Successful Vaginal Delivery in Spite of a Large Pelvic Neurofibroma

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Neurofibromatosis (NF) is an autosomal dominant genetic disorder and pregnancy in a patient with NF is associated with very poor perinatal outcome and life-threatening complications. In addition, the growth of pelvic tumors during pregnancy can be caused to dystocia and delivery is accomplished by cesarean section. We presented a case with a large pelvic nerve tumor not causing any dystocia or obstruction to the labor, allowing vaginal delivery

A 26 year old primigravida, diagnosed NF, had a large neurofibroma (about 8x9 cm diameter size) in pelvis infront of the sacrum. No significant change was detected in NF lesions. After an uneventfully antenatal follow-up, the patient gave a birth with vaginal delivery at term, even had a pelvic mass almost equal to the size of the fetal head causing no birth dystocia.

If not otherwise fetally or maternally indicated, initiation of labor and the course of labor should be observed and vaginal delivery should be attempted for the pregnant has a pelvic neurofibroma, before a decision for cesarean section is made.

Key Words: Neurofibromatosis, Pregnancy

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Introduction

Neurofibromatosis Type 1 (NF1, von Recklinghausen's disease, peripheral neurofibromatosis) is an autosomal dominant genetic disorder. The NF1 gene locates on chromosome 17q and the protein product termed neurofibromin acts as a tumor suppressor. Type 1 NF relates to gene abnormality. Mutations in the NF1 gene result in loss of function of neurofibromin, and this in turn results in increased proliferation and tumorogenesis in neurocutaneous tissues. It is characterized by numerous tumors affecting somatic, cranial and autonomic nerves.

The incidence of pregnancy among NF1 patients is relatively low and is inversely correlated with the severity of the disease; approximately 1/5.000 to 1/18.000 of obstetrical patients have NF1, compared with the 1/3.000 to 1/4.000 overall NF1 incidence.[1,2] However, Segal et al found the prevalence of NF1 as 1 in 2434 deliveries.[3]

The rate of spontaneous mutations in NF-1 is very high. Approximately 50% of cases of NF-1 result from de novo mutations, occurring for the first time in the family; NF is inherited as an autosomal dominant trait and penetration rate is almost 100% with variable expressivity and grades of phenotypic severity.[4] NF has markedly variable clinical expressivity, which is both inter- and intra-familial, with manifestations ranging from mild cutaneous lesions to severe cosmetic effects, orthopedic complications and functional impairment, and the risk of malignant degeneration of the tumors. The manifestations of NF may appear or worsen with age in affected individuals. This may explain the findings that none of the offspring of women with NF seemed to be affected at birth.

The diagnostic criteria for NF-1 are met in an individual if two or more of the following are present (others may develop over time): >6 café-au-lait spots (>15 mm in adults, >5 mm in children), two or more neurofibromas of any type or at least 1 plexiform neurofibroma, freckling in the axillary or inguinal regions, optic glioma, two or more Lisch nodules (iris hamartomas), a distinctive bony lesion and a first-degree relative with NF1 with the above criteria.[4]

The growth of pelvic tumors during pregnancy was reported to cause dystocia and delivery was accomplished by cesarean section. In this article, we reported a case with a large pelvic nerve tumor not causing any dystocia or obstruction to the labor, allowing vaginal delivery, and we discussed a pregnancy with NF1 in the light of the literature.

