MRI investigation for neurofibromatosis type 1 lesions during pregnancy - A case report

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Summary

Background: Neurofibromatosis type 1 (NF1) is a genetic disorder of the nervous system that causes tumor growth on the nerves, skin changes and bone deformities, and it is reported to be associated with adverse perinatal outcome in pregnant women.

Case: We report a case of MRI investigation of a pregnant woman with a known NF1 disease who showed exacerbation of skin lesions during pregnancy. No dangerous lesions that could compromise pregnancy outcome were detected, thus allowing the pregnancy to continue to term uneventfully.

Conclusion: MRI has strong indications for the follow-up of pregnant patients with neurofibromatosis as it can detect lesions that can serve as risk factors for pregnancy complications.

Key words: MRI; Neurofibromatosis type 1; Pregnancy.

Case report

A 30-year old primiparous woman was first seen at our institution at the 7th week of her first pregnancy. The woman was suffering from neurofibromatosus type 1 (NF1) and had undergone several surgical operations for skull bone deformity since childhood. She was not suffering from any other disease and was not on any medication. Her condition seemed to be in a steady phase without major complications at the time. She and her husband received extensive counseling regarding the risk of the fetus inheriting the disease and were fold that there was no way to predict the severity of the disease in the child. Both parents understood the risks but were determined to go on with the pregnancy, avoiding unnecessary interventions. Detailed physical examination, blood pressure and neurological tests were found normal. No scoliosis was found. First trimester screening for chromosomal abnormalities using nuchal translucency and biochemical markers showed an extremely low risk. The midtrimester anomaly scan was also normal. The patient was kept under close observation with regular monitoring for hypertensive complications. She had monthly ultrasound scans to monitor fetal growth rate and status of the placenta and amniotic fluid, as well as Doppler examinations of the fetal vasculature. She also had biweekly non-stress-tests starting from 32 weeks. Fortunately all the tests were normal and the patient seemed to have an uneventful pregnancy. A major concern of both the mother and the medical team involved in her care was that the skin lesions were exacerbated, especially after 26 weeks. Café au lait spots had increased in size and number from the late midtrimester. We could not possibly know whether this was associated with growth of internal neurofibromas in the patient's body and if this was the case, if this would affect the pregnancy or her general medical status. Ultrasonography was thought to be inadequate to examine the lower abdomen (due to presence of the gravid uterus) and the woman declined the alternative of a computed tomography (CT) for fear of irradiation causing damage to the fetus. We proposed magnetic resonance imaging (MRI) of the brain and the thorax (the locations of her neurofibromatoses before she became pregnant as confirmed by previous CT), as well as of the abdomen to check for the development of new lesions that could cause obstetrical complications. The brain MRI demonstrated postoperative alterations of the left cerebellum with encephalomeningocele and atrophy (Figure 1), bone deficit in the occipital region and other postoperative alterations as well as a soft tissue mass attributed to neurofibromatosis in the left parietal, temporal and parotitic area. MRI of the thorax and abdomen showed a pregnant uterus with a fetus in breech presentation corresponding to 29 weeks of gestation

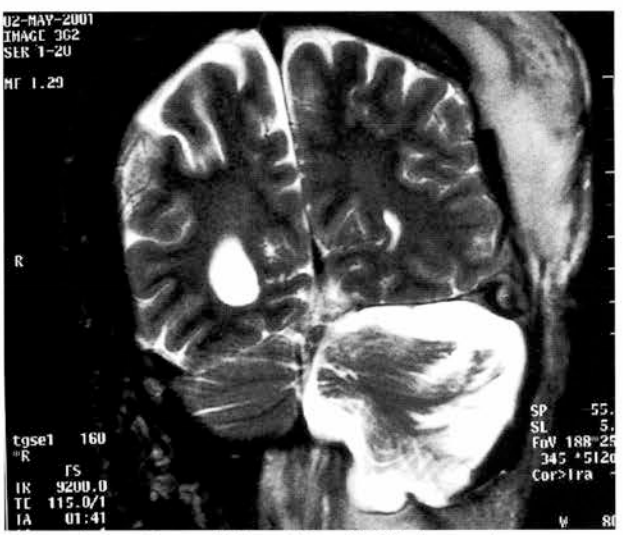

Figure 1. — Coronal T2-weighted TSE image demonstrates postoperative alterations in the left cerebellum with atrophy of the hemisphere and encephalomeningocele.

