

# Case Report: 8 years old boy with neurofibromatosis type one and Alopecia from KING ABDULAZIZ MEDICAL CITY OF THE NATIONAL GUARD. Saudi Arabia

Dr. Ahmed Moosa Yahya assery

Paediatrics ambulatory care consultant. King Abdulaziz Medical City, National Guard Medical Affairs, Family Medicine- Ambulatory care, NGCSC

DOI: <https://doi.org/10.5281/zenodo.7578973>

Published Date: 28-January-2023

---

**Abstract:** <sup>1</sup>Neurofibromatosis (NF) is a group of genetic disorders: NF1, NF2 and schwannomatosis (shwon-oh-ma-toe-sis).NF1 (also known as von Recklinghausen disease) is the most common type, with a prevalence of 2.500 to 3.000 .NF1 is a variable disorder, which means that it can affect children in many different ways. It usually causes freckles or spots, most often on the skin or eyes, and benign (not cancer) swelling or lumps around the nerves. Many children with NF1 have no or very few medical problems, but about four in 10 children will have some complications. Treatment for NF1 depends on the medical problems your child develops.NF1 is something you are born with, and it is not contagious. Some of the signs of NF1 do not appear until later in life, often around puberty. in such current case I faced here two unusual associating disorders with NF1- according to many references I just reviewed-presented in form of (almost rare) hair disorder called trichothiodystrophy (TTDY) and the other one is a rare ophthalmological problem presented as a retinal atrophy (which I will not focus in it so deep) that motivated me to present such interesting case.

**Keywords:** KingAbdullAziz Medical City: KAMC, Neurofibromatosis Type One: NF1, Neurofibromatosis Type two:NF2, Trichothiodystrophy: TTDY, HELLP: Homolysis-Elevated liver enzymes and low platelets, National Guard Comprehensive Specialized Clinics: NGCSC, NO: Number, Intelligence Quotient: IQ, café au lait: CAL.

---

## 1. INTRODUCTION

(NF-1): <sup>2</sup>is a genetic disease that predisposes to malignancies in many parts of the body, especially the in central nervous system. It is the most common form of neurocutaneous diseases and has an autosomal dominant pattern of inheritance with a reported incidence of 1/3000 to 1/4000 NF-1 has a wide clinical spectrum and can affect almost any organ. The disease might present with different clinical pictures in affected family members, and some of the findings might appear with increasing age. Therefore, it is a challenge to diagnose and follow-up for both patients and clinicians. The most common clinical features are café au lait (CAL) spots, axillary/inguinal freckling, neurofibromas, and Lisch nodules.

The children with NF-1 could admit to different departments with a wide range of symptoms like skin lesions, visual problems, seizures, learning difficulties, growth retardation, endocrine disorders, and unexplained hypertension. Although it has familial characteristics, the course and severity of the disease can be quite different within the same family. The diagnosis and follow-up of these children should be pursued with a multidisciplinary approach.

**Signs and symptoms of NF1:**

NF1 can affect many different organs in the body in very different ways, but some features of the condition are more common. The major features of NF1 include:

- **Café au lait spots:** These are flat coffee-coloured patches on the skin. It's common to have one or two of these spots, but people with NF1 always have six or more of these birthmarks, which are always present before the age of five years. The number of cafés au lait spots is not related to the severity of the disease and they cause no problems or symptoms.
- **Freckling:** People with NF1 often have freckles in unusual places such as the armpit or the groin. They also tend to appear later (between ages three and five). Skin freckles are smaller than café au lait spots.
- **Neurofibromas:** These are benign swellings around nerves, which are usually seen as small lumps in or under the skin. Neurofibromas are not normally seen in young children but tend to increase in number and size around puberty and in adult life. They are usually small and generally cause no physical symptoms.
- **Lisch nodules:** These are small freckles within the iris (the coloured part of the eye). Lisch nodules develop by puberty in people with NF1. They are usually seen only on examination with a special lamp, and never affect vision or cause other symptoms.

Other common features of NF1 include:

- learning difficulties, which may cause some problems at school.
- headaches, which are more common in children with NF1 than in the general population.

Rarely, children with NF1 may have other complications, such as:

- high blood pressure (hypertension)
- curvature of the spine (scoliosis)
- bone problems (seen in the first two years of life)
- large benign nerve tumours (called plexiform neurofibromas – these are usually present at birth)
- spine and brain tumours that are usually benign.
- speech delay
- epilepsy.