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Case Report

A 26-year old primigravid woman with 15 weeks of gestation according to last menstrual date presented for routine control. Physical examination revealed multiple café-au-lait spots (the largest 11 cm) and multiple neurofibromas (the largest 3 cm). A 5 cm scar was present at the gluteal region, which seemed to belong to a biopsy procedure performed 8 years ago and yielded neurofibroma. Obstetric ultrasonography revealed live pregnancy at 15 weeks of gestation as well as a 90x80 mm homogenous solid lesion with regular contours at the Douglas pouch. The tumor resembled a pedinculated uterine leiomyoma, ovarian mass or soft tissue sarcoma. Bimanuel examination revealed a firm, nontender, nonfixed mass in the posterior aspect of the pelvis in front of the sacrum behind the pregnant uterus. MRI used for the differential diagnosis of the mass detected a 8.5x6.5x7.cm soft solid tissue mass (neurinoma?) with regular contours, no adipose content and no invasion to the adjacent soft tissue, arising from the spinal canal at sacral 1-2 level and extending from the dilated right foramina into the presacral area. While on T1-weighted MRI the mass had heterogenous-hypointense images with higher intensity of the central portion than the periphery, on T2-weighted images the central portion had hypointensity and the periphery had higher signal intensity (Figure 1). The lesion seemed to be positioned at the rectouterine space and to extend to the posterior plane of the uterine cervix. Sections of images were not conclusive for the presence of fetal lesions.

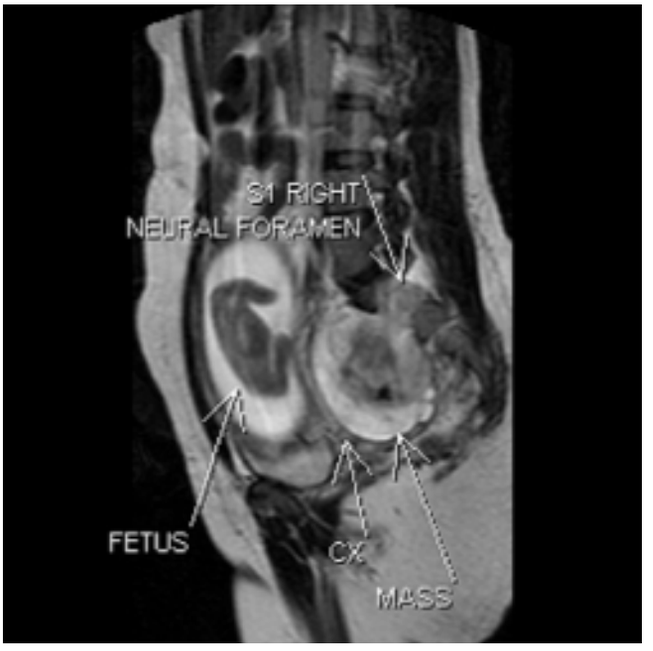

Figure 1: The magnetic resonance image of the pelvic neurofibroma and the fetus

The patient had a history of NF1 diagnosed both by histological findings and by diagnostic criteria on Neurofibromatosis. She also did not have a family history of NF.

Genetic counseling was given to the family. The 50% rate of transmitting the disease to offspring was emphasized, and they were informed on the possibility of developing complications during pregnancy and in the future life of the fetus. After the discussion, the parents opted to continue with the pregnancy.

The follow-up of pregnancy was uneventful. No significant change was detected in NF lesions and the pelvic mass. The patient had a 3250 g girl with vaginal delivery through normal labor process at 41 weeks +1 day of gestation with the pelvic mass almost equal to the size of the fetal head causing no birth dystocia. Blood pressure of the patient was normal throughout pregnancy and at pre- and post-partum periods. No complications were observed during the postpartum period. The newborn had no signs of NF1 on physical examination.

The patient who was recruited for the reevaluation of the mass in the postpartum period did not respond to this call and was lost to follow-up.

Oral informed consent was obtained from the patient for publication of this case report and accompanying image.

Discussion

Neurofibromas in NF1 manifest in several ways: cutaneous neurofibromas, subcutaneous neurofibromas, nodular plexiform neurofibromas and diffuse plexiform neurofibromas.

The skin lesions may increase in size and number during pregnancy and tend to regress following delivery.[5] A possible explanation for the puberty/pregnancy-associated tumor growth is direct or indirect (nerve growth factor-mediated) sex hormone stimulation.[6] Large neurofibromas can also arise from multiple nerves within plexuses, termed plexiform neurofibromas. Those type pelvic nerve tumors are capable of aggressive growth particularly during pregnancy.[2] The increased incidence of malpresentation and cephalopelvic disproportion can be partially attributable to these diagnosed or undiagnosed pelvic (possibly retroperitoneal) neurofibromas and pelvic contractures.

