Type I Neurofibromatosis With Cutaneous Localization: A Study Of 63 Cases

A Sankale, M Foba, A Ndiaye

Citation

A Sankale, M Foba, A Ndiaye. *Type I Neurofibromatosis With Cutaneous Localization: A Study Of 63 Cases*. The Internet Journal of Plastic Surgery. 2019 Volume 14 Number 1.

DOI: 10.5580/IJPS.53741

Abstract

Skin manifestations of neurofibromatosis present both an aesthetic and functional handicap. The objective of this study was to report our experience in the surgical management of these plexiform neurofibromas.

A retrospective study was conducted over a period of 16 years (January 2001 to December 2017) and involved 63 patients with neurofibromatosis type 1. The sex ratio was 0.85 (29 men and 34 women) and the average age was 24 years. Aesthetic discomfort was the main reason for consultation. All the patients presented with cutaneous neurofibromas at multiple locations and plexiform neurofibromas were present in 96.7% of cases. The major tumor was located in the cervical-cephalic region in 60% of cases. The average size of the neurofibromas was 10.49 cm [1.5 to 45 cm].

The indication of excision was considered based on the size and number of tumors as well as the extent of aesthetic and functional discomfort. Thirty-nine patients were operated on, or 60.9% of the cases. Extra-tumoral excisions with cutaneous closure by direct suture plus or minus local plasty accounted for 43.6% of the interventions, or 17 patients. Intra-tumor reshaping resections represented 56.4% of cases or 23 patients. Superinfection of the operative site and hypertrophic scars were the two complications encountered (n = 2). The average follow-up was 5 months [10 days - 5 years]. The different procedures gave aesthetic (69%) and functional (100%) satisfactory results.

INTRODUCTION

Von Recklinghausen disease or neurofibromatosis type I (NF1) is the most common type of phakomatosis (approximately 1/3000 births) [4]. It is a genetic disease with autosomal dominant transmission [2]. However, the frequency of neo-mutations is about 40 % [5]. The gene responsible is located on the long arm of chromosome 17. The protein encoded by this gene is neurofibromin, which is involved in the control of cell proliferation. Neurofibromatosis type I is a multi-systemic pathology but with a particular neuro-cutaneous tropism.

The purpose of this work is to conduct an epidemiological and clinical study of patients with NF1 followed in our department, and to report the results of the various surgical procedures as practiced in our working environment.

PATIENTS AND METHODS

This is a retrospective study over a period of 16 years (January 2001 to December 2017) on 63 patients with neurofibromatosis type 1 skin manifestations, managed in the Plastic and Reconstructive Surgery Department of the Aristide Le Dantec Hospital, in Dakar, Senegal. We studied epidemiological, clinical and therapeutic parameters.

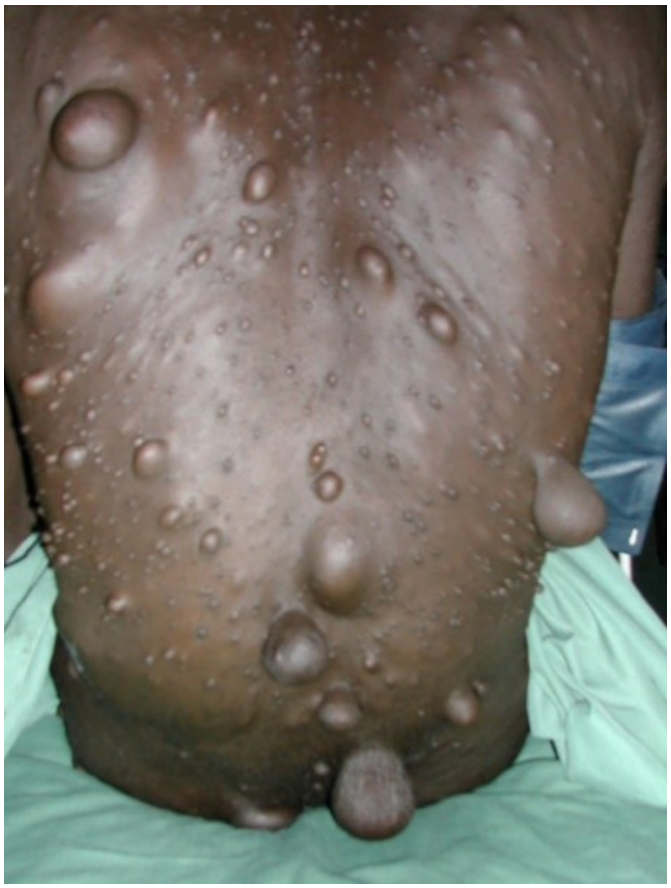
RESULTS

There were 29 men and 34 women for a 0.85 sex ratio. The average age was 24 years. The sporadic isolated cases accounted for 90.5% of cases. The esthetic discomfort constituted the main reason for consultation, followed by the functional discomfort in 58.3% of the cases.