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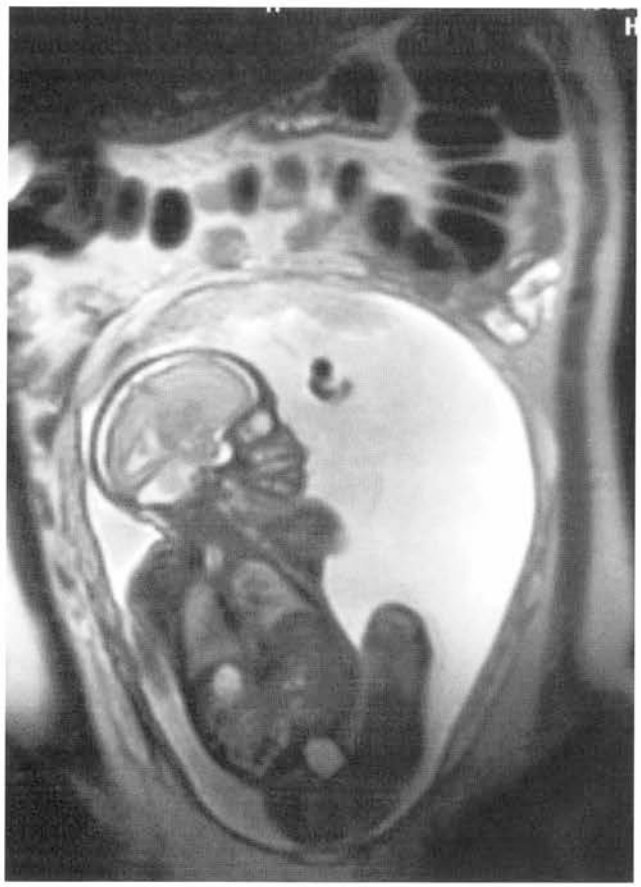

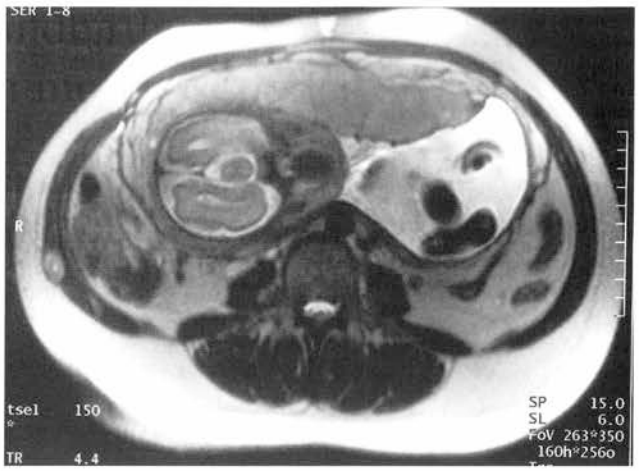

Fig. 2b

Figure 2a. — Coronal T2-weighted TSE image demonstrates the fetus corresponding to 29 weeks of gestation.

Figure 2b. — Axial T2-weighted TSE image. In the right abdominal wall there is a small neurofibroma.

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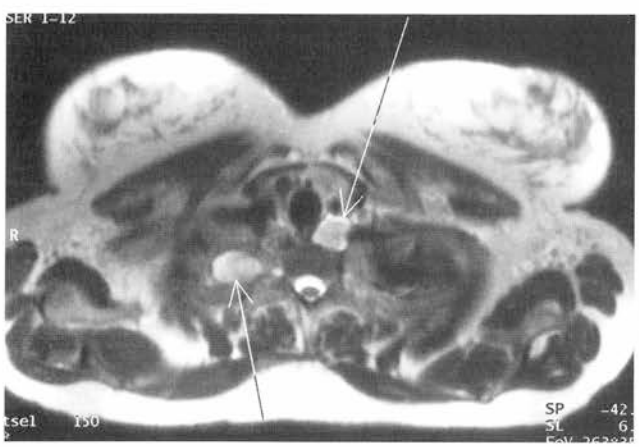

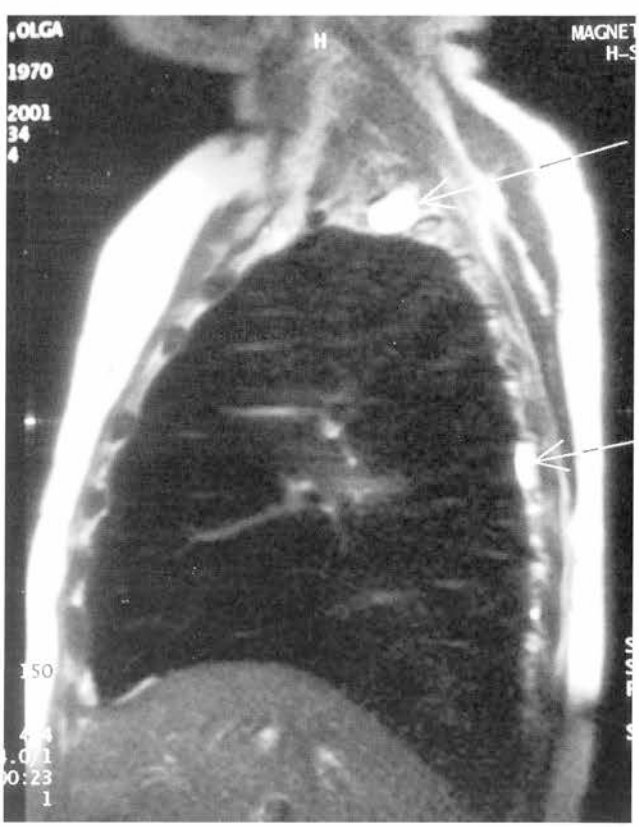

Fig. 4

Figure 3. — Axial T2-weighted TSE image displays the two cystic neurofibromas located in the apex of the right hemithorax and at the left of the trachea, posterior to the great vessels, respectively.

