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# Rare Case of C4 Perineurinoma with Lambdatic Parieto-occipital Neuroma in Von Recklinghausen Disease in a 5 Years Old Child: A Case Report

**Fondop Joseph [a], Atemkem Tsatedem Faustin [a], Atyam Ekoto Marie Christine [b], C. A. Djam [a*], Eyenga Victor [c], Scavarda Didier [d] and V. D. P. Djientcheu [c]**

*[a] FMSP UDs, Cameroon.*
*[b] HD Dschang, Cameroon.*
*[c] CHU, Yaoundé, UY1, Cameroon.*
*[d] CHU, Timone, Marseille, France.*

***Authors' contributions***

*This work was carried out in collaboration among all authors. All authors read and approved the final manuscript.*

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***Case Report***

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## ABSTRACT

Neuro fibromatosis groups together distinct diseases that most often share only certain cutaneous signs. Neurofibromatosis (NF) is a neurocutaneous syndrome characterized by the development of tumors of the central or peripheral nervous system including the brain, spinal cord, organs, skin, and bones. Prineurinoma is a relatively rare tumour that has been shown to be a proliferation of perineural cells with an incomplete basal lamina and no onion bulb appearance. Neuroma also known as traumatic neuroma or amputation neuroma or stump neuroma, is a focal non-neoplastic

**Corresponding author: Email: djamalain@yahoo.com;*

area of proliferative hyperplastic reaction secondary to peripheral nerve damage that commonly occurs after a focal trauma (acute or chronic) or surgery, such as amputation or partial transection; We report an extremely rare clinical case of a 5 year old child with neurfibromatosis congenita type 1 with asyntomatic medullary comrepssion and incidental discovery of a left intraductal C4 perineurinoma during the exploration of an occipital-parietal neuroma associated with a dehiscence of the lambda suture. He was operated on, with the postoperative course marked by paresis of the cervical muscles, which was in the process of recovery by physiotherapy.

*Keywords: Neurofibromatosis type 1; C4 perineurinoma; intracanalicular; neuroma; surgery.*

## 1. INTRODUCTION

- **Neuro fibromatosis** groups together distinct diseases that most often share only certain cutaneous signs. Neurofibromatosis (NF) is a neurocutaneous syndrome characterized by the development of tumors of the central or peripheral nervous system including the brain, spinal cord, organs, skin, and bones. (The term Recklinghausen refers to fibromatosis type 1 (NF1). Its prevalence is estimated at 1/3000 [1-3]. Since the 1988 National Institute of Heath (NIH) consensus conference, diagnostic criteria for NF1 have been established, their validity confirmed by biology. The clinical diagnostic criteria for NF1 have been validated and defined (Table 1) [2,4].

The cardinal signs of neurofibromatosis type 1 have been described:

- Café au Lait spots are the first manifestation of NF1, they are congenital, and rarely appear after the age of 2 years, the histological abnormalities are summarized by a hyperpigmentation of the basal ketinocytes, they are found in 99% of cases from the age of 1 year. Their size is greater than 0.5 cm in childhood and at least 1.5 cm after puberty have diagnostic value provided they are more than 6 in number.
- Lentigines have the appearance of a small café au lait spot, and correspond to macules of 1 to 3 cm in diameter located preferentially in the axillary, inguinal and sub-mammary region, they are rarely present before the age of 2 years, they are found in 90% of children aged 7 years.
- Cutaneous neurofibromas are small soft tumours, mobile with the skin, sessile or pedunculated, of variable size and number. They appear only at puberty and are exceptionally absent in adulthood; however, plexiform neurofibromas are located on the trunk, head, limbs and neck; they are always visible before 5 years of age, with 1/3 of cases developing in adolescence. Plexiform and/or nodular neurofibromas are sometimes the cause of peripheral motor and/or sensory, spinal cord, visceral nerve compression [3].

- **Perineurioma**: Prineurinoma is a fairly rare tumour, representing about 1% of peripheral nerve tumours but with very specific clinical, electrophysiological and radiological features [2,5]. The term perineurioma was first used by S.S. Lazarus and L.D. Trombetta [2]. For a nerve tumour that appeared to be a neurofibroma but whose histology, by electron microscopic study, turned out to be a proliferation of perineural cells with an incomplete basal lamina and without an onion bulb appearance. Until the fin 1970s, localized nerve hypertrophies were considered to be special forms of hypertrophic neuritis. There is still some confusion about the concept of localized hypertrophic neuropathy (LHN). H. Mitsumoto et al. have shown that the appearance of localized hypertrophic neuropathy can be due to a localized form of generalized hypertrophic neuropathy or to an intranervous tumour, in this case a perineurioma, a benign intranervous tumour developed from the perineural cells of the nerve [3,6].

