

CASE REPORT

PREGNANCY WITH ELEPHANTIASIS NEUROFIBROMATOSA: WITH A GOOD MATERNAL AND FETAL OUTCOME: A CASE REPORT

R. Chauhan¹, Rowena Walker², B. Sahu³

HOW TO CITE THIS ARTICLE:

R. Chauhan, Rowena Walker, B. Sahu. "Pregnancy with Elephantiasis Neurofibromatosa: with a Good Maternal and Fetal Outcome: A Case Report". Journal of Evolution of Medical and Dental Sciences 2014; Vol. 3, Issue 23, June 09; Page: 6504-6508, DOI: 10.14260/jemds/2014/2772

ABSTRACT: Elephantiasis neurofibromatosis, most striking of NF-1 type of neurofibromatosis. We report a rare case of neurofibromatosis type-1 with elephantiasis of right lower limb, with full term pregnancy in labour. Patient underwent caesarean section. A healthy mother and baby was discharged, after detailed counselling about the fifty percent chances of transmission to the progeny, whereas the mother can have various complications like carcinoma breast, brain tumour, MSNT or leukaemia, etc. Hence regular follow up and screening was advocated, so that early detection of complications and effective treatment could be offered.

KEYWORDS: Elephantiasis neurofibromatosa, Pregnancy, Carcinoma breast, MSNT, CT scan.

INTRODUCTION: Neurofibromatosis (NF) is an autosomal dominant, phakomatosis or neurocutaneous syndrome affecting the neuroectoderm and the mesoderm. However, in 50% there is no family history, reflecting the incidence of new mutations. It is classified into two distinct types, neurofibromatosis 1 (NF1) the peripheral variety and neurofibromatosis 2 (NF2) the central one. The birth incidence of type 1 is One in 2, 500-3, 000 but may be slightly higher because of failure to diagnose milder cases. NF type 2 has a prevalence of about One in 25,000. The sex ratio is equal.¹ It appears to be more common in white races.

NF1, Von Recklinghausen disease characterizes²

- 5-6 or more café-au-lait spots (defined oval-shaped light brown patches greater than 0.5cm in diameter).
- Multiple neurofibromas (tumors on, under, or hanging off the skin).
- Freckling (under the armpits and areas of skin folds such as the groin).
- Lisch nodules (tiny tumors on the iris of the eye).
- NF often first appears in infants or during early childhood.



Café-au-lait spots



Neurofibroma

CASE REPORT

NF2, also known as bilateral acoustic neurofibromatosis, characterizes:³

- Multiple tumors, lesions on the brain and spinal cord.
- The tumors grow on the auditory nerves and lead to hearing loss; this is usually the first symptom of the disease. Often this is not apparent until the late teens or early twenties.

Another rarest variety of neurofibromatosis is Schwannomatosis characterized by intense pain. NF-1 is divided into three types, the most common being localized neurofibroma, the least common being diffuse neurofibroma and the most characteristic lesion of the disease being plexiform neurofibroma, exhibits a characteristic “bag of worms” appearance on gross examination and cross-sectional imaging because of the involvement of the long segment of a major nerve trunk and its branches.

This lesion, in its extreme form, may involve an entire extremity, with gigantic hypertrophy of the skin, soft tissues and the underlying skeleton. They may become very large and deformed, therefore named as “elephantiasis neuromatosa” by Virchow¹.

CASE REPORT: Mrs. G, 21 years old married, Primigravidae, unbooked, residing at a remote village about 150kms from medical college was admitted, with amenorrhea of nine months. She also had multiple neurofibromatosis and Elephantiasis right leg since childhood. Patients-general condition was fair, afebrile, pulse 90 per minute regular, pallor- Absent, BP-110/80 mm of Hg, height 156cms; body weight 103kgs. Right lower extremity, buttocks, perineal region and waist presented with elephantiasis.

Visible dark brown pigmentation were present all over the body with multiple soft nodule of the diameter of 3 to 40 mm, present on the neck, chest, back, both arms, and left lower limb. The patient started developing the generalized Café-au-lait spots, starting to arise first from the abdomen by the age of about 5years, followed by small generalized, neurofibromatotic lesions. Patient also noted a painless swelling on her calf that progressed distally and proximally to involve the hip joint.

By the age of about 13years, she had marked swelling with-out pain leading to marked disfigurement. Elephantiasis in right limb extending up to the hip joint the largest circumference of the right thigh was 102 cms and the right calf was 110 cms. The left lower limb was normal.

The circumference of the left thigh (precisely at 10 cm above the knee joint) was 35cms, and the largest circumference of the lower left calf was 28cms, lungs clear, with no cardiac anomaly. Family history: Patient's father has Café-au-lait spots with diffuse neurofibromatosis but no enlargement of any area or limbs. Per Abdomen examination –Uterus full term, cephalic presentation, fetal heart sound and regular 140 per minute.

Per vaginal examination – Cervical os 3-4 cm dilated and 50-60 percent effaced, vertex at -3 station, membrane present early bag forming both sidewalls were convergent. Routine investigations showed the blood cell count as well as the liver function tests and glucose level in the normal range. The patient was AB+ positive and tested negative for hepatitis A and B as well as for acquired immune deficiency syndrome and VDRL.

