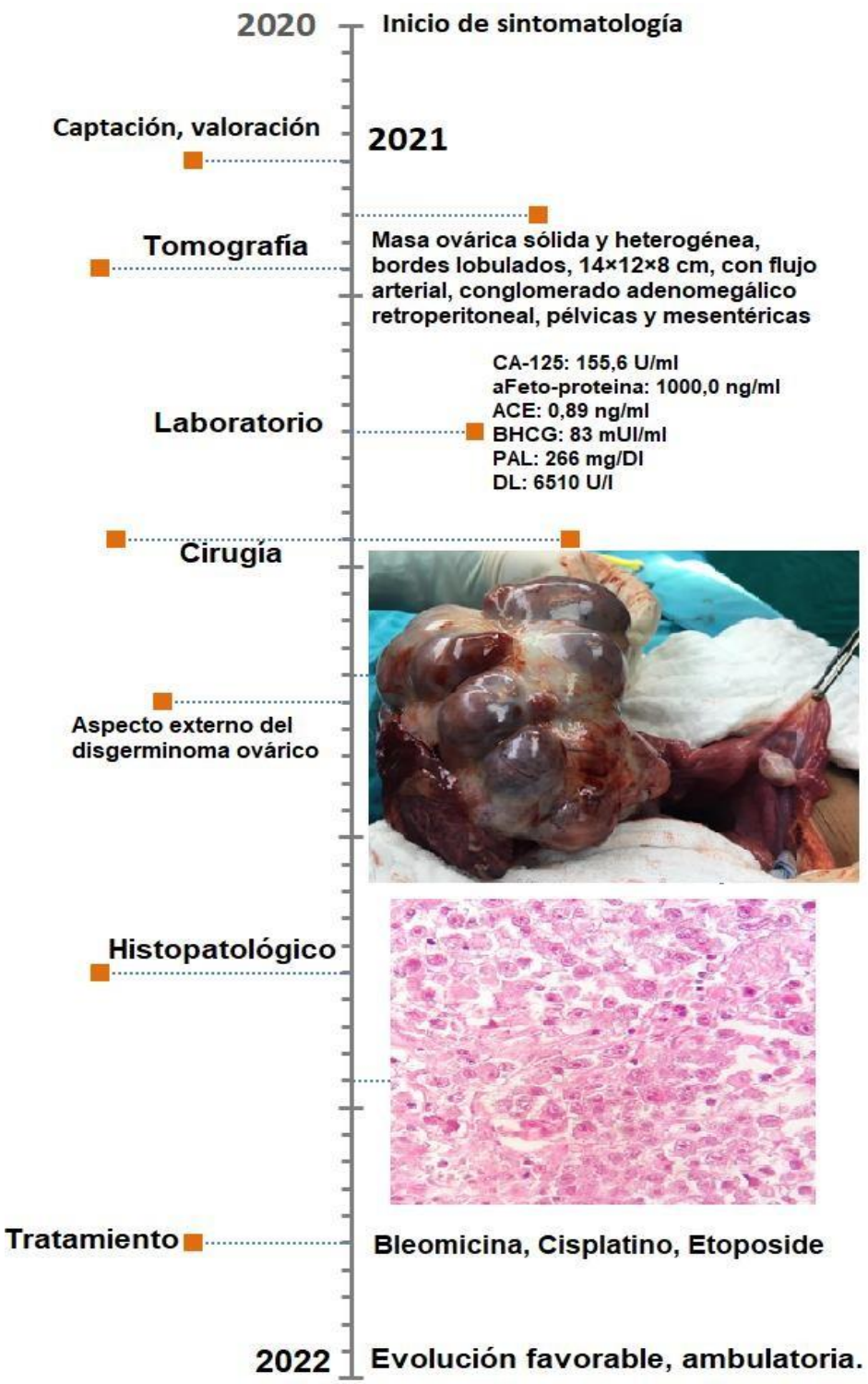

Figure 2. Timeline, dysgerminoma in an adolescent with Down syndrome

CA-125: cancer antigen 125. ACE: carcinoembryonic antigen. BHCG: human chorionic gonadotropin. PAL: alkaline phosphatase. DL: lactic dehydrogenase.

In the present case, salpingo-oophorectomy, omentectomy, excision of para-aortic lymph nodes, and appendectomy were performed due to ovarian cancer stage IIIA1 and pathological stage IV pT3-pN1-pM1. Zogbi L recommended that surgery be as conservative as possible; the majority (75%) of the patients were diagnosed as FIGO-I stage, with a 5-year survival of 95%. In his case report, the patient was in stage IIC (T2C, N0, M0), opting for radical cytoreductive surgery, lymphadenectomy, hysterectomy, and omentectomy [7].

The use of chemotherapy increases survival rates; hysterectomy and bilateral salpingo - oophorectomy do not change the prognosis since it depends on the size of the tumor, histological type, and clinical stage. The most widely used chemotherapy schemes include bleomycin, etoposide, cisplatin or vincristine, dactinomycin, and cisplatin. Our patient received bleomycin, cisplatin, and etoposide. The survival rate is 95% in the early stages, 80% in stage III, and 60% in stage IV [8].