- **Neuroma:** Neuroma also known as traumatic neuroma or amputation neuroma or stump neuroma, is a focal non-neoplastic area of proliferative hyperplastic reaction secondary to peripheral nerve damage that commonly occurs after a focal trauma (acute or chronic) or surgery, such as amputation or partial transection. Neuromas are more commonly located in the lower limbs, followed by head and neck; other extremely rare sites include the ulnar nerve followed by the radial nerve and the brachial plexus [5]. A neuroma is a benign tumor of nerve tissue

that is often associated with pain or specific types of various other symptoms. Neuromas most commonly arise from non-neuronal nervous tissue after amputation or trauma, or they can be true neoplasms [7,8].

We report here a rare clinical case of a 5 years old child, whose mother and sisters have NF1, and who presents with a left intracanal C4 peri neuroma with laterocolic spinal cord compression and a subcutaneous neuroma at the occipito parietal junction on dehiscence with lambdatic above a venous plexus.

## 2. CLINICAL CASE

Child KTZ 6 years old, male, came in consultation for occipital mass, occurred after the age of 4 months and since the age of 1 month is budding, He is the 4th of 4 siblings, with the mother also showing plexiform skin nodules, café au lait spots and Lentigines as well as his older sister (Fig. 1).

On physical examination, there is limited head movement, with a discrete right latero coli and a firm, puffy subcutaneous occipital mass. The skin is scattered with café au lait spots and more than 6 skin nodules of different ages.

No neurological deficits are observed in, osteotendinous reflexes are normal.

Cerebral CT shows a cervical mass with lambdatic parieto-occipital dehiscence and peri-lambdatic occipito parietal dysplasia, containing a venous plexus (Fig. 3).

Craniocervical MRI shows a cervical mass opposite C4 on the left, very suggestive of a benign tumour (Fig. 4).

After informed consent, the child was operated on, the incision is occipitocervical in order to have the removal of the occipital mass and the cervical mass at C4 level.

**In Per operative:** In the occipital region, there is an occipital mass under the galea whose almost complete removal reveals a dehiscence of the parieto-occipital bone containing a venous plexus. The anatomopathological analysis of this mass shows an amputation neuroma.

**Table 1. Diagnostic criteria for neurofibromatosis type 1 (NF1)**

| Original Diagnostic Criteria (1988) | Updated Diagnostic Criteria (2021) |
|---|---|
| A diagnosis of NF1 can be made if a person person has two or more of the following manifestations:<br>- Six or more "café au lait" macules<br>Six or more café au lait macules (brown spots) "greater than 5 mm in children<br>more than 15 mm in prepubertal children " more than 15 mm in post-pubescent<br>post-puberty - Freckles in the armpit (armpit) or in the groin<br>- Two or more neurofibromas of any type of any type, or a plexiform neurofibroma plexiform<br>- Two Lisch nodules<br>Two or more Lisch nodules of the iris (iris hamartomas)- Glioma<br>A distinctive bone lesion: dysplasia (abnormal growth) of the sphenoid bone behind the eye, or dysplasia of the bones, often in the lower part of the leg<br>- Having a close relative (parent, sibling or child) with NF1 | A diagnosis of NF1 can be made if a person person has two or more of the following manifestations:<br>- Six or more "café-au-lait-macules*" (brown spots on the skin) "greater than 5 mm in children<br>greater than 15 mm in prepubertal children " greater than 15 mm in post-pubertal individuals<br>post-puberty - Freckles in the armpit (armpit) or groin*.<br>- Two or more neurofibromas of any type of any type, or a plexiform neurofibroma plexiform<br>- Two or more Lisch nodules or two or more choroidal anomalies or<br>- Optic tract glioma (tumor of the visual pathway)<br>- A distinctive bony lesion such as: Sphenoid dysplasia; anterolateral curvature of the tibia (tibial dysplasia); or pseudarthrosis of a long bone<br>- A parent with NF1 according to the above criteria freckles)must be bilateral. |

