International Journal of Medical Science and Clinical Research Studies

ISSN(print): 2767-8326, ISSN(online): 2767-8342

Volume 04 Issue 12 December 2024

Page No: 2147-2150

DOI: https://doi.org/10.47191/ijmscrs/v4-i12-07, Impact Factor: 7.949

Neurofibromatosis type 1 and High-risk Pregnancy: A Case Report

Orlando Jesus Sarmiento Haydar[1], Catalina Ivonne Chan Sierra[2], Juan Carlos Cachón Alpuche[2], Manuel Andrés Miranda Guillermo[2]

[1]Gynecology and Obstetrics Resident, Facultad de Medicina de la Universidad Autónoma de Yucatán. Clínica Hospital Mérida ISSSTE. Calle 21 s/n Comisaría de Susulá, Mérida, Yucatán, México. CP 97314.

[2]Gynecology and Obstetrics Service, Clínica Hospital Mérida ISSSTE. Facultad de Medicina de la Universidad Autónoma de Yucatán

ABSTRACT

Introduction: A high-risk pregnancy is the result of a disease present before pregnancy. Neurofibromatosis type 1 is a rare disease during pregnancy, autosomal dominant, with a mutation in the NF1 gene, on chromosome 17q11.2.

Clinical case: 38-year-old woman, Gestational age 1. Mother with neurofibromatosis type 1. The patient is hypertensive, diagnosed 7 years ago and currently treated with alpha-methyldopa 250 mg every 12 hours orally. She reports the diagnosis of neurofibromatosis type 1 since childhood. Without adequate prenatal control, with alterations in the Doppler ultrasound and complication of intrauterine growth restriction stage 1. A referral is made to third level of care and evaluation by maternal-fetal medicine

Discussion: Pregnancies in patients with neurofibromatosis are predisposed to obstetric complications due to the multisystemic nature of the disease. These patients have a higher incidence of spontaneous abortions, stillbirth, preeclampsia, intrauterine growth restriction (IUGR), oligohydramnios, premature birth and cerebrovascular complications.

KEYWORDS: neurofibromatosis type 1, high-risk pregnancy, chronic arterial hypertension, fetal growth restriction, maternal-fetal medicine.

ARTICLE DETAILS

Published On:
03 December 2024

Available on:
https://ijmscr.org/

INTRODUCTION

Neurofibromatosis (NF) is a group of hereditary neurocutaneous syndromes associated with tumors of the central and peripheral nervous system. There are 3 subtypes: Neurofibromatosis type 1 (NF1), neurofibromatosis type 2 (NF2) and Schwannomatosis; NF1 is the most representative with 96% of all cases, also known as Von Recklinghausen disease [1].

Neurofibromatosis type 1 is an autosomal dominant disease, with a mutation in the NF1 gene, on chromosome 17q11.2. Clinically, it is characterized by the presence of neurofibromas, café-au-lait spots, ephelides, Lisch nodules, optic gliomas, skeletal and neurological abnormalities, among others [2].

The cutaneous manifestations of neurofibromatosis 1 significantly reduce health-related quality of life. Neurofibromas can contribute to a deterioration in quality of life, anxiety and mood in patients [3].

For the presence of café-au-lait pigmentations and benign neurofibromas, no treatment is applied, except if the patient presents symptomatic lesions, which leads to a surgical intervention for their excision [4].

A high-risk pregnancy is the result of a disease present before pregnancy. High-risk pregnancies require strict control and should be referred to a perinatal center [5].

It is essential from the beginning to evaluate and categorize obstetric risk factors such as: family history, economic-social factors, nutritional factors, disability, pelvic-genital pathologies and current pregnancy, with hypertensive disorders being the most prevalent pathologies [6].

CLINICAL CASE

38-year-old woman, first pregnancy, who comes to our hospital for the first time, without adequate prenatal care.

Upon direct questioning: mother with neurofibromatosis type 1. The patient is hypertensive diagnosed 7 years ago

and currently on treatment with alpha-methyldopa 250 mg every 12 hours orally. She reports the diagnosis of neurofibromatosis type 1 since childhood without follow-up or treatment. First pregnancy, she reports the start of prenatal care at 2 months of pregnancy, however, she does not present any recent laboratory or ultrasound studies; she only shows an ultrasound from March 5, 2024 that reports 8.6 weeks of pregnancy. She does not remember the date of her last menstruation.

Physical examination with vital signs in normal parameters: BP 130/80 MMHG. Uterine fundal height of 27 centimeters, without uterine activity or cervical dilation.

Abdomen and back with the presence of café-au-lait spots and cutaneous neurofibromas on the abdomen, back and extremities (figures 1, 2 and 3).