From a clinical point of view, all patients had multiple localized skin neurofibromas and "café au lait" spots (figure1).

Figure 1

Multiple cutaneous Neurofibromas (dorsal location)



Plexiform neurofibromas were almost always present (96.7%). The major tumor was located in 60 % of the cases in the cervical-cephalic region (Figures 2 and 3), followed by the upper limb (14%). The average size of the neurofibromas was 10.49 cm [1.5-45 cm].

Figure 2

right hemi-facial neurofibroma in an 8-year-old girl

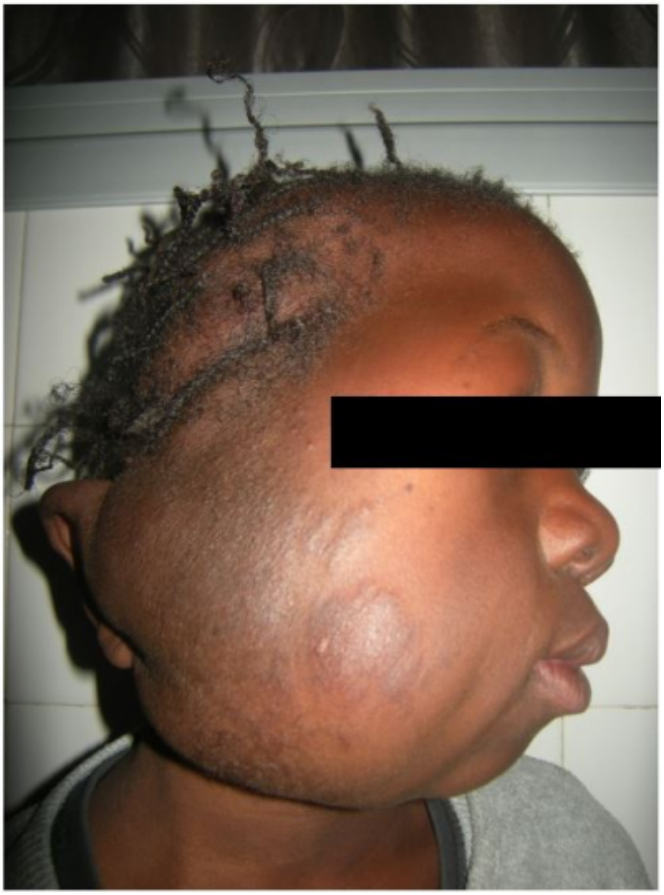


Figure 3

fronto-temporal neurofibroma

The indication of excision was considered based on the size and number of tumors as well as the extent of aesthetic and functional discomfort. Thirty-nine patients were operated representing 60.9 % of the cases. Extra-tumoral resection was the procedure employed in 43.6% of cases or 17 patients, followed by skin closure by direct suture with or without an associated local cutaneous plasty. Intra-tumoral reshaping resections accounted for 56.4 % of procedures or 23 patients. Supra-infection of the operative site and hypertrophic scars were the two complications encountered in 2 patients. The average follow-up was 5 months [10 days - 6 years]. The different procedures gave satisfactory results both at aesthetic (69% of cases) and functional (100% of cases) levels (Figures 4 and 5).

Figure 4a

plexiform neurofibroma of the face preoperative aspect (face and profile)

