SAS Journal of Surgery
Abbreviated Key Title: SAS J Surg
ISSN 2454-5104
Journal homepage: https://www.saspublishers.com

OPEN ACCESS

Plastic Surgery

Labial Reconstruction for Congenital Insensitivity to Pain: A Case Report

Dr. Jihane Hamdaoui[1*], Pr. Samir El Mazouz1, Pr. Noureddine Gharib[1], Pr. Abdellah Abbassi[1], Pr. Jawad Hafidi[1]

[1]Plastic Surgery Department, Hospital University Ibn Sina, Rabat, Morocco

DOI: 10.36347/sasjs.2024.v10i04.011 | **Received:** 23.02.2024 | **Accepted:** 06.04.2024 | **Published:** 16.04.2024

***Corresponding author:** Dr. Jihane Hamdaoui
Plastic Surgery Department, Hospital University Ibn Sina, Rabat, Morocco

Abstract **Case Report**

Congenital insensitivity to pain (CIP) is a very rare condition, most often of genetic origin. The authors report the case of a 10-year-old girl, followed for CIP following self-mutilation, particularly serious oro-digital, which is addressed in our training for lip reconstruction. CIP with anhidrosis is a very rare condition. It is characterized by feverish attacks, anhidrosis, absence of painful sensation, self-harm and sometimes mental retardation. Complications of this insensitivity (neglected fractures, burns, oro-digital mutilation) can be life-threatening. The treatment remains preventive. Hence the interest in educating patients and their families.

Keywords: Congenital insensitivity to pain, lip reconstruction, mutilation.

INTRODUCTION

Congenital insensitivity to pain (CIP) is a very rare clinical condition, most often of genetic origin (autosomal recessive transmission).

Its most severe form is characterized by an absence or radical reduction in pain perception, recurrent episodes of fever due to the absence of sweating (anhidrosis) and mental retardation.

The various accidents and complications, particularly oral-digital self-mutilation, pose a real problem for multidisciplinary care. Prevention, often difficult, seems to be the only alternative.

CASE REPORT

We report a case of a 10-year-old girl, from a non-consanguineous marriage, the only one of her parents, and without any notion of a similar case in the family, who has presented insensitivity to pain since the age of 4 months, a notion of self-mutilation, particularly oro-digital, as well as psychomotor delay. She was referred by his pediatrician for surgical treatment of a loss of labial substance hindering salivary continence (Fig 1) following self-mutilation and in whom the examination found pulpal amputations of the 1st and 2nd fingers of the right hand, 2nd, 3rd and 4th fingers of the left hand (Fig 2). An amputation of the big and 5th toes of the right foot (Fig 3).

The neurological examination noted respect for tactile, vibratory and thermal sensitivity with the presence of tendon reflexes. The treatment was based on reconstruction of the lower lip using an Estlander heterolabial flap (Fig 4), as well as raising awareness of the child and his family on measures to prevent trauma.

The functional and aesthetic result is considered satisfactory by the surgeon and the patient's family, allowing the young girl to regain salivary continence, speaking and a normal smile.