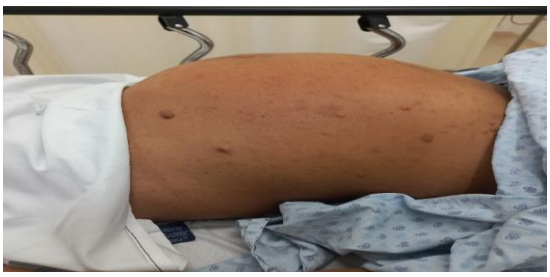

Figure 1. Cutaneous neurofibromas on the abdomen and hyperpigmented macules (café au lait macules).

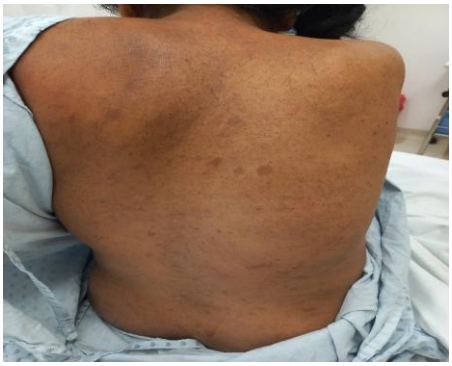

Figure 2. Back with hyperpigmented macules (café au lait macules).

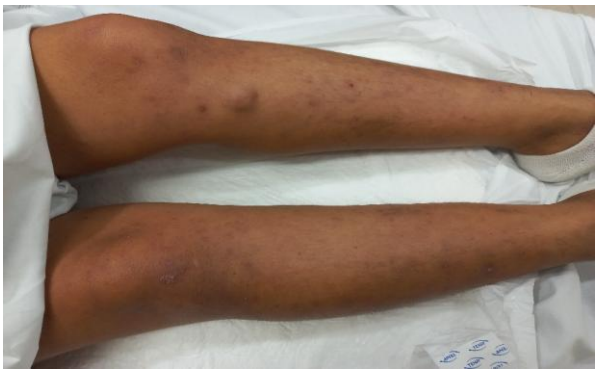

Figure 3. Enlarged cutaneous neurofibroma on the left leg.

A cardiotocographic record was performed in 20 minutes, classifying as category 1.

Laboratory tests upon admission reported: hemoglobin 12.8 g/dl, hematocrit 41.0%, platelets 214,000, total leukocytes 9420, PT 11.9 sec, PTT 29.5 sec, INR 1.18, glucose 55 mg/dl, urea 24.3 mg/dl, creatinine 0.49 mg/dl, uric acid 4.8 mg/dl, ALT 20 u/l, AST 19 u/l, DHL 137 u/l, GGT 12 u/l, blood group O positive, VDRL negative, HIV negative.

An obstetric ultrasound was performed with findings of a single fetus, cephalic presentation and longitudinal position, with a fetal heart rate of 139 beats per minute. The biparietal diameter measures 80 mm, corresponding to 32.1 weeks of gestation and is below the 3% percentile. The head circumference is 306 mm, corresponding to 34.1 weeks and is below the 3% percentile. The abdominal circumference measures 285 mm, equivalent to 32.4 weeks, below the 3% percentile. The femoral length is 58 mm, corresponding to 30.4 weeks of gestation, located below the 3% percentile. The middle cerebral artery has a pulsatility index (PI) of 1.36, at the 4% percentile (Figure 4).

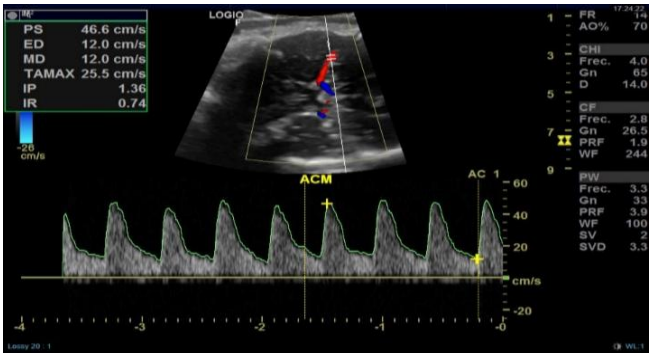

Figure 4. Middle Cerebral Artery Pulsatility Index.

The umbilical artery is located at the 32nd percentile (Figure 5).

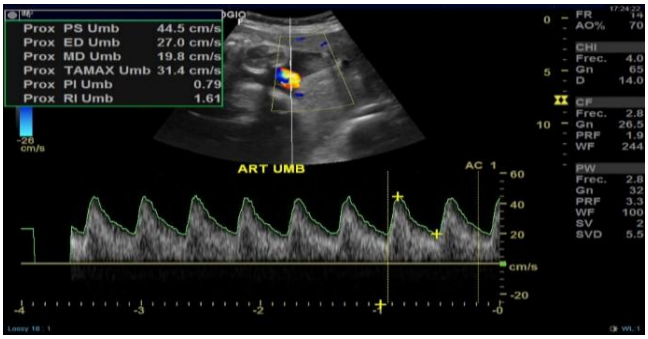

Figure 5. Umbilical Artery Pulsatility Index.