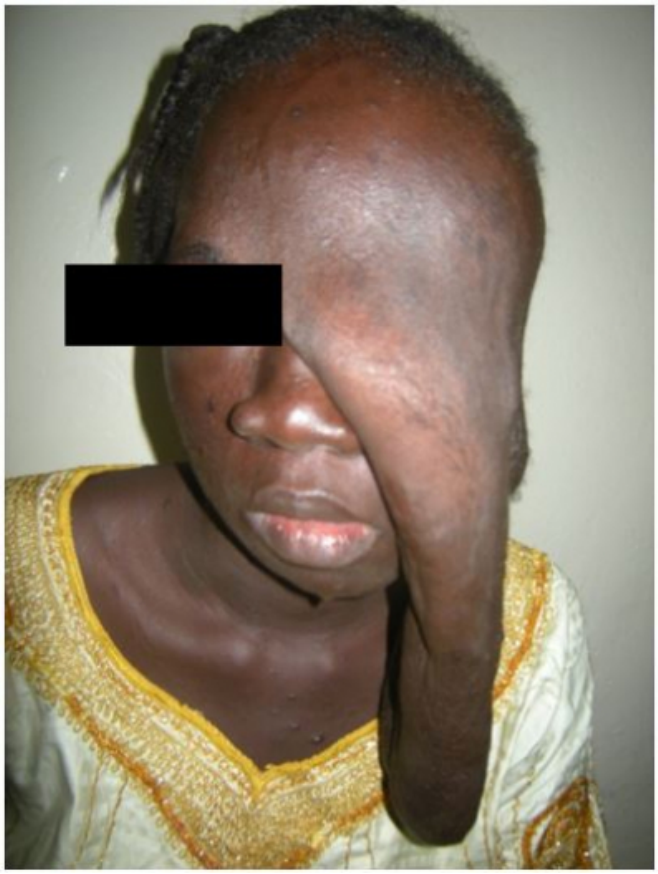


Figure 4b

plexiform neurofibroma of the face preoperative aspect (face and profile)

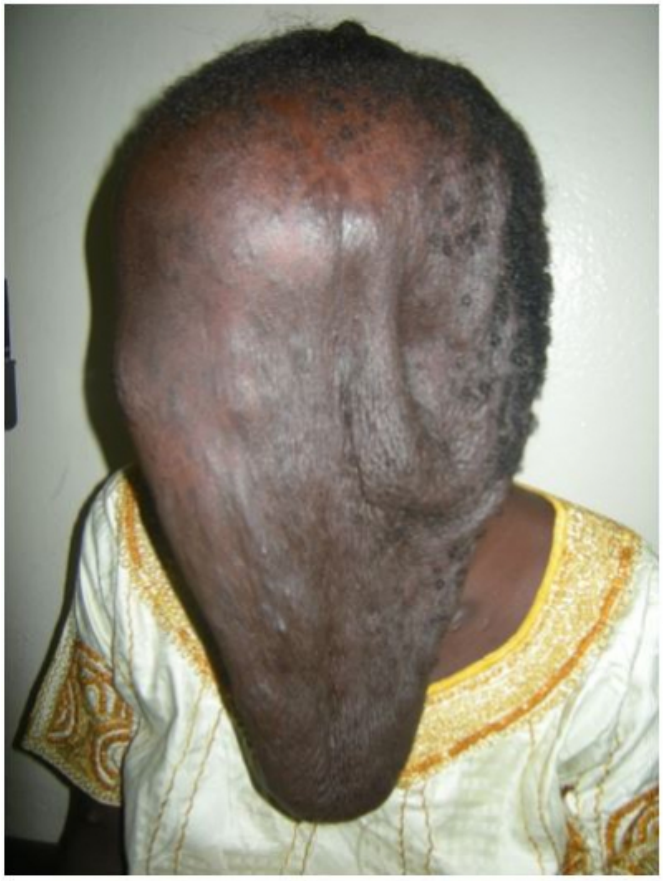


Figure 4c

plexiform neurofibroma of the face result (face and profile) after 2 interventions with left eye enucleation (4 years follow-up)

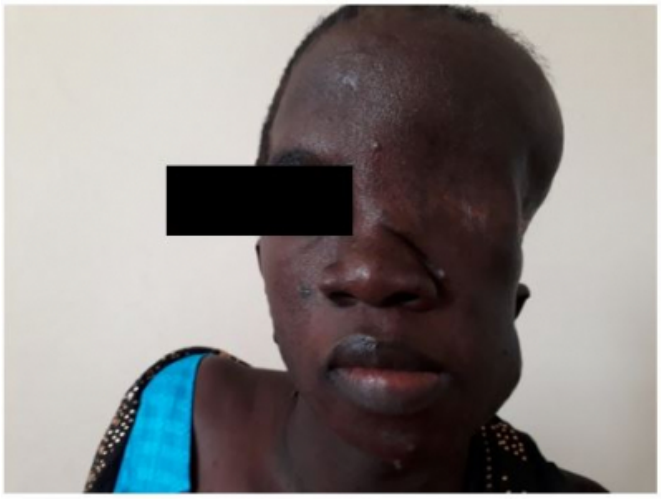


Figure 4d

plexiform neurofibroma of the face result (face and profile) after 2 interventions with left eye enucleation (4 years follow-up)