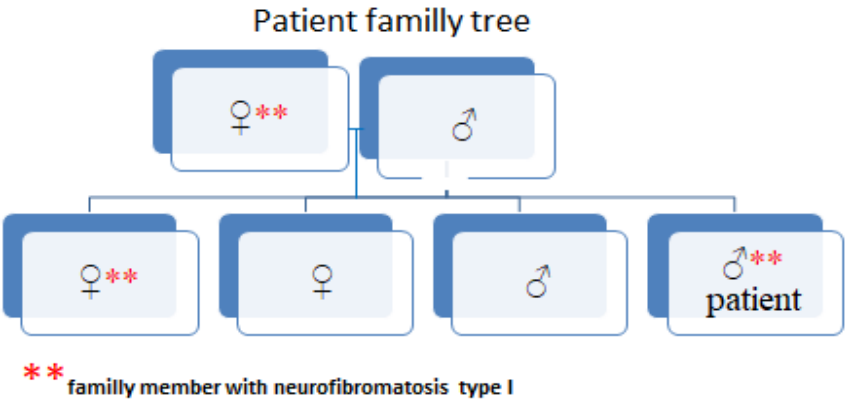


**Fig. 1. Patent familly tree**

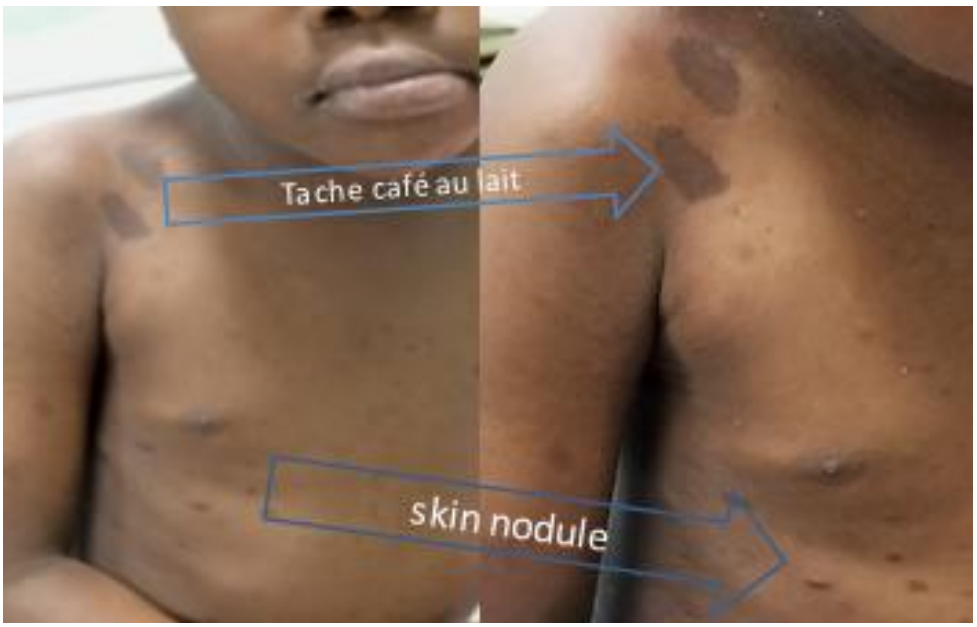


**Fig. 2. Examination of the patient's skin**

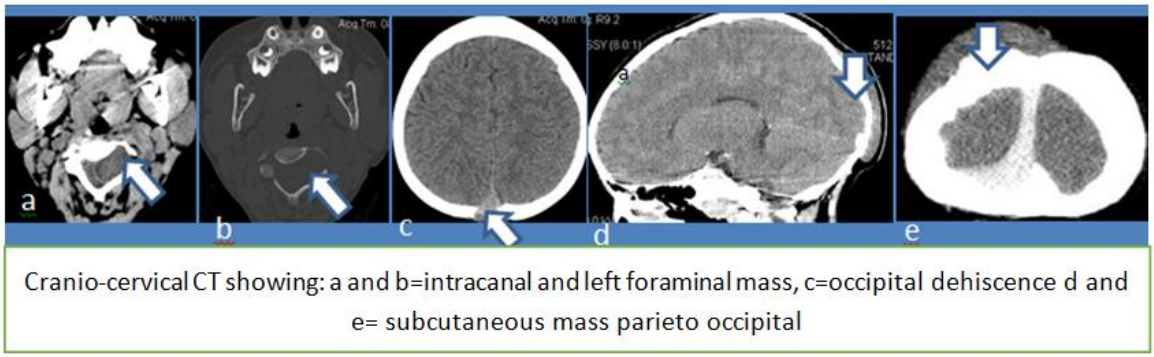


**Fig. 3. Cervical CT scan, showing the left side of the vertebral canal and an occipital mass under the galactic vertebrae with dehiscence of the lambda suture in relation to the sagittal sinus**