The growth of pelvic tumors during pregnancy was reported to cause dystocia and delivery was accomplished by cesarean section. Several case reports suggest that asymptomatic neural neoplasms arising from the pelvic nerve sheath and reaching the size of 7-11 cm (schwannoma, neurofibroma, neurofibrosarcoma) may cause dystocia by obstruction, thus complicating labor and requiring cesarean section.[7-10]

While no information on the frequency of pelvic neurinomas is present in cohorts in the literature,[2,3] only Dugoff and

Sujansky in their largest series of 105 women and 247 pregnancies, reported that 3 cases with pelvic neurinoma required cesarean section. The sizes of the so-called tumors designated as large neurofibromas protruding into the pelvis was not reported.[5]

Cesarean section may be difficult in NF patients since tumors arising from retroperitoneal autonomic plexuses may distort the normal anatomy of the retroperitoneum, bladder or vagina.

Otherwise, cephalopelvic disproportion due to the orthopedic deformities associated with pelvic bone contractions may cause dystocia during labor.[5] General physical and gynecologic examination of our patient that was performed to evaluate and exclude the cephalopelvic disproportion did not reveal any pelvic or spinal deformity.

Higher rates of maternal and neonatal complications were reported in the series including spontaneous abortion (18-21%),[2,5] hypertension/preeclampsia/eclampsia (31-70%),[3,11] IUGR (13-46%),[2,3] preterm labor (31-60%),[3,11] stillbirth (9-50%),[2,3,11] low-birth weight,[3] cesarean section due to obstetric and maternal complications of NF-1 (28-39%),[2,3,5] perinatal mortality (16%)[2] and maternal mortality (10%) directly caused by disease of NF-1 during pregnancy.

However, controversial information on neurofibromatosis and pregnancy also exists. Dugoff and Sujansky did not detect a higher rate of stillbirth, preeclampsia and preterm delivery, and subsequently concluded that common obstetrical complications were not more frequent in women with NF1 than in the general population, but there might be a subgroup of women with NF1 who could be at increased risk for obstetrical complications and might require close monitoring at a high risk obstetric center.[5] Nevertheless, Jarvis and Crompton did not observe any increased incidence of obstetric complications compared to the risk of the general population.[12]

NF patients can be safely carried to term if adequate monitoring is provided.[2] In our case, we did not observe any obstetrical complication.

Prenatal diagnosis of NF1 was not possible because of the nonspecific nature of the findings and because such disseminated disease is rare in the prenatal and neonatal periods. Requests for prenatal testing are limited because of the inability to predict disease severity and the phenotype of an individual.[13] It is difficult to provide parents with accurate data concerning specific NF1 complications. Also there is little evidence to support phenotype-genotype correlations in NF1. As the phenotype of NF1 is variable, it is difficult to predict the risks of complications in any individual. The risk of an individual with NF1 having a severely affected child is 8%.[13]

In this respect, NF1 provides a particular challenge in genetic counseling and will serve as a model for disease with high penetrance of the mutation, but extreme variability in the disease phenotype.

These tumors are rarely present at birth. The neonatal features of NF1 are usually solitary and cutaneous and therefore ultrasound alone cannot provide a specific prenatal diagnosis in most cases. MRI routinely used in the postnatal evaluation of affected individuals can provide additional useful information in the diagnosis of fetal NF1.

Currently, prenatal diagnosis of NF1 with MRI is still limited due to structural and technical difficulties. McEwing et al [14] presented a case diagnosed by MRI at 32 weeks of gestation after suspected lesions were detected with USG.

In conclusion, the pregnancy of a patient with neurofibromatosis should be regarded as high risk and requires ideally close antenatal monitoring at tertiary centers in order to detect early obstetric and maternal complications.[1-3] The presence of pelvic neurofibromas should be noted as they cause dystocia or malpresentation, necessitating a cesarean delivery.

In our case who delivered vaginally without any complication of pregnancy, it is notable that a 9 cm pelvic mass originating from the sacral neurons and was adjacent to the cervix did not cause dystocia.