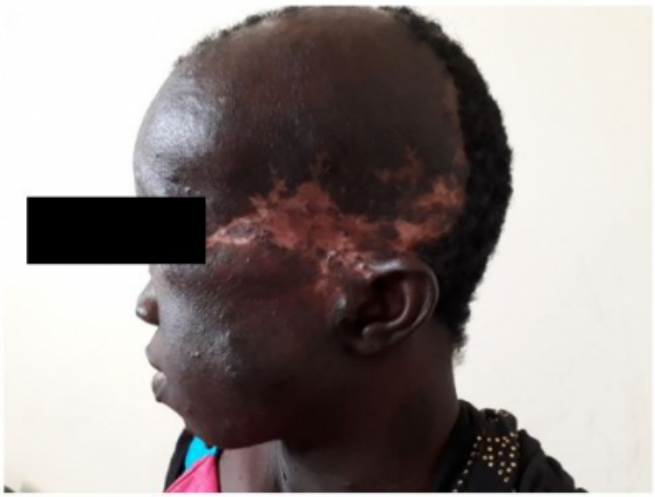


Figure 5a

Neurofibromatosis of the head and left shoulder pre-operative aspect

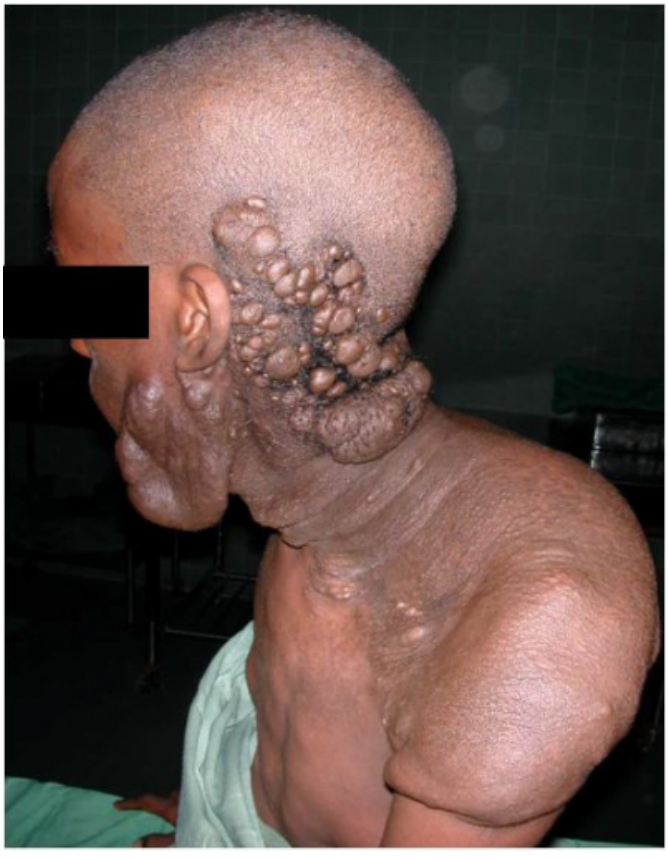
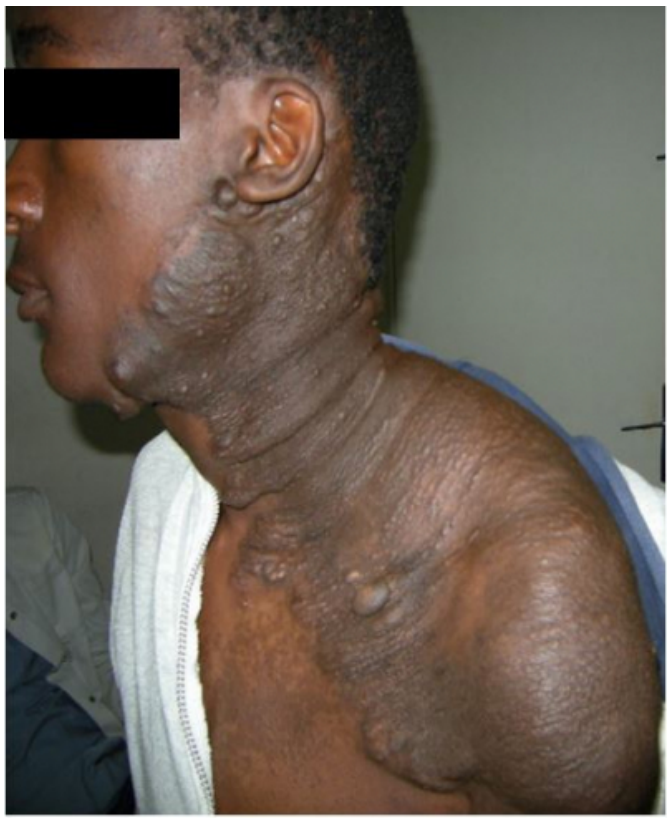


