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Case Study

NEUROFIBROMATOSIS TYPE 1 (NF1): A CLASSIC CASE PRESENTATION AND ITS AYURVEDIC UNDERSTANDING

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ABSTRACT

Neurofibromatosis type 1 (NF1), is an autosomal dominant tumor-predisposition disorder that develops from loss-of-function variants in the tumor-suppressor gene neurofibromin 1 called NF1 gene with a prevalence of 1 in 2,000–3,000 live births caused by mutations in the NF1 gene on chromosome 17. Cardinal features of NF1 include multiple café-au-lait spots, benign neurofibromas, and Lisch nodules of the iris. In addition to these hallmark features, NF1 exhibits a wide spectrum of clinical manifestations, including cutaneous, neurological, skeletal, and other systemic abnormalities. This case study examines a 19-year-old female patient who presented with multiple café-au-lait spots, axillary freckling, Lisch's nodules and 3 cutaneous neurofibromas. She also reported a family history of NF1, with her father exhibiting similar symptoms and diagnosed with NF1. The patient's neurological evaluation revealed no significant deficits, but eye examination identified the presence of Lisch nodules. The diagnosis of NF-1 was made according to the presence of two or more diagnostic criteria of the National Institute of Health 1987. This case highlights the characteristic clinical features of NF1, including café-au-lait macules, axillary freckling, cutaneous neurofibromas, Lisch nodules, and a positive family history, fulfilling the NIH 1987 diagnostic criteria. From the Ayurveda perspective, several fundamental concepts provide the base to understand the neurofibromatosis, such as *Anukta Vyadhi*- the group of diseases that are not directly mentioned in the classical texts; *Aadibalapravritta Vyadhi*- diseases of hereditary nature; and *Arbuda*- tumors/malignant growths. These concepts provide a strong outline for conceptual understanding of neurofibromatosis through Ayurveda perspective.

INTRODUCTION

Neurofibromatosis type 1 (NF1), is an autosomal dominant tumor-predisposition disorder that develops from loss-of-function variants in the tumor-suppressor gene neurofibromin 1 called NF1 gene with a prevalence of 1 in 2,000–3,000 live births caused by mutations in the NF1 gene on chromosome 17 with a prevalence of 1 in 2,000–3,000 live births caused by mutations in the NF1 gene on chromosome 17.[1] This gene encodes neurofibromin, a protein that helps regulate cell growth. NF1 is caused by either de novo or inherited, on the NF1 gene. Nearly half of all NF1 cases are hereditary in origin, while remaining cases result due to de novo mutations.

Cardinal features of NF1 include multiple café-au-lait spots, benign neurofibromas, and Lisch nodules of the iris. Other common features include learning disabilities, mild shortness of stature, and skeletal abnormalities.[2] There is a significantly increased risk of certain cancers, including female breast cancer <50 years, malignant peripheral nerve sheath tumors (MPNSTs), and brain tumors. There are 3 distinct types of neurofibromatosis: type 1 (NF1), type 2 (NF2), and schwannomatosis. NF1 and NF2 are the most common, while schwannomatosis is rare.[3] NF1 accounts for approximately 96% of all neurofibromatosis cases. In the present case, the patient also demonstrated NF1. It occurs equally among both genders and races. The disorder is

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associated with an 8-15 year reduction in average life expectancy in both men and women, primarily due to malignant neoplasms and cardiovascular causes. [4]

Case Report

An unmarried, 19-year-old female Hindu patient visited the outpatient department (OPD) of Kayachikitsa department of the Institute of Teaching and Research in Ayurveda (ITRA), Jamnagar, Gujarat, on 09/05/2024 with the complaints of multiple hyperpigmented macules, which were present all over the body, and 2-3 palpable masses.

History of Present Illness

The patient presented with a history of multiple hyperpigmented macular lesions, initially observed on the right arm and face during childhood. Over the preceding 3–4 years, these lesions had progressively increased in number and distribution, involving the bilateral hands, back, face, and lower limbs. At the age of 13 years, she developed two painless cutaneous neurofibromas, located on the right arm and left side of the chest, followed by the appearance of another painless neurofibroma in the left loin region. Fifteen days prior to presentation, she noticed a new palpable nodule over the forehead and near the left eye. The patient reported having consulted homeopathic and allopathic practitioners for these complaints before presenting to the Ayurvedic outpatient department. A positive family history was noted, as her father had similar clinical manifestations suggestive of NF1, with a reportedly similar pattern of disease progression.