**Criteria of diagnosing NF1:**

It's important to know that an accurate diagnosis of NF1 can only be made by a physician with expertise in the diagnosis and treatment of neurofibromatosis. Because people with NF1 often have multiple café-au-lait spots, the disorder should be suspected in someone with six or more spots, at least five millimetres in size before puberty and 15 millimetres in size after puberty. There is no relationship between the number of café-au-lait spots and the severity or prognosis of the disorder.

While the presence of café-au-lait spots is one criterion for the diagnosis, at least two features of the disorder need to be present to confirm the condition. A diagnosis of NF1 is made through an extensive physical examination and, in some cases, biopsies, imaging studies, or additional medical tests to confirm the presence of any of the following diagnostic criteria of NF1 established by the National Institutes of Health (NIH) in 1987 based on a consensus of experts in the field.

- Six or more **café-au-lait spots** measuring at least 5 mm in size before puberty and 15 mm in size after puberty
- **Freckles** in the axilla (under the arms) or groin
- Two or more **neurofibromas** or one **plexiform neurofibroma** (tumour involving multiple nerves)
- **Lisch nodules** on the iris of the eye

- **Optic glioma**

- Specific skeletal abnormalities such as **tibial dysplasia** (bowing of shin bone) or **abnormality of the orbit**
- NF1 by above criteria in a parent, sibling, or offspring

(TTDY): <sup>3</sup>Trichothiodystrophy, which is commonly called TTD, is a rare inherited condition that affects many parts of the body. The hallmark of this condition is brittle hair that is sparse and easily broken. Tests show that the hair is lacking sulphur, an element that normally gives hair its strength.

The signs and symptoms of trichothiodystrophy vary widely. Mild cases may involve only the hair. More severe cases also cause delayed development, significant intellectual disability, and recurrent infections; severely affected individuals may survive only into infancy or early childhood.

Mothers of children with trichothiodystrophy may experience problems during pregnancy including pregnancy-induced high blood pressure (preeclampsia) and a related condition called HELLP syndrome that can damage the liver. Babies with trichothiodystrophy are at increased risk of premature birth, low birth weight, and slow growth. about half of all people with trichothiodystrophy have a photosensitive form of the disorder, which causes them to be extremely sensitive to ultraviolet rays from sunlight. They develop a severe sunburn after spending just a few minutes in the sun. However, for reasons that are unclear, they do not develop other sun-related problems such as excessive freckling of the skin or an increased risk of skin cancer. Many people with trichothiodystrophy report that they do not sweat.

## 2. OUR CASE PRESENTATION

This is 8 years old boy who has been seen more than 10 times in our NGCSC by my colleges before he attended my clinic and since of the big numbers of the patients we are managing daily nobody gave a concern of the father inquiries about his son that were concentrated in major three thing, First one was about the black extending skin spots all over the body (Figure No1-A, B,C)



(Figure No 1-A: café-au-lait spots ).



(Figure No 1-B: Another café-au-lait spots ).



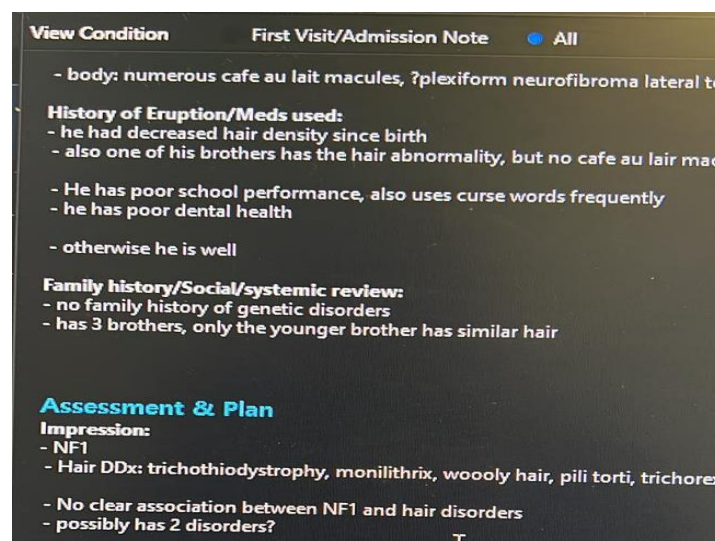
(Figure NO 1-C: Freckles in the axilla).

