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CASE REPORT

Giant congenital melanocytic nevus, Clinical Case

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ABSTRACT

Introduction: Congenital melanocytic nevi (CMN) are skin lesions that are frequently present from birth; however, the presence of a giant CMN greater > 20 cm is infrequent and forms the basis for this case report.

Case: A boy of two years and five months, who presented congenital nevi of different diameters scattered throughout the skin area, the largest being a dark-colored embossed one in the posterior thorax area along the mid-dorsal line. The nevus rose from the skin and started from the occiput and extended through the midline until it reached the sacral region and buttocks. It covered the shoulders in an inverse triangular shape with diameters of 27 by 25 cm. It was accompanied by numerous satellite nevi ranging from 3 mm to 15 cm. The presence of two neurofibromas on the fingers was noted.

Evolution: A consultation with pediatric neurology concluded in a neurological examination without alteration, the study of brain nuclear magnetic resonance and of the spinal canal, were normal, as well as the complementary tests of hematic biometry, blood chemistry, liver and thyroid profiles, and abdominal echo. The skin biopsy reported a histological pattern of melanocytic nevus. Due to the extent of the injury, observation was decided. The pruritus was treated symptomatically.

Conclusion: CMN Syndrome is associated with multiple classic phenotypic findings among which are pigmentation patterns that occupy Blaschko's lines, neurofibromas, and multiple satellite melanomas. Diagnosis is based on clinical features, and its treatment requires surgical procedures that take into account the extent of the lesion. Comprehensive management in an interdisciplinary manner is essential for treatment of CMN.

Key Words:
MESH: Nevus, Nevi and Melanomas, Skin Neoplasms, Pediatrics.
Free text: Melanocytic Nevus Syndrome.

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INTRODUCTION

The term nevus from the Latin "naevus or lunar" is described histologically as a Hamartoma, a malformative lesion or circumscribed non-hereditary dysplasia. Melanocytic nevi are benign proliferations of melanocytes that are part of normal skin. The incidence is variable. Although it is believed that in the newborn this incidence is approximately 1%, it increases throughout childhood and puberty and acquires the maximum number of elements by 25 years of age. [1]

The excessive presence of melanocytic cells in the skin and the central nervous system was described by Virchow in 1859 and called neurocutaneous melanosis by van Bogaert in 1948. In 1972, Fox proposed the criteria that define this entity, later revised in 1991 by Kadonga and Friedman. Currently, the term "congenital melanocytic nevus syndrome" (CMN) is preferred for referring to this entity and is defined by the presence at birth of a melanocytic nevus of > 5 cm or the presence of more than one nevus of any size that is associated with neurological compromise. [2]

CMNs are considered neural crest-derived hamartomas, which are produced by postzygotic mutations that determine defects in the melanocyte migration and/or differentiation. They originate between gestation weeks 5 and 24. It has been proposed that the loss of growth regulation of melanoblasts (melanocyte precursors) during embryogenesis determines melanocyte proliferation. The earlier this process occurs, the deeper and larger the congenital melanocytic nevi will be. [3]

Nevi are found in approximately 0.2%–1.5% of all live newborns. In the case of the giant congenital melanocytic nevus, its incidence is < 1 case per 500 thousand live births.

The prevalence according to gender is three women for two men. They are more frequent in children with lighter skin.[4]

Nevi are also more common in preterm female newborns. Maternal age, the number of previous pregnancies, and birth weight do not seem to influence the frequency of appearance of congenital melanocytic nevus.[5]

CMNs have been classified by their size according to their diameter: (1) small (< 1.5 cm), (2) medium (between 1.5 and 19 cm), and (3) giant (> 20 cm). In addition, subgroups (G1, G2, G3) are proposed for every 10 cm that exceeds 20 cm, and when predicting the risk of melanoma, the existence of more than 50 satellite nevi would increase the category by one. The surface of the nevus can be smooth, nodular, hairy, plexiform, or just pigmented and corrugated.