The cerebroplacental ratio is 0.84, at the <1% percentile. With an estimated fetal weight of 1903 grams, less than the 3rd percentile. The amniotic fluid index is 9.4 cm. The placenta is located in the fundic region and has a degree of maturation 2.

Finally, the diagnosis of high-risk pregnancy at 35.6 weeks is established by ultrasound of the first trimester / neurofibromatosis type 1 / fetal growth restriction stage 1 / chronic arterial hypertension.

As we are a second level hospital and based on the ISSSTE reference criteria for high-risk pregnant patients, a referral is made to a third level of care for evaluation by the Maternal-Fetal Medicine service.

DISCUSSION

Neurofibromatosis type 1 occurs with a frequency during pregnancy in 1/2 500 to 1/18 500 births, with a 50% probability that the offspring will have it, regardless of sex [7].

Giliyar Shobha, et al [8] published a retrospective study that identified a total of 60 patients with neurological disorders during the 3-year study period. They identified that 36 women had epilepsy (60%), seven cerebrovascular diseases (11.6%), six polio survivors (10%), four central nervous system infections (6.6%) and one with neurofibromatosis (1.6%).

Obstetric risk refers to the early evaluation of risk factors that compromise maternal-fetal well-being during gestation, among these are hypertensive disorders that cause systemic alterations during pregnancy [9]. Our patient is chronically hypertensive with a neurological and dermatological disease diagnosed since childhood, however, she did not attend adequate prenatal care to prevent future complications.

It has been observed that cutaneous neurofibromas increase in size or number in up to 82% of pregnancies and in rare cases, they can appear in large numbers at the same time (eruptive neurofibromas) [10]. In our case, the patient reported that during pregnancy she had an increase in size of two neurofibromas located in the abdomen and one in the left lower extremity; she denied the eruption of new cutaneous neurofibromas.

Bharathi, et al [11] published 7 cases of pregnancy with neurofibromatosis type 1, of which 4 presented the following obstetric complications: 1 case of a mother with epilepsy that worsened her seizure activity, 1 case that presented preeclampsia with severity criteria and 2 cases with fetal growth restriction.

Intrauterine growth restriction (IUGR) is the inability of the fetus to reach its full genetic and growth potential. Children with IUGR are five times more likely to die during the neonatal period, four times more likely to die during their postnatal period [12]. When performing the obstetric ultrasound, we obtained an estimated fetal weight of 1903 grams, below the 3rd percentile, so Doppler ultrasound was subsequently performed to stage the disease.

Fetal growth restriction (FGR) is a pathological decrease in the fetal growth rate, generally associated with placental insufficiency. It is diagnosed by obstetric ultrasound and Doppler velocimetry [13]. The Doppler ultrasound performed in our hospital reports the following: the middle cerebral artery presents a percentile of 4%, which is considered pathological. The umbilical artery is at the 32nd percentile, within normal limits. The cerebroplacental relationship with a percentile of <1%, which is pathological. We concluded that fetal growth restriction was stage 1, repeating the Doppler study in 1 week, considering termination of pregnancy at 37 weeks and referral for evaluation by maternal-fetal medicine.

Pregnancies in patients with neurofibromatosis are predisposed to obstetric complications due to the multisystemic nature of the disease. These patients have a higher incidence of spontaneous abortions, fetal death, preeclampsia, IUGR, oligohydramnios, premature birth and cerebrovascular complications. Preeclampsia and IUGR can be attributed to vasculopathy leading to placental hypoxic changes [14]. In our case, the patient did not undergo preventive screening for preeclampsia or fetal growth restriction with Doppler ultrasound of the uterine arteries in the first trimester, nor did she have adequate prenatal monitoring to prevent these diseases and for this reason, she was not evaluated by the neurology, dermatology, internal medicine or maternal-fetal medicine services in relation to her chronic diseases and high-risk pregnancy.

CONCLUSIONS

In a high-risk pregnancy, the importance of adequate prenatal monitoring to identify risk factors and prevent future obstetric and perinatal complications is highlighted.

Neurofibromatosis type 1 is a rare disease during pregnancy, timely diagnosis of complications associated with the disease is important, using laboratory studies and performing Doppler ultrasound. It is important to carry out adequate monitoring of the pregnancy by maternal-fetal medicine, as well as multidisciplinary management and close monitoring in a third-level perinatal care center.