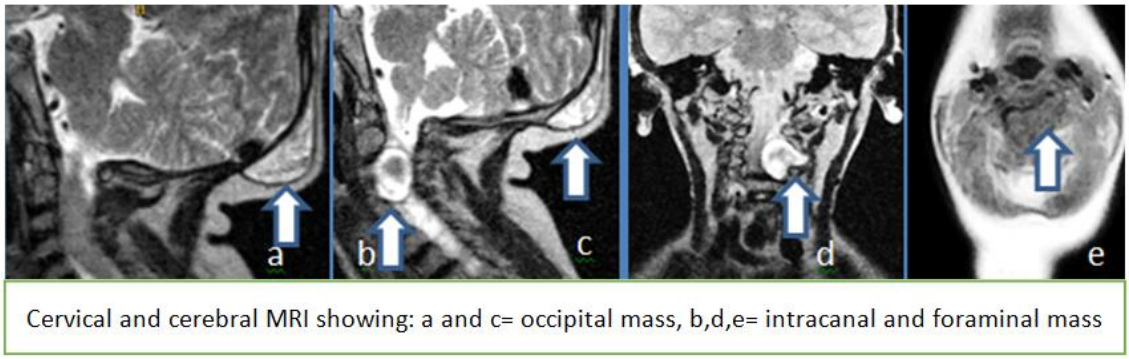


**Fig. 4. Craniocervical Magnetic resonance image (MRI) showing an intra canal mass opposite C4 with spinal cord compression**

And in the posterior cervical region, intra-canal, we noted a mass enveloping the left C4 nerve root, measuring 3cmx2cmx1cm. Anatomical pathology analysis described it as a peri-neuroma.

The immediate operative follow-up was marked by pain treated by analgesics, resumption of feeding after 24 hours, latero coli with the head tilted to the right, paresis of the elevation of the left upper limb, without any infectious sign. He was discharged at 7 days post-op to continue the dressing and the rehabilitation in external, with good scarring, followed 4 days later by an emergency rehospitalization for suppuration of the surgical wound with signs of meningeal irritation, of which the sampling and culture allowed the isolation of Pseudomonas aeruginosa. He was discharged from hospital after 4 weeks to home with physiotherapy and home dressing. The wound is completely healed.

At 3 months post-surgery he presented with hypotonia of the left cervico-lateral muscles resulting in a rightward tilt of the head, hypotonia of the left deltoid muscle limiting the extension of the left upper limb.

## 3. DISCUSSION

Our patient is 5 years old, has a mother and a sister with neurofibromatosis type 1, he has several cafe au lait spots on the skin, with skin nodules and discrete axillary lines, and has occipito parietal dysplasia. He is a carrier of congenital neurofibromatosis; he presents the cardinal signs of neurofibromatosis typ1. Although biological tests have not been carried out, he has had VON Recklinghausen disease [2,5,6].

The appearance of neurofibromatosis nodules at the age of 5 years is as is in favour of plexiform neurofibromas [3]. These nodules are disseminated on the patient's skin, and may even develop in the spinal canal, causing spinal cord compression [9].