Figure 4. — Sagittal TSE T2-weighted image displays a small cystic intercostal neurofibroma on the right and a second one at the thoracic inlet.

(Figure 2a). There were two cystic neurofibromas in the apex of the right hemithorax and in the thoracic inlet posterior to the trachea and the great vessels on the left, respectively (Figure 3). Multiple smaller intercostals and intervertebral neurofibromas were also detected in the thoracic region without outside growth (Figure 4). There was also a small lesion in the right abdominal wall attributed to a neurofibroma as well (Figure 2b).

Since the MRI did not detect any new fibromatoses in the brain, the vessels or the pelvis, no additional measures were taken. The pregnancy was uneventful and the patient had a cesarean section at 38 weeks of gestation due to persistent breech presentation, and a live healthy female fetus weighing 3,050 g was born. The postoperative period was also uneventful and the patient and her newborn were discharged on the fifth postoperative day in good condition.

Discussion

In patients with neurofibromatosis fertility is unaffected and pregnancy is possible. Complications during pregnancy may have an impact on the fetus, causing intrauterine demise, mental retardation and body deformities, or on the mother causing hypertensive disorders, vasomotor manifestations of pheochromocytoma, aneurysm rupture, hemolysis, coagulation defects, or even disseminated intravascular coagulation and impaired liver function tests, resulting in intrauterine demise, intrauterine growth retardation, premature delivery and increased cesarean section rates [3-6].

A recent report has mentioned up to a 46% rate for intrauterine growth restriction, 23% stillbirth rate and higher cesarean section rate of 37% [4]. Dugoff and Sujansky presenting the pregnancy outcome of 105 women with NF 1 however, did not show the previously reported increased incidence of preeclampsia, preterm delivery, intrauterine growth restriction, pregnancy-induced hypertension, stillbirth, spontaneous abortion, or perinatal mortality [3]. Nevertheless, neurofibromas and café-au-lait spots are known to increase in size during gestation [5]. Sixty percent of the women reported by Dugoff and Sujansky showed growth of new neurofibromas during pregnancy and 52% noted enlargement of existing ones. Less than 20% observed no changes in the size of their neurofibromas and no growth of new ones during pregnancy in this study [3].

Vascular changes may predispose the patient to hypertension. Growth of central nervous system tumors can occur that require intervention. Patients with neurofibromatosis are known to have an increased risk of hypertension throughout their lives, especially those who have hypertension during pregnancy. It is possible that a process similar to that present in skin lesions occurs also in neurofibromatosis-associated vascular lesions. Such progression during pregnancy may be caused by a variety of known angioactive factors (placenta and other growth factors, tumor necrosis factor, angiogenin, interleukin-8, etc.) or the recently discovered vascular endothelial growth factor that has also been considered to play an important role in the regulation of angiogenesis including placental angiogenesis [5].

In our case there was great concern when the skin lesions of the patient were enlarged, because we did not know whether it was associated with an enlargement of internal neurofibromatoses located either within the pelvis potentially causing obstetric complications, or located elsewhere (in the brain or even vessels) which could cause severe problems such as aneurysm rupture or urgent medical complications. Ultrasonography would only be helpful in detecting large pelvic masses that could cause obstetric complications during delivery but could not inform us about growth of smaller masses and their relationship with the vessels, either in the abdomen or elsewhere. The patient was unwilling to be subjected to CT scan for the fear of irradiation. We then suggested MRI of the brain, thorax and abdomen, which were our major concerns. MRI has been proven efficient to evaluate neurofibromatosis lesions within the pelvis [7] and is considered safe during pregnancy [8]. MRI results were reassuring – no major problem had appeared in the pelvis or the vessels so the pregnancy was allowed to proceed with no additional tests and ended with a good perinatal outcome.

This case report shows that MRI has strong indications for the follow-up of pregnant patients with NF 1 as it can detect potentially dangerous lesions that serve as risk factors for pregnancy complications and adverse perinatal outcome. MRI in the areas of interest will help us determine the management plan. If no lesions are detected we can allow the pregnancy to proceed under the close observation already mentioned. Unless there is an obvious malformation of the pelvis from detected neurofibromas, a trial of labor is appropriate. If vessel lesions are detected on MRI, increased surveillance should be the rule and a surgeon's team informed and prepared for immediate intervention in the case of rupture.

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