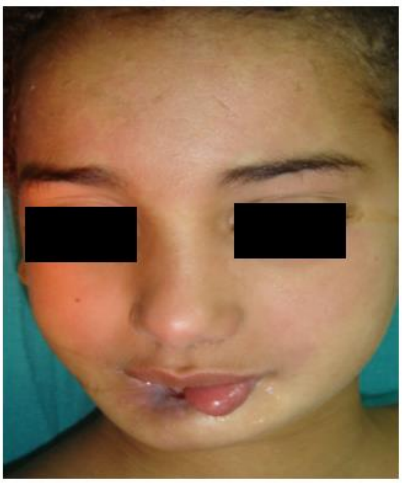

Figure 1: Labial loss of substance

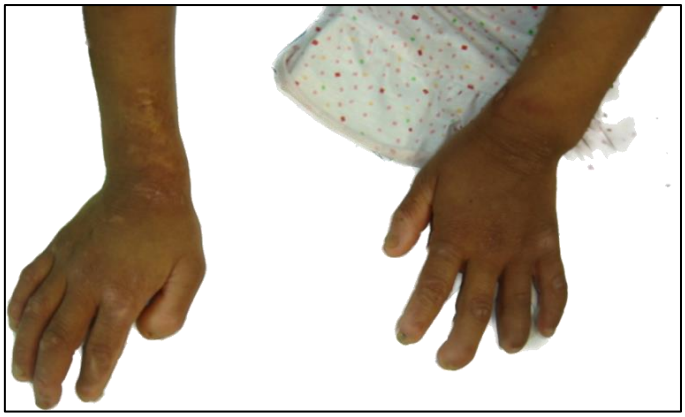

Figure 2: Pulp amputations following self-mutilation

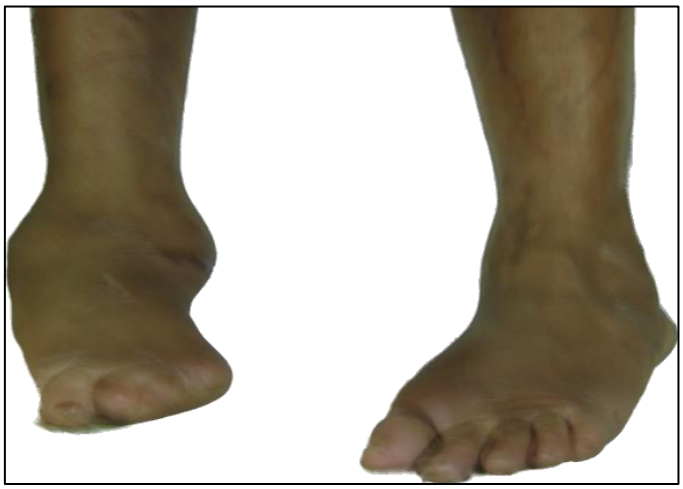

Figure 3: Amputation of 1st and 5th toes of right foot

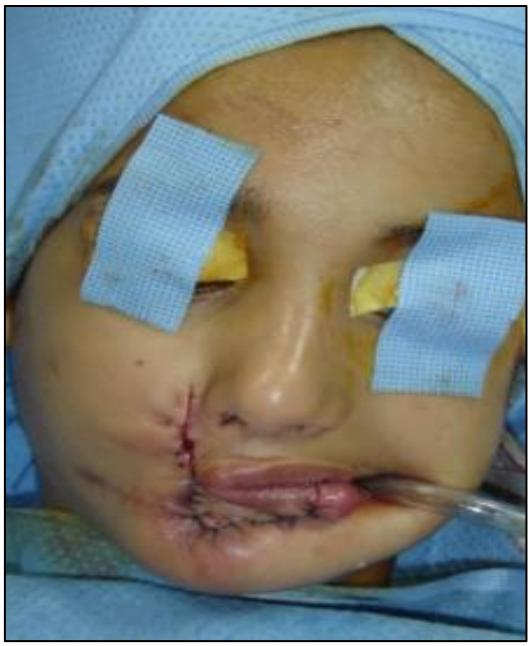

Figure 4: Estlander flap for labial reconstruction

DISCUSSION

Congenital insensitivity to pain (CIP) with anhidrosis is a very rare genetic condition. Its initial description dates back to 1932 by Dearbon [1]. Since then, the number of cases diagnosed worldwide remains uncertain. An international association (Tomorrow) lists these cases from Japan. It officially reported nationwide 300 cases in 2007. Eighty-seven other cases were

diagnosed in the United States, only two cases in New Zealand, two cases in Morocco, and two cases in France [2-4]. Since then, other sporadic cases have been reported.