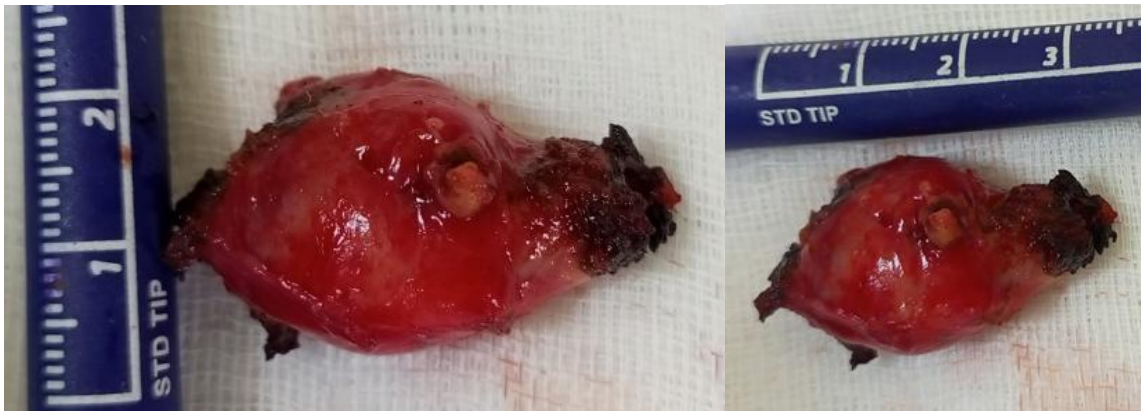


**Fig. 5. Enveloping C4 mass, pathology examination describes peri-neuroma**

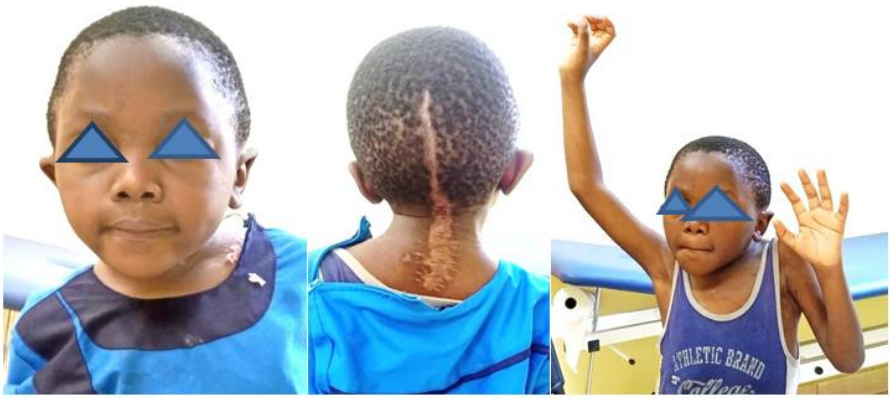

**Fig. 6. 3 months post: Right aerocolic with healing screw and left deltoid paresis limiting left upper limb extension**

Intra neural perineuroma is a rather rare tumour, which was considered by some authors as a neurofibroma before being considered as a perineurinoma by H. Mitsumoto et al. [10], who had the honour of being the first to have attributed to a perineu rioma, previously defined by S.S. Lazarus and L.D. Trombetta [11], the origin of certain cases of localised hypertrophic neuropathy, the perineurinoma representing about 1% of peripheral nerve tumours but with very particular clinical, electrophysiological and radiological characteristics [6,10-12], the evolution is very slow, with a clear predominance of motor involvement, and the absence of extension to adjacent nerve trunks during follow-up [12]. Of 97 cases reported in the literature by P. Bouche [6].

The age of discovery of perineurinomas varied. It was less than or equal to 10 years of age in 20 cases out of 97, i.e. nearly 20% of cases; it is between 11 and 18 years of age in 35% of cases, and less than 30 years of age in 77% of cases, i.e. three quarters of cases. The male/female ratio is 47/50. The nerves affected were, in order of frequency, the trunk of the sciatic nerve in 28 cases, then the common fibular in 8 cases [6]. In the series by M.L. Mauermann et al. the average age at diagnosis was the average age of onset was 17 years (range 2-56), but the average time from symptom onset to diagnosis was 3 years (range 6 months to 30 years). The main complaint was a motor deficit in the territory of interest, and considering the entire course, all patients had a motor deficit. Of the 32 patients, 24 had sensory symptoms, 13 with pain. The neuropathy was purely motor in 5 cases [6,13].

In our 5 year old patient, the tumour was intra canal with a medullary compression at the left C4 level, the only clinical sign described was the latera coli, the removal of this mass prevented medullary complications, especially at the C4 level which could be complicated by a phrenic nerve deficit. The postoperative period was marked by neck muscle deficits that had to be re-educated along with the other muscles innervated by the cervical plexus.

Amputation neuroma is a benign tumour formed by more or less abnormal fibres. It is a rare benign lesion affection peripheral nerves characterized by non-neoplastic proliferation of the proximal end of a partially or completely transected nerve [5]; No case has been described in the literature without any notion of trauma or surgery, and the same is true for the association with neurofibromatosis. As in our patient, given that we had a dehiscence of the lambdatic suture with an occipito parietal dystrophy opposite the dehiscence, this neuroma could be explained by minimal nerve trauma, linked to the gestures of everyday life. But it may also be an atypical case occurring in a traumatic context.

## 4. CONCLUSION

Neurofibromatosis type 1 associated with perineurinoma and neuroma in the same 5-year-old patient is a very rare clinical picture. The existence of this condition in our patient's mother and sister attests to the fact that it is a condition related to a chromosomal anomaly. The perinerinoma developed in the cervical spinal canal perimedullary, discovered in a 5 year old child on neurofibromatosis can be responsible for spinal cord compression and requires a surgical intervention followed by a reduction of the post operative nerve deficit.