ACKNOWLEDGMENT

To “Instituto de Seguridad y Servicios Sociales de los Trabajadores del Estado (ISSSTE)” and the “Facultad de Medicina de la Universidad Autónoma de Yucatán (UADY)”.

.

REFERENCES

I. Hinojoza Remache, R. A., Moreno Carillo, A. B., Valero Ortega , A. E., & Burgos Paladines, A. J. (2024). Neurofibromatosis, Generalidades y Manifestaciones Clínicas Una Revisión Bibliográfica. Ciencia Latina Revista Científica Multidisciplinar, 8(1), 919-928. https://doi.org/10.37811/cl_rcm.v8i1.9465

II. Khare A, K. Verma K, Sambangi A, Dhavala M, B. Tarbox M. Detecting Manifestations of Neurofibromatosis: Cutaneous Diagnostic Criteria

for NF1 and NF2. Journal of Clinical Dermatology and Surgery. 2024;2(2). DOI:10.61853/z7cg8k08

III. Daniela, Rosales & Gradis, Olman & María, Enríquez & Shirley, Andino. (2024). Neurofibromatosis tipo 1 en paciente puérpera. Medicina Clínica Práctica. 7. https://doi.org/10.1016/j.mcpsp.2024.100454.

IV. Dance B, Dardare A, Fleming J, Siow SF, Schlub TE, Crawford H, Saunderson RB, Wong C, Berman Y. The impact of cutaneous neurofibromas on quality of life and mental health in neurofibromatosis type 1. J Dermatol. 2024 Aug;51(8):1050-1059. DOI: 10.1111/1346-8138.17276.

V. Guzmán, Juana & Viteri Rojas, Ana & Vallejo Lopez, Alida. (2023). Embarazo de alto riesgo. RECIAMUC. 7. 371-380. DOI:10.26820/reciamuc/7.(1).enero.2023.371-380.

VI. Arroyo Cadena, L. M., Navas Almeida, A. R., & Hidrobo Guzmán, J. F. (2023). Riesgo obstétrico y su relación con los trastornos hipertensivos en gestantes. Ciencia Latina Revista Científica Multidisciplinar, 7(1), 9707-9720. https://doi.org/10.37811/cl_rcm.v7i1.5168

VII. García Odio, A., Rives González, Y., Reina Arroyo, A., & Álvarez Bolívar, D. (2016). Embarazada con neurofibromatosis tipo 1. Revista de Ciencias Médicas de Pinar del Río, 20(5), 645-650.

VIII. Giliyar S, Kanavi JV, Thomas A. Neurological Disorders Complicating Pregnancy and its Obstetric Outcomes. Int J Infertil Fetal Med 2023;14(2):59–64.

IX. Arroyo Cadena, L. M., Navas Almeida, A. R., & Hidrobo Guzmán, J. F (2023) Riesgo obstétrico y su relación con los trastornos hipertensivos en gestantes. 7(1), 1-16. https://doi.org/10.37811/cl_rcm.v6i5.2903

X. Yahya, Husain; Sani, Hadiza. Eruptive Neurofibromas in Pregnancy. Annals of African Medicine 19(2):p 150-152, Apr–Jun 2020. DOI:10.4103/aam.aam_37_19

XI. Bharathi, Shree & N S, Kubera & Jha, Nivedita & Chanu, Sairem. (2021).. CASE SERIES OF NEUROFIBROMATOSIS AND ITS VARIED MANIFESTATIONS IN PREGNANCY: AN OBSTETRICIAN'S PERSPECTIVE. INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH, 68–70. https://doi.org/10.36106/ijsr/7824991

XII. Cabezas, Bertha & Canchingre, Karen & Tobar Blandón, Maria & Vaca, Diego & Cabezas, Giovanny & Ochoa Andrade, Miguel. (2021). Restricción del crecimiento intrauterino asociado a factores de riesgo maternos, en el servicio de neonatología del Hospital General IESS-Ibarra. Enero 2016-abril 2018. Revista Ecuatoriana de Medicina EUGENIO ESPEJO. DOI: 10.23936/ree.v7i13.95

XIII. Sandoval, Isidora, Véliz, Romina, Sepúlveda, Álvaro, Candia, Alejandro A., & Herrera, Emilio A.. (2022). Utilidad de la ecografía Doppler en el diagnóstico, el pronóstico y el manejo de la restricción del crecimiento fetal: situación en Chile. Revista chilena de obstetricia y ginecología, 87(3), 218-228. https://dx.doi.org/10.24875/rechog.21000011

XIV. Dr. Venus Bansal, Dr. Muskaan Chhabra, Dr. Sonam Gilhotra. Neurofibromatosis and its varied manifestations in pregnancy: A case report. Int J Clin Obstet Gynaecol 2021;5(3):94-98. DOI: 10.33545/gynae.2021.v5.i3b.909