Physical Examination

Dermatological Findings

- Hundreds of hyperpigmented skin macules on the chest, back, abdomen, B/L UL, B/L LL, and face (Figure 1).
- Multiple café-au-lait spots with diameter >1.5 cm (Figure 2).
- Axillary freckling (Figure 3).
- Cutaneous neurofibroma present over the right arm, left loin region and near right axilla. (Figure 4).



Figure 1- Multiple café-au-lait spots over Abdomen (Lt.) and back (Rt.)



Figure 2- café-au-lait spots with diameter >1.5 cm




Figure 3- Axillary Freckling





Figure 4: Cutaneous Neurofibroma over right arm, left loin region and near Rt. axilla

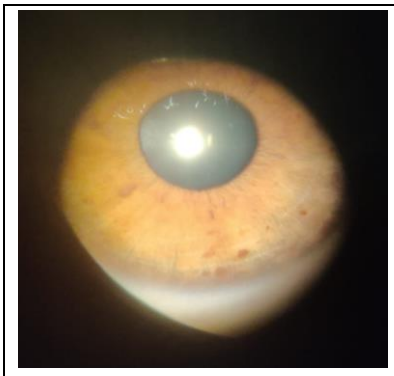
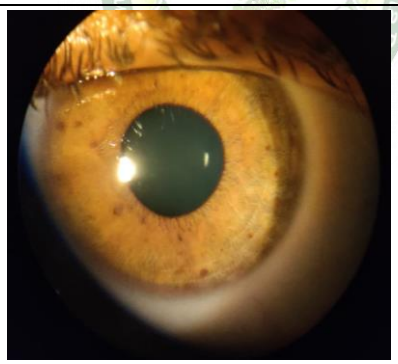



Figure 5: Lisch's nodules (iris hamartomas)



Figure 6: Plexiform Neurofibroma on hand (Father, Proband of NF1 in family)

Ocular Findings- Lisch's nodules (iris hamartomas) on the iris of both eyes were present and visualized on slit lamp examination without clinical visual impairment. (Figure 5)

1. Musculoskeletal Examination- No evidence of scoliosis, kyphosis, long-bone bowing, limb length discrepancy, chest wall deformity, or other NF1-associated skeletal abnormalities was noted.
2. Neurologic Manifestations- The patient demonstrated normal motor function, with age-appropriate intellectual and learning abilities. No behavioral disturbances or cognitive deficits were observed during the clinical assessment.
3. Laboratory Findings- Standard laboratory test values were in the normal range.
4. Vitals: The vital parameters were within normal physiological limits, with a pulse rate of 78 beats/min, blood pressure of 124/78 mmHg, respiratory rate of 22 breaths/min, and body temperature of 37.5°C.

Clinical Diagnosis- Clinical diagnosis of NF-1 was made based on the presence of multiple characteristic clinical features in accordance with the NIH diagnostic criteria (Table 1). The patient fulfilled more than two diagnostic criteria, including multiple café-au-lait macules, axillary freckling, multiple cutaneous

neurofibromas, bilateral Lisch nodules, and a first-degree relative with clinically diagnosed NF1. Molecular genetic testing was not performed.

Table 1: Diagnostic criteria for neurofibromatosis-1 (NF1) [5]

Patients had two or more of following symptoms:
a) 6 or more café-au-lait macules (>0.5 cm in children of >1.5 cm in adults)
b) 2 or more cutaneous or subcutaneous neurofibromas, one plexiform neurofibroma
c) Axillary or groin freckling
d) Optic glioma
e) 2 or more Lisch nodules
f) Sphenoid wing dysplasia or bowing of long bone (those of or those of pseudarthrosis)
g) First-degree relative with an NF1 diagnosis

In 2021, an international consensus group updated the original NIH criteria to incorporate genetic testing and advanced ophthalmological imaging, primarily to improve diagnostic accuracy and prevent misdiagnosis in young children. [6]

However, the 2021 Revised Criteria are unnecessary in the present case. Because this patient fulfills five distinct clinical features, far exceeding the minimum diagnostic threshold, there is no diagnostic ambiguity. Consequently, the original 1987 NIH diagnostic criteria remain entirely sufficient for establishing a definitive diagnosis. But, in the present case, the patients fulfill five distinct criteria, far exceeding the minimum requirement and hence not having diagnostic ambiguity. So, the 2021 criteria is not necessary in this case. The original 1987 NIH diagnostic criteria are sufficient.