In conclusion, if not otherwise fetally or maternally indicated, initiation of labor and the course of labor should be observed and vaginal delivery should be attempted before a decision for cesarean section is made. Cesarean delivery should be considered only if obstructed labor due to dystocia is present.

Büyük Pelvik Nörofibroma Rağmen Başarılı Vajinal Doğum

Nörofibromatosis (NF) otozomal dominant geçişli bir hastalıktır ve NF'li olgularda gebelik, olumsuz sonuçlar ve hayatı tehdit edici komplikasyonlarla birliktelik göstermektedir. Bununla birlikte, pelvisde bulunan tümörlerdeki büyüme distosiye, buna bağlı olarak da sezaryen ile doğuma neden olabilmektedir. Bu makalede amacımız büyük pelvik nörofibroma rağmen distosiye veya doğum eyleminin ilerlemesine engel olmadan başarılı vajinal doğum gerçekleşen bir olguyu sunmaktır.

26 yaşında, primigravid ve NF tanısı almış olan gebe olguda pelvisde sakrumun ön tarafında yaklaşık 8x9 cm çapında nörofibrom mevcut idi. Gebelik boyunca olgunun NF lezyonlarında bir değişiklik saptanmamıştı. Sorunsuz bir antenatal takip sonrasında, yaklaşık fetal başa benzer bir büyüklüğe sahip pelvik kitleye rağmen distosiye neden olmadan termde, vajinal yolla doğum gerçekleşti.

Pelvik nörofibroma sahip bir gebede başka bir nedenden dolayı fetal veya maternal endikasyon olmadığı durumda sezaryen ile doğum kararı verilmeden önce, doğum eyleminin başlaması ve ilerlemesi izlenmeli ve vajinal doğum denenmelidir.

Anahtar Kelimeler: Nörofibromatosis, Gebelik

References

1. Blickstein I, Lancet M, Shoham Z. The obstetric perspective of neurofibromatosis. Am J Obstet Gynecol 1988; 158:385-8.
2. Weissman A, Jakobi P, Zaidise I, Drugan A. Neurofibromatosis and pregnancy. An update. J Reprod Med 1993;38:890-6.
3. Segal D, Holcberg G, Sapir O, Sheiner E, Mazor M, Katz M. Neurofibromatosis in pregnancy. Maternal and perinatal outcome. Eur J Obstet Gynecol Reprod Biol 1999; 84:59-61.
4. Ferner RE, Huson SM, Thomas N et al. Guidelines for the diagnosis and management of individuals with neurofibromatosis. J Med Genet 2007;44:81-8.
5. Dugoff L, Sujansky E. Neurofibromatosis type 1 and pregnancy. Am J Med Genet 1996;66:7-10.
6. Roth TM, Petty EM, Barald KF. The role of steroid hormones in the NF1 phenotype: focus on pregnancy Am J Med Genet A 2008;146A:1624-33.
7. Andrews S, Capen CV. Pelvic schwannoma in pregnancy. A case report. J Reprod Med 1993;38:826-8.
8. Golan A. Retroperitoneal neural tumours obstructing labour: A report of 2 cases. S Afr Med J 1979;56:1163-6.
9. Griffiths ML, Theron EJ. Obstructed labour from pelvic neurofibroma. S Afr Med J 1978;53:781.
10. Baker VV, Hatch KD, Shingleton HM. Neurofibrosarcoma complicating pregnancy. Gynecol Oncol 1989;34:237-9.
11. Sharma JB, Gulati N, Malik S. Maternal and perinatal complications in neurofibromatosis during pregnancy. Int J Gynaecol Obstet 1991;34:221-7.
12. Jarvis GJ, Crompton AC. Neurofibromatosis and pregnancy. Br J Obstet Gynaecol 1978;85:844-6.
13. Ferner RE. Neurofibromatosis 1. Eur J Hum Genet 2007;15:131-8.
14. McEwing RL, Joelle R, Mohlo M, Bernard JP, Hillion Y, Ville Y. Prenatal diagnosis of neurofibromatosis type 1: sonographic and MRI findings. Prenat Diagn 2006; 26:1110-4.