The therapeutic objective in young people is to preserve fertility. For stages III and IV, hysterectomy and bilateral adnexectomy, resection of the omentum and appendix are recommended due to their 5-year survival rate of 96% and relapse rate of 17%. The recurrent tumor remains doubtful and can be treated in a second operation; however, fertility-sparing surgery should be carefully considered in cases with exogenous papillary structure and peritoneal infiltration and implantation. In the present case, peritoneal fluid was positive for malignancy, dysgerminoma-type ovarian tumor, left adnexal, and omentum positive for malignancy [9]. The presence of postoperative residual tumors is the most important poor prognostic factor; furthermore, emphasizing the chemotherapy-sensitive nature of these tumors, an adequate attempt at maximal debulking without compromising fertility seems a reasonable surgical approach in the initial treatment for these patients [10].

Conclusions

We present a clinical case of an adolescent patient with Down syndrome who developed an ovarian dysgerminoma, underwent surgery, and was managed with chemotherapy, with better clinical evolution and quality of life. The classic symptoms of germ cell tumors in Down syndrome are not very indicative; in most cases, it is about preserving fertility, even when girls are carriers of Down Syndrome. Follow-up, in this case, has been favorable for 16 months.

Acknowledgments

Does not apply.

Administrative information

Abbreviations

CA-125: cancer antigen 125.
CEA: carcinoembryonic antigen.
BHCG: humanchorionic gonadotropin.
PAL: alkaline phosphatase.

DL: lactic dehydrogenase.

Additional Files

None declared by the authors.

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Availability of data and materials

Data are available upon request to the corresponding author. No other materials are reported.

Author contributions

Gabriela Peñaherrera Cepeda: Conceptualization, Formal analysis, Research, methodology, Project administration, Supervision, Validation, Visualization, Writing-original draft, Writing-revision, and editing.
Kathia Maldonado Merino: Conceptualization, formal analysis, research, validation, writing-revision, and editing.
Estefanía Maldonado Merino: Conceptualization, formal analysis, research, validation, writing-revision, and editing.
All authors read and approved the final version of the manuscript.

Ethics committee approval

It does not apply to observational studies and narrative reviews.

Consent for publication

The authors have written authorization from the patient's mother to publish images presentedin this article.

References

1. Pérez-Ortiz V, Reyna-Villasmil E. Malignant mixed ovarian germ cell tumor. Case report. Peruvian Journal of Gy- necology and Obstetrics [Internet]. 2020 February 3 [cited 2020 September 4];66 (1):107-11. https://doi.org/10.31403/rpgo.v66i2241

2. Socorro Castro C, Chávez Valdivia M, Martínez Navarro J. Pure ovarian dysgerminoma in adolescents: presen- tation of a case and review of the literature. Finlay Magazine [Internet]. 2018 Dec [cited 2020 Sep 11];8(4):321 -6. SCIELO: S2221

3. Philip J. DiSaia P, William T. Creasman, Robert S Mannel, Scott McMeekin, David G Mutch4. Clinical gynecologic oncology. Available at: books.google

5. Satgé D, Honoré L, Sasco AJ, Vekemans M. An ovarian dysgerminoma in Down syndrome. Hypothesis aboutthe association. International Journal of Gynecological Cancer [Internet]. 2006;16 (S1):375-9. https://doi.org/10.1111/j.1525-1438.2006.00211.x
PMid: 16515627

6. Tsuboyama T, Hori Y, Hori M, et al. Imaging findings of ovarian dysgerminoma with emphasis on multiplicityand vascular architecture: pathogenic implications. abdomen Radiol [Internet]. 2018 July 1;43 (7):1515-23. https://doi.org/10.1007/s00261-018-1503-6
PMid: 29450608

7. Zogbi L, Gonçalves CV, Tejada VF, et al. Treatment of bilateral ovarian dysgerminoma with 11-year follow-up: A case report. Ann Med Surg (London) [Internet]. 2018 August 21;33:50 -2. https://doi.org/10.1016/j.amsu.2018.08.009
PMid: 30186597 PMCid: PMC6122391

8. Polanco-Sosa AL, Peña-Montemayor AK, Mireles-García AM. Mixed ovarian germ cell tumor, an unusual combi- nation. Gynecology and Obstetrics of Mexico. 2020;4. SCIELO: mx/S0300

9. Su M, Chang W, Xu T, et al. Characteristics of diagnosis and therapy of adolescent malignant ovarian tumors.Eur J Gynecol Oncol. 2013;34 (6):565-8.

10. Li J, Wu X. Current Strategy for the Treatment of Ovarian Germ Cell Tumors: Role of Extensive Surgery. CurryTreat Options in Oncol [Internet]. 2016 June 29;17(8):44. https://doi.org/10.1007/s11864-016-0416-2
PMid: 27357180

Editor's Note