Family History

The patient had a positive family history of NF1. Her father was diagnosed with NF1 and is the proband in the family. He exhibited clinical features consistent with the disorder, including multiple plexiform neurofibromas and café-au-lait macules. The occurrence of similar manifestations in the patient's father supports the hereditary nature of the disease and is consistent with the autosomal dominant pattern of inheritance characteristic of NF1. Moreover, her father was diagnosed clinically as having NF1, not genetically confirmed.

- Father's age- 55 yrs.
- Age at onset of café-au-lait macules- 9 yrs.
- Age at onset of multiple plexiform neurofibromas- 20 yrs.

DISCUSSION

In the present case, the patient exhibited several characteristic clinical features of NF1, including multiple café-au-lait macules, axillary freckling, multiple cutaneous neurofibromas, and bilateral Lisch nodules. According to the diagnostic criteria proposed by the National Institutes of Health Consensus Development Conference, the presence of two or more specific clinical findings confirms the diagnosis of NF1. The patient fulfilled this criterion, thereby supporting the clinical diagnosis. The presence of similar clinical manifestations in the patient's father further supports a hereditary pattern consistent with the autosomal dominant inheritance of NF1. The patient's father is the proband in the family, being the first affected individual to receive the diagnosis of NF1.

The chronological progression of the disease in this patient also corresponds with the natural history of NF1. Pigmentary lesions such as café-au-lait macules are usually the earliest manifestation and may appear during infancy or early childhood, whereas neurofibromas typically develop during adolescence or after puberty. In this case, macular hyperpigmentation was present since childhood, while neurofibromas appeared later during adolescence and gradually increased in number and distribution over time. Moreover, the patient did not present with any behavioral disturbances or cognitive deficits or NF1-associated skeletal abnormalities.

Ayurveda Approach to Understand Neurofibromatosis

From Ayurveda perspective, several fundamental concepts provide the base for understanding neurofibromatosis (NF). Because NF may not have a direct, single equivalent condition in ancient texts, it can be clinically approached through the triad of Ayurveda principles, i.e., *Anukta Vyadhi* (untold diseases), *Aadibalapravritta Vyadhi* (hereditary or genetically transmitted diseases), and *Arbuda* (tumour-like growth).

Understanding Neurofibromatosis through the Ayurvedic Concept of *Anukta Vyadhi*

Various scattered references regarding *Anukta Vyadhi* are available in the classical texts of Ayurveda. *Acharya Charaka* has stated that one should not be ashamed of one's inability to name a disease, since all disorders cannot be given standard names.[7] Diseases are innumerable, as the same vitiated *Dosha* can produce different disorders depending on variations in their etiology and site of manifestation. *Acharya Charaka* further elaborates the line of management for *Anukta Vyadhi*, stating that treatment should be initiated only after a thorough understanding of the

disease (*Vikara Prakruti*), including its pathogenesis (*Adhisthan*), location, and causative factors (*Samutthan*).[8]

Rather than limiting a disease to a particular name, this concept encourages the physician to understand the condition through its clinical features, causative factors, site of manifestation, and underlying pathogenesis. Moreover, based on the principles of *Anukta Vyadhi*, the treatment of NF1 may be planned after a comprehensive clinical assessment of the nature and pathogenesis of the disease, the site of manifestation, and the involvement of *Dosha* (functional regulatory principles), *Dushya* (vitiated bodily tissues), *Srotas* (circulatory pathways), *Agni* (digestive and metabolic capacity), and *Rogamarga,* etc.