The second inquiry was about the ingrowing brittle hair his son was having since birth (Figure NO 2):



(Figure NO 2: Alopecia totalise):

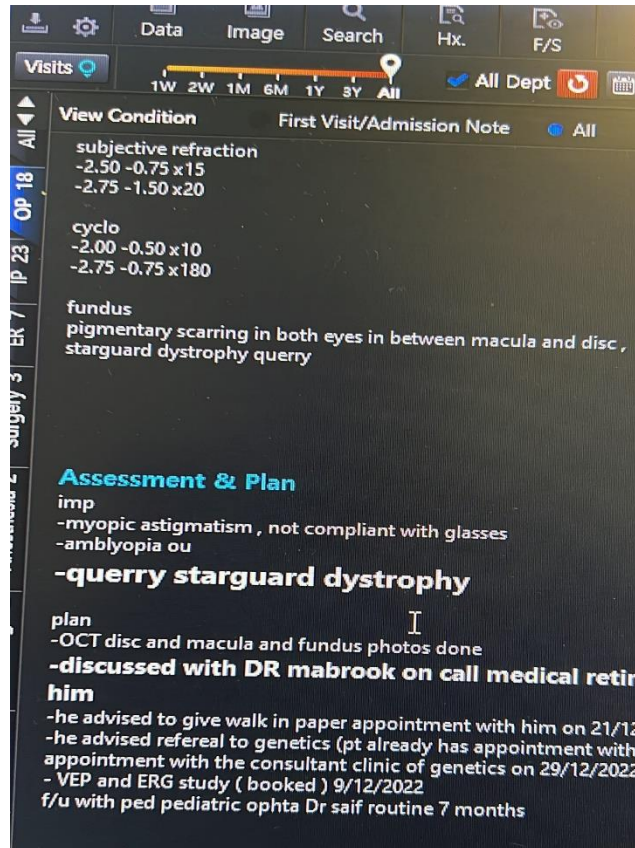
The third concern was the learning difficulties his son was facing in the school, I started the comprehensive work up of the patient thoroughly beginning by the blood works up that came all within normal (especially what was related the alopecia he had, one of them Vitamin D resistant *rickette* I presumed initially) and since of my initial working diagnosis was NF1 which needs a multiple paediatrics subspecialities opinions I decided to consult the paediatrics *Neurology, Dermatology, Orthopaedic, Ophthalmology, Behavioural team, Psychiatry*, and within couple of weeks the diagnosis came as NF1 with one striking exception related to alopecia totalise particularly replayed from the paediatrics dermatologist who reported that the alopecia he had is due to one rare dermatological disorder called TTDY, (Figure NO 3): which also not related to his major neuro-cutaneous problem i.e.,NF1.



(Figure NO 3: The initial Paediatrics dermatology replay that confirmed the TTDY disorder later):

Unfortunately, while we are waiting the different subspecialties replies, the ophthalmology report also was taking about the query stargardt dystrophy that the patient was most likely having (Figure NO 4):

which also uncommon presentation of NF1 but here in this case report I will not high light deeply on such disease and I will leave for the interested ophthalmologists to search about the simultaneous occurrence of both disorders.



(Figure NO 4: The initial Paediatrics ophthalmology replay that mentioned *Stargardt disease* which is a <sup>4</sup> *Stargardt disease* is a genetic eye disorder that causes retinal degeneration and vision loss. *Stargardt Disease* is the most common form of inherited macular degeneration, affecting about 30,000 people in the U.S. The progressive vision loss associated with *Stargardt disease* is caused by the degeneration of photoreceptor cells in the central portion of the retina called the macula.

*The retina is the delicate light-sensing tissue lining the inside wall of the back of the eye. Photoreceptor cells in the retina convert light into electrical signals, which are sent to the brain where they are processed to create the images we see. The macula, which is rich in cone photoreceptors, is responsible for sharp central vision — for tasks like reading, watching television, and looking at faces. Cones also provide vision in lighted settings and colour perception).*

Later among the subsequent management of the child he was labelled ultimately as NF-1 with moderate cognitive disability (relayed on his IQ of 65/100 reported later) and with incidentally conjoining such rare hair disorder of TTDY.