Figure 5b

Neurofibromatosis of the head and left shoulder result after 2 interventions (5 years follow-up)



DISCUSSION

From the epidemiological point of view, there was a slight feminine predominance in our study: this could be explained by the fact that aesthetic problems may have a stronger psycho - social impact in women than in men. Data from literature [4] confirms these results. The aesthetic discomfort due to cutaneous neurofibromas especially cervical- cephalic becomes particularly important in young adult patients between 20 and 30 years [6] as in our study where the average age at consultation was 24 years old.

The average duration of evolution was 18 years in our study but there is variability in the period of onset of the first skin localization, around the age of 10 years. Several authors agree that cutaneous neurofibromas do not usually appear before puberty [5]. On the other hand, plexiform neurofibromas are most often visible before the age of 5 years.

Among the functional signs documented in our patients, cosmetic discomfort followed by functional impairment (58.3%) were the main functional signs found. In fact, the unsightly appearance alters the quality of life and has an impact on the patient's psycho - social development. [11].

The study of Ferner in 2017 [5] based on a questionnaire concerning the quality of life of patients with type 1 neurofibromatosis found that 32% of patients had moderate aesthetic problems and 16% of patients had severe problems.

Systematic physical signs were "café au lait" skin lesions and cutaneous neurofibromas in all patients and plexiform neurofibromas in 96.7% of our patients. The study of Jeeblaoui in Tunisia [8] found 100% plexiform neurofibromas. This can be explained by the fact that patients consult primarily when disease manifestations are socially unbearable because of bulky and unsightly tumors.

Surgery is the treatment for plexiform neurofibromas. In fact, no medical treatment has hitherto made it possible to slow down their growth [6, 13]. Their excision is more or less complex depending on their size and their location [12].

The lag to intervention time is relatively long in our study because of the psycho- social and economic context of patients and factors related to our practice environment. Marchac [11] advocates early surgery to prevent recurrence.

Plexiform neurofibromas of the face [8, 10] pose different problems. Indeed, if their size is large, they can deform underlying bone and cartilage structures. If the tumor is small, simple excision is appropriate. If it is too large to allow direct closure, all techniques of reconstructive surgery can be used: skin grafts, flaps, or more elaborate procedures such as reconstructive microsurgery and skin expansion [1, 15]. The use of these various methods depends on the location of the tumor. On the forehead and the scalp, reconstruction rarely raises problems, these regions being sources of many flaps for closure [3, 1 4, 16]

Two opposing surgical technics are possible: intra-tumoral resection, and total excision. Indeed, intra-tumoral resection, which is practiced by most authors [12], is recommended in cases where the complete excision of the tumor would cause significant mutilation and/or significant functional impairment. Bearing in mind the natural evolution of neurofibromas, intra-tumor resection is conceivable only if making repeated interventions to optimize the aesthetic result.

Extra-tumoral excision can go as far as amputation but face localizations pose reconstruction problems.

The advent of microsurgery has enabled some teams to make subtotal resections with pre and post-operative MRIs. Unfortunately, we do not have the technological platform to

perform microsurgery in our setting.

Other techniques have been reported such as skin grafting, facial plasty, and subcutaneous lifting [9, 16].

The surgery of neurofibromas is very hemorrhagic, and in our context infiltration adrenaline serum, monopolar coagulation and ligation of large vessels are the only means used to control bleeding. In China, Zhao proposes a procedure of preoperative endovascular embolization [17] hardly feasible in our context.

The esthetic results were judged satisfactory in 69% of the cases. These results are based on subjective criteria including the initial appearance of neurofibromas, the psycho-social impact and the final appearance after intervention and the degree of satisfaction of the patients.

CONCLUSION

Despite the limitations of our practice environment, surgery for neurofibromas with cutaneous localization is possible. Improving the quality of life of patients with NF1 involves informing patients and their families.