Ultrasonography of right limb [Grey scale]:

Longitudinal Scan: Multilobulated tortuous tumors, oriented along the long axis of the leg.
Neurofibroma: The Target sign seen.

CASE REPORT

Plain Computed tomography (CT) T2W/STIR:

Bone Window: Large periosteal defect giving way to infiltrating mass lesion.

Axial – Periosteal / endosteal thickening.

Sagittal – Cortical erosion at talo calcaneal joint.

Tissue Window: Thick wavy cords of low attenuated masses traversing the soft tissue and giving a reticular network appearance of right leg.

Color Doppler: B-mode ultrasound imaging of the right lower limb revealed multiple, non-uniform echo lumps, although pathologic results were unconfirmed. In addition, the right tibio-fibular periosteum was intermittent, which was consistent with lumps in the lower limb invading the periosteum.

Patient under- went a cesarean section due to the contracted pelvis. She delivered a full term alive, female baby of 2.8 kg, by vertex, the whole procedure was uneventful. Stitches removed on the 8th day, stitch line was healthy. She was discharged after counseling about the progression and rare chances of cancerous development of the elephantiasis neurofibromatosa of right limb, development of breast cancer, transmitting the genes and affecting 50% of the progeny.

DISCUSSION: Common complications of NF1 include:

- Neurological problems. Learning and thinking (cognitive) difficulties occur in up to 60 percent. Uncommon complications include epilepsy, stroke and hydrocephalus.⁴
- Cosmetic. Visible signs of neurofibromatosis-such as extensive cafe au lait spots, numerous nerve tumors (neurofibromas) in the facial area or large neurofibromas- can cause anxiety and emotional distress, even if they're not medically serious.
- Skeletal problems. Some children have abnormally formed bones, which can result in bowing of legs, scoliosis, decreased bone mineral density and fractures that sometimes don't heal.
- Visual difficulties. Occasionally in children, optic pathway glioma grows on the optic nerve leading to visual problems.
- Increase in neurofibromas. Hormonal changes associated with puberty, pregnancy or menopause may cause an increase in neurofibromas. Most women with NF1 have healthy pregnancies but will likely need monitoring by an obstetrician familiar with NF1, in addition to their NF1 specialists.
- Cardiovascular problems. An increased risk of high blood pressure and, rarely, blood vessel abnormalities.⁴
- Cancer. Less than 10 percent of people develop malignant peripheral nerve sheath tumor (MPNST). Breast cancer, leukemia, brain tumors and some types of soft tissue cancer.
- Monitor neurofibromas vigilantly for any change in appearance, size or number. Changes may indicate cancerous growth. The earlier a malignancy is detected, the better the chances for effective treatment.

NF1 is caused by a mutation on a gene located on chromosome17 and NF 2 on chromosome 22. NF1 has a marked effect on the lives of affected subjects and their families. The psychosocial areas of greatest concern were, the difficulties faced at school, and later in accepting the cosmetically disfiguring aspects of the condition. Women with neurofibromatosis 1 are at a moderately increased risk of developing breast cancer and should be considered for early screening.

CASE REPORT



Pictures showing the hypertrophied right limb with normal left limb with generalized Café-au-lait spots and neurofibromatosis



Marked difference in the size of the right and left foot seen

REFERENCES:

1. Ferner RE, Huson SM, Thomas N, et al; Guidelines for the diagnosis and management of individuals with neurofibromatosis 1. J Med Genet. 2007 Feb; 44 (2): 81-8. Epub 2006.
2. Friedman, J.M. (2009). Neurofibromatosis 1. Gene Reviews. University of Washington at Seattle.
3. Evans, D.G. (2009). Neurofibromatosis 2. Gene Reviews. University of Washington at Seattle.
3. NIH. Neurofibromatosis. Conference statement. National Institutes of Health Consensus Development Conference. Arch Neurol. May 1988; 45 (5): 575-8. [Medline].
4. National Institutes of Neurological Disorders and Stroke (NINDS). (2009). Neurofibromatosis fact sheet.

CASE REPORT

AUTHORS:

1. R. Chauhan
2. Rowena Walker
3. B. Sahu

PARTICULARS OF CONTRIBUTORS:

1. HOD, Department of Obstetrics and Gynaecology, NSCB Medical College, Jabalpur, M. P.
2. Senior Resident, Department of Obstetrics and Gynaecology, NSCB Medical College, Jabalpur, M. P.
3. Assistant Professor, Department of Obstetrics and Gynaecology, NSCB Medical College, Jabalpur, M.P.

NAME ADDRESS EMAIL ID OF THE CORRESPONDING AUTHOR:

Dr. Rowena Walker,
#1330, Diwanbada,
P. O-Westland, Khamaria,
Jabalpur-482005, M. P.
E-mail: rowe_eagle@yahoo.com

Date of Submission: 07/05/2014.
Date of Peer Review: 08/05/2014.
Date of Acceptance: 26/05/2014.
Date of Publishing: 07/06/2014.