This pathology is characterized in its severe form by total analgesia, which explains the occurrence of various injuries during the most trivial actions of daily life. Chewing, for example, can lead to self-mutilation of the lips as in our patient who had a loss of labial substance, the tongue and the inner side of the cheeks. In the same context, severe trauma can occur (burns, wounds, trophic disorders, fractures and other osteoarticular lesions) wrongly directing (especially in the case of mental retardation) towards a psychiatric disorder, Silverman syndrome or Lesch-Nyhan.

The absence of crying and protective behavior of the injured area is characteristic of CIP [3, 4].

Episodes of fever due to anhidrosis are recurrent. The osteotendinous reflexes and other modes of sensitivity are preserved.

The CIP of this young patient is linked to a genetic anomaly of recessive inheritance. In fact, the genetic study has revealed a hereditary sensory neuropathic disease type IV (HSAN IV: hereditary and sensory automatic neuropathy IV), which is due to an abnormality of the TRKA or NTRK1 gene (neurotrophic tyrosine kinase receptor), responsible of massive neuronal death affecting these two populations of peripheral nerve fibers and which corresponds well to the clinical phenotype [5].

In this type IV neuropathy, skin biopsy shows the absence of epidermal innervation, a very marked reduction in dermal innervation and a disappearance of the innervation of the sweat glands. The classification of this HSAN more or less clearly differentiates five types from I to V [6].

CIP must be differentiated from congenital indifference to pain, described in 2006 by Cox *et al.*, [7]. In this last condition, the patients present the same clinical symptoms but the paraclinical assessment is normal. This indifference would be due to an increase in the production of endorphins in the brain with a loss of the protective thalamic mechanism which interprets traumatic attacks as painful.

Treatment is often difficult. It must be multidisciplinary, combining neurologist, surgeon, dermatologist and child psychiatrist. Very often, the prevention of these accidents and their complications is the only alternative with regular monitoring of patients.

CONCLUSION

Pain plays a fundamental role as an alarm signal contributing to human survival. The danger of CDI is thus essentially linked to serious traumatic complications, hence the importance of preventive education for patients and their families.

Conflicts of Interest: The authors declare no conflict of interest.

Author Contributions: All authors contributed to the preparation of this article. They have read and approved the contents of this article.

REFERENCES

1. van Ness Dearborn, G. (1932). A case of congenital general pure analgesia. *The Journal of Nervous and Mental Disease*, *75*(6), 612-615.
2. Axelrod, F. B., & Gold-von Simson, G. (2007). Hereditary sensory and autonomic neuropathies: types II, III, and IV. *Orphanet journal of rare diseases*, *2*, 1-12.
3. Bonin, B., Labarte, F., Van Hove, A., Picard, A., & Goga, D. (2002). Automutilations orales et insensibilité congénitale à la douleur: diagnostic et difficultés thérapeutiques: A propos d'un cas. *Revue de stomatologie et de chirurgie maxillo-faciale*, *103*(1), 30-32.
4. Redouani, L., Léauté-Labrèze, C., Ramirez de Villar, S., Taieb, A., & Sarlangue, J. (2002). Difficulty in the management of congenital insensitivity to pain. *Archives de Pediatrie: Organe Officiel de la Societe Francaise de Pediatrie*, *9*(7), 701-704.
5. Houlden, H., King, R. H. M., Hashemi-Nejad, A., Wood, N. W., Mathias, C. J., Reilly, M., & Thomas, P. K. (2001). A novel TRK A (NTRK1) mutation associated with hereditary sensory and autonomic neuropathy type V. *Annals of neurology*, *49*(4), 521-525.
6. Dyck, P. J., Mellinger, J. F., Reagan, T. J., Horowitz, S. J., Mcdonald, J. W., Litchy, W. J., ... & Lambert, E. H. (1983). Not 'indifference to pain'but varieties of hereditary sensory and autonomic neuropathy. *Brain*, *106*(2), 373-390.
7. Cox, J. J., Reimann, F., Nicholas, A. K., Thornton, G., Roberts, E., Springell, K., ... & Woods, C. G. (2006). An SCN9A channelopathy causes congenital inability to experience pain. *Nature*, *444*(7121), 894-898.