References

1 . Angel MF, Persing JA, Edgerton MT 1994. Reconstructive surgery for neurofibromatosis. In: Huson SM, ed. The neurofibromatosis: a pathogenetic and clinical overview. London res : Chapmann and Hall, 332-350
2 . Barker D, Wright E, Nguyen K, Cannon L, Fain P, Goldgar D, Bishop DT, Carey J, Baty B, Kivlin J, et al. Gene for Von Recklinghausen neurofibromatosis is in the pericentromeric region of chromosome 17. Science 1987; 236 (4805): 1100-1102
3 . Bessede JP. Skin flaps. Medicine 2017; 8: 8p.
4 . Cnossen MH, Goede-Bolder A, Van den Broek KM, Waasdorp CME, Oranje AP, Stroink H, Simonsz HJ, AMW van den Ouweland , Halley DJJ, Niermeijer MF. A prospective 10 years follow up study of patients with neurofibromatosis type 1. Arch Dis child 1998; 78: 408-412.
5. Ferner RE, Huson SM, Thomas N, Mercer G, Williams V; Leschziner GD, SK Afridi, Golding JF. Evaluation of quality of life in adults with neurofibromatosis 1 (NF1) using the impact of NF1 quality of life (INF1-QOL) questionnaire. Health Qual Life Outcomes [en line] .2017; 15; 34. Available at https://www.nbci.nlm.nih.gov/pmc/articles/PMC5307827/ Accessed 06/05/2017.
6 . Granström S, Langen Bruch A, Augustin M, Mautner VF. Psychological burden in adult neurofibromatosis type 1. Dermatology 2012 ; 224 (2): 160-167.
7 . High Authority of Health: Center of reference labeled neurofibromatoses. National Diagnostic and Care Protocol (P NDS) Neurofibromatosis 2016: 1-65.
8 . Jeblaoui Y, Neji B , Haddad S, D. Mnif , S. Hchicha. Difficulties in the management of cephalic extremity involvement in neurofibromatosis. Annals of Aesthetic Plastic Surgery . 2007; 52 (1): 43-50.
9 . Lantieri L. Neurofibromatosis. Innovatio n Plastic Surgery . 2015: 3p.
10 . Latham K, Buchanan EP, Suver D, Gruss JS. Neurofibromatosis of the Head and Neck. Plast Reconstr Surg [online]. 2014; 135 (3): 845-853. Available on:
11 . Marchac D, Britto JA. Remodeling the upper eyelid in the management of orbitopalpebral neurofibromatosis. Br J Plastic Surgery [online]. 2005; 58 (7): 944-956.
12 . National Institutes of Health Consensus Development Conference Neurofibromatosis. Conference statement. Arch Neurol [en line]. 1988; 45 (5): 575-578
13 . Nguyen R, E Dombi , Widemann CB. Growth dynamics of plexiform neurofibromas : a retrospective cohort study of 201 patients with neurofibromatosis 1. Orphanet J Rare Dis . 2012; 7 (75): 3-11p.
14 . PZ Page, Page GP, Scotland E, K rf BR, Leplege A, Wolkenstein P. Impact of neurofibromatosis 1 on Quality of Life: A cross-sectional study of 176 American cases. Am J Med Genet A [en line]. 2006; 140 (18): 1893-1898.
15 . Rilliet B, Pittet B, D Montandonb, Narada AP From Sch they Ribaupierre SN F Boscherini D, Di Rocco C. Achieving orbitofrontal -temporo-face in the neurofibromatosis type 1 (NF1). Neurosurgery 2010; 56 (2010): 257-270.
16 . Thornton JF, Gosman AA. Skin grafts and skin substitutes and princesses of flaps. Selected Readin g in Plastic Surgery. 2004; 10 (1): 1-42.
17 . Zhao M, Qiang J, Li Y, Tang Y, W Chen, Yang Z, Ma N, W Wang, Xu L, Feng J. Plastic surgery of scalp and facial plexiform neurofibromas. Zhongguo Xiu Fu Chong Jian Wai Ke Za Zhi . 2015; 29 (11): 1401-4.

Author Information

Anne-Aurore Sankale
Department of Plastic, Reconstructive and Aesthetic Surgery, Le Dantec Hospital
Dakar, Senegal

Mamadou Lassana Foba
Department of Plastic, Reconstructive and Aesthetic Surgery, Le Dantec Hospital
Dakar, Senegal

Aïnina Ndiaye
Department of Plastic, Reconstructive and Aesthetic Surgery, Le Dantec Hospital
Dakar, Senegal