Understanding of Neurofibromatosis as a Genetic Disease

NF1 is indeed a genetic disorder caused by a mutation in the NF1 gene on chromosome 17, it can be explained through Ayurvedic concepts such as *Beeja* (sperm and ovum), *Beeja Bhaga* (gene), and *Beeja Bhagavayava* (chromosome). *Acharya Charaka* stated that vitiation of *Doshas* in these specific structural components results in congenital or hereditary anomalies in the developing foetus.[9] Similarly, the concept of genetic disease is described by *Acharya Sushruta* in his classification of diseases.[10] He classified diseases into seven major categories, one of which is *Aadibalapravritta Vyadhi*. As mentioned in the following *Shloka*, this category refers to disorders that arise due to hereditary or congenital factors. [11]

तत्र, आदिबलप्रवृत्ता ये शुक्रशोणितदोषान्वयाः कुष्ठार्शःप्रभृतयः;

तेऽपि द्विविधाः- मातृजाः, पितृजाश्च |

This means *Aadibalapravritta Vyadhi* are those that arise due to defects in the reproductive elements (*Sukra* and *Shonita*) of the parents, such as *Kushtha* (skin disorders), *Arsha* (hemorrhoids), and similar conditions. These are further classified as maternal (*Matruja*) and paternal (*Pitruja*) in origin. This description reflects the Ayurvedic concept of hereditary disorders transmitted through the parental reproductive elements. It can be correlated with modern genetic diseases such as NF1, an autosomal dominant disorder caused by pathogenic variants in the NF1 gene that are inherited from either the mother or the father.

An Ayurvedic Interpretation of Neurofibromatosis through the Concept of *Arbuda*

From an Ayurvedic perspective, neurofibromas, one of the characteristic features of NF-1, exhibit clinical similarities to *Arbuda*. Both present as deep-seated, firm, rounded masses with a slow and progressive pattern of growth. Based on these similar clinical features, neurofibromatosis can be understood as the tumor-like growths seen in *Arbuda* characterized by *Vrittam* (rounded shape), *Sthiram* (firm or stable), *Mandaruja* (mild pain), *Mahantam* (progressively enlarging mass), and *Chiravriddhi* (slow growth over time) resulting from the vitiation of the *Kapha dosha* along with the involvement of *Mamsa Dhatu*. [12] These characteristics of *Arbuda* resemble those of plexiform neurofibromas, a type of neurofibroma.

***Dosha–Dushya* Involvement in Neurofibromatosis Type 1**

In terms of pathogenesis, the clinical features of NF1 may be interpreted primarily through the imbalance of *Vata Dosha*, which governs neurological functions. Vitiation of *Vata* may lead to abnormal growth patterns, neuromuscular involvement, and progressive tissue changes. Additionally, the involvement of *Kapha* may contribute to the formation of nodular masses, while *Rakta Dushti* may explain pigmentary abnormalities such as café-au-lait macules. Thus, the disease process may represent a combined disturbance of *Vata*, *Kapha*, and *Rakta* at the level of neurons.

Thus, the interpretation of NF1 through the Ayurvedic concepts discussed above emphasizes the relevance of correlating this hereditary disorder with classical Ayurvedic entities such as *Aadibala Pravritta Vyadhi*, *Anukta Vyadhi*, and *Arbuda*. Such an integrative approach may contribute to a broader understanding of NF1 while also offering insights into its prognosis, severity, and disease progression. Furthermore, it may help in developing appropriate therapeutic strategies by bridging classical Ayurvedic perspectives with contemporary understanding of hereditary disorders.

CONCLUSION

Neurofibromatosis type 1 (NF1) is a hereditary disorder with variable clinical manifestations observed in different patients according to the type and stage and severity of disease. In the present case, the diagnosis of NF1 was established based on characteristic clinical features and a positive family history.

Three concepts discussed above can be viewed as addressing different aspects of the NF1 from an Ayurvedic perspective. *Anukta Vyadhi* helps in approaching a disease for which there is no direct equivalent in the classical texts, while *Aadibala-pravritta Vyadhi* provides a conceptual basis for understanding its hereditary or genetic origin. The characteristic tumor-like lesions of NF1 may, in turn, be discussed in relation to *Arbuda* based on their clinical features. Thus, rather than considering these concepts as interchangeable, they can be understood as offering different perspectives on the disease- its clinical interpretation, its origin, and the nature of its lesions.

Limitations: As this is a single case report, restricting the generalizability of the findings. The absence of molecular genetic testing, radiological investigations, and histopathological examination limits further characterization of the condition and associated nodular lesions. The Ayurvedic perspective presented here is an attempt to understand the clinical manifestations through classical Ayurvedic concepts and does not imply a direct equivalence between NF1 and any specific Ayurvedic disease. Further studies involving more patients and long-term follow-up are needed to explore this approach in greater detail.

Informed Consent: Written informed consent was obtained from the patient for publication of her clinical details and photographs.

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