### 3. DISCUSSION ABOUT THE CASE

Among my Medline medical searches about any coexistence reported previously between the NF1 and any alopecia totalise (or TTDY), I just found the following two case reports connected both (NF1 and alopecia totalise):

FIRST ONE: A case report published in *2008 May-Jun;25(3):392-3* by *Pediatr Dermatol* who described a case of neurofibromatosis type 1 having an association with alopecia areata and the author mentioned further That associations of neurofibromatosis type 1 with alopecia areata has not been reported earlier and previously. Whether these associations reflect an autoimmune causal relationship with neurofibromatosis type 1, or something was coincidental which needs really to be searched deeply about it.

## International Journal of Novel Research in Healthcare and Nursing

Vol. 10, Issue 1, pp: (51-57), Month: January - April 2023, Available at: [www.noveltyjournals.com](http://www.noveltyjournals.com)

SECOND ONE: a case report Published on *February 27, 2020*, by *I-Ting Lee Jian-Min Chang Yu Fu* of a 32-year-old man with loss of overlying hair on the anterior aspect of his scalp (alopecia areata). The Dermatology report also confirmed presence of multiple cafe-au-lait spots and an ultimate NF1. The interesting thing in such case that the initial working diagnosis was plaque disease the patient was having, and the health care team could not get the real correlation between the plaque and any of the other elements which are the alopecia and NF1 or even the relation between the alopecia and NF1.

From to what mentioned previously from the case I presented and from the medical data I have collected about the NF1, this medical disorder (i.e., NF1) being still almost rare in our community, but it is a must to review frequently its diagnostic criteria that everybody know long time and try to seek about the new clinical signs that incidentally conjoining it even not involved in the original criteria, why is that ? since of the continues mutations we are facing every years and century in our human World that makes it a probable extension in modifying the diagnostic criteria of the NF1 in the coming days.

Cutaneous manifestations are one the mandatory keys of diagnosing the disorder and as in our case report the attendance of the alopecia caused by such TTDY may add a new member of the criteria of NF1 and still it remains very crucial to search about any underlying explanation of the co-existence between the NF1 and TTDY.

So, from the previous I may succeed on giving you a rapid review on this big title of the NF1 in children's, not only that and I may succeed on adding a newly undiscovered dermatological (Hair) disorder that may be considered as a futural new major criteria of NF1 that justified by a new mutation or other hidden reason.

### 4. CONCLUSION

**Though the already long-time established of NF1 criteria in both children's and adults, for the first time TTDY cojoin such disorder, is it an incidental thing or will be a new fixed criteria of the disorder?**

**Rationale for consent of the patient's family to publish the case:** The patient's father talked about all the rationales behind publishing the case in this medical journal and he agreed on that and signed on a paper that kept secretly in our institution's medical records to respect the patient's privacy and also for any unsuspected further medico-legal claims from the family side.

### REFERENCES

- [1] [https://www.rch.org.au/kidsinfo/fact\\_sheets/Neurofibromatosis/](https://www.rch.org.au/kidsinfo/fact_sheets/Neurofibromatosis/).
- [2] [https://www.ncbi.nlm.nih.gov/pmc/articles/PMC8655961/#:~:text=Introduction-,Neurofibromatosis%20type%20\(NF%201\)%20is%20a%20genetic%20disease,especially%20the%20central%20nervous%20system.&text=1%2C2-,It%20is%20the%20most%20common%20form%20of%20neurocutaneous%20diseases%20and,%2F3000%20to%201%2F4000](https://www.ncbi.nlm.nih.gov/pmc/articles/PMC8655961/#:~:text=Introduction-,Neurofibromatosis%20type%20(NF%201)%20is%20a%20genetic%20disease,especially%20the%20central%20nervous%20system.&text=1%2C2-,It%20is%20the%20most%20common%20form%20of%20neurocutaneous%20diseases%20and,%2F3000%20to%201%2F4000).
- [3] <https://medlineplus.gov/genetics/condition/trichothiodystrophy/>
- [4] [https://www.fightingblindness.org/diseases/stargardt-disease?gclid=Cj0KCQiAic6eBhCoARIsANlox8417Q65pTuH\\_zpuJSqJ6bKwOIRtkwpmKy-MyovkXG2z-rL3Yu2dyn4aAkyZEALw\\_wcB](https://www.fightingblindness.org/diseases/stargardt-disease?gclid=Cj0KCQiAic6eBhCoARIsANlox8417Q65pTuH_zpuJSqJ6bKwOIRtkwpmKy-MyovkXG2z-rL3Yu2dyn4aAkyZEALw_wcB).