The amputation neuroma is a benign tumor caused by pseudo-nervous lesions of slow and progressive growth; it is rare in children, especially without any notion of amputation; its location in a region of bony dehiscence of the lamda suture made us evoke its physiopathology by nerve lesions caused by the bony edges of the dehiscence; its development would be favored in our patient by the terrain of neurofibromatosis type 1, making the exceptional particularity of our clinical case. The association of neurofibromatosis type 1 with cervical intraductal neuroma with medullary compression and occipital subcutaneous neuroma in front of a dehiscence of the lambda suture in a non-traumatic context in a child is exceptional and makes the particularity of our clinical case.

Neurofibromatosis type 1 is a terrain that can favor the development of several types of benign nerve tumors, isolated or associated.

## CONSENT

As per international standard, parental written consent has been collected and preserved by the author(s).

## ETHICAL APPROVAL

As per international standard or university standard written ethical approval has been collected and preserved by the author(s).

## COMPETING INTERESTS

Authors have declared that no competing interests exist.

## REFERENCES


1. Ryota Tamura. Current understanding of Neurofibromatosis type 1, 2 and schwannomatosis, Int. J. Mol. Sci. 2021; 22:5850.
   Available:https://doi.org/10.3390/ijms22115850 https://www.mdpi.com/journal/ijms
2. Ina Ly K, Jaishri O. Blakeley. The diagnosis and management of neurofibromatosis type 1, medical clinics of North America. 2019;103(6): 1035-1054.
3. Valeyrie-Allonore P. Wolkenstein, neuro-fibromatosis 1 and variant forms EMC. Neurology. 11-170-A-65.
4. Neurofibromatose Midwest Cliniques, sensibilisation, recherche, éducation et soutien Mise à jour des critères diagnostiques NF2021 1 - Neurofibromatose Midwest (nfmidwest.org) Publié par NF Midwest | Le 20 mai ; 2021
   Available:https://www.nfmidwest.org/blog/2021-nf1-diagnostic-criteria-update/
5. Fabio Sandomenico, Antonio Corvino, Sergio Venanzio Setola, Igino Simonetti, Mauro Porcaro, Piero Trovato, Orlando Catalano, Antonella Petrillo. Post-amputation neuroma of radial nerve in a patient with ephitelioid sarcoma: Case report and literature review. Acta Biomed. 2020;91(1): 122-127. Published online 2020 Mar 19.
   DOI: 10.23750/abm.v91i1. 8510, PMCID: PMC7569598PMID: 32191665
6. Bouche P. Perineuriomas, perineurioma, La Lettre du Neurologue Nerf, Muscle. 2011;15:n°1.
7. Mate Zabaglo, Mark A. Dreyer. Neuroma. NIH ?Stat Pearls [Internet]. Last Update; 2022.
8. Na Rae Kim, Hyun Yee Cho, Dong Hae Chung, Keon Kuk Kim, Jae Hee Cho, Seung Joon Choi Post-transplant Amputation Traumatic Neuroma of the Hilum and Extrahepatic Duct in a Liver Donor,J Pathol Transl Med. 2018; 52(3):191-194.
   Published online 2017 Aug 4.
   DOI: 10.4132/jptm.2017.01.20 ,PMCID: PMC5964281PMID: 28774160
9. Fondop J, Atemkem TF, Amougou B, Ekoto Atyam MC, Nkono Banga D, Dogmo A, Ndouma A, Fuentes S. Cervical spinal cord compression by a plexiform nodule in neurofibromatosis type1 treated surgically by posterolateral approach: About a case with post traumatic revelation. Journal of Spine.
10. Mitsumoto H, Wilbourn AJ, Goren H. Perineurioma as the cause of localized hypertrophic neuropathy. Muscle Nerve and Neurological Disorders. Schol Arena. 1980;3:403-12.
    Available:www.scholarena.com
11. Lazarus SS, Trombetta LD. Ultrastructural identification of a benign perineurial cell tumor. Cancer 1978;41:1823-9.
12. Rachelle Ai Nahed, Christophe Vvandendries, Thierry maisonobe,

guillaume forgeot, Intraneural perineurioma: Retrospective analysis of a series of 9 patients. Revue Eurologique. 2022; 178:S4. DOI:10.1016/j.neurol.2022.02.242

13. Mauermann ML, Amrani KK, Kuntz NL et al. Longitu dinal study of intraneural perineurioma-A benign, focal hypertrophic neuropathy of youth. Brain 2009;132: 2265-76.



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