It is essential to differentiate the nevi present at birth or in the first months of life (congenital melanocytic nevus) and those appearing during the life of the individual (acquired melanocytic nevus).

The most frequent location of a nevus is the trunk (47%) followed by the extremities (30%) and the head (22%). Lesions can be single or, more frequently, multiple. Up to 78% of patients present with satellite lesions (small CMNs) that when numerous (generally more than 20) increase the risk of melanoma or neurocutaneous melanosis. [6] They can affect any segment and frequently more than one body segment thus resembling a bathing suit. The morphology varies with age; at birth, nevi are flat, light brown neoformations that evolve into neoformations with a rough, irregular surface, with papules of different sizes, rangining in color from dark brown to black. [7]

The presence of giant CMNs, in addition to their size, have some clinical characteristics that facilitate their diagnosis, such as the almost constant presence of hypertrichosis in terminal borders on their surface, different shades of brown and black in their coloration and distribution in descending order on the trunk, the extremities, and the head. In addition, these structures frequently present surrounding melanocytic nevi, and

their importance lies in the risk of development of melanoma and neurocutaneous melanocytosis.

The diagnosis of a giant congenital melanocytic nevus is based on clinical features. The imaging test of choice for detecting lesions in the central nervous system is brain magnetic resonance imaging, which is recommended during the first six months of life, especially in the presence of ≥ 2 congenital melanocytic nevi.

Histopathological characteristics include nevic cells in the reticular dermis and subcutaneous cell tissue and between collagen fibers with different invasion patterns of skin attachments, vessels, and nerves among others.[8]

The risk of malignant transformation has been described for congenital nevi according to specific criteria: (1) small and medium 0%–4.9% and (2) giant 4.5%–10%. It is clear that individuals with giant CMNs are at higher risk of malignant transformation (melanoma). This risk varies depending on the characteristics of the nevus, such as size, depth, homogeneity, and whether it is macular or rough. In patients with congenital nevi, melanoma can develop at any age; however, the risk of malignant transformation is highest in the first years of life, and approximately 70% of melanomas are diagnosed in children under 10 years of age.

The risk of transforming any one nevus into melanoma is generally considered limited. It has been estimated that the risk of melanoma development on any individual cumulative nevus up to age 80 years would be 0.03% (one in 3,164 nevi) in men and 0.009% (one in 10,800) in women.[9]

The risk of malignancy of benign melanocytic lesions has recently been studied in a meta-analysis, with a 2% incidence of melanoma being observed, especially for congenital nevi > 40 cm and located on the trunk. [10]

Invasion of lymphatic vessels in melanocytic nevi is an incidental and infrequent finding in acquired melanocytic nevi and is somewhat more frequent in congenital melanocytic nevi. From a practical point of view, it is important to avoid a false diagnosis of malignancy. [11]

Primary melanomas of the central nervous system are unusual and are associated in up to 40% to 60% of cases with neurocutaneous melanosis. The location of the nevi on the scalp, face, neck, and/or posterior midline is associated with an increased risk of malignancy in the leptomeninges.

Neurocutaneous melanosis is believed to be related to an abnormality in the development and migration of melanoblasts in the skin and pia mater. One of the criteria for diagnosis is to present with a giant congenital melanocytic nevus at birth measuring > 20 cm in adults or multiple satellite nevi. Symptoms of cutaneous melanosis are hydrocephalus, lethargy, emesis, and irritability. Hydrocephalus is believed to be caused by impaired reabsorption of cerebrospinal fluid due to infiltration of melanocytes into the spinal cord.

Other symptoms are seizures, motor deficits, and/or cranial nerve disorders. However, it can appear in up to 25% of neurologically asymptomatic patients. Cases (40%–60%) of cases of neurocutaneous melanosis present malignant transformation of leptomeningeal melanocytes to melanoma. [12]

The association with social and psychosocial alterations has been observed in more than 50% of patients with giant CMN, who have experienced serious emotional and behavioral disorders and rejection and lack of acceptance by the family and social circle, aspects that should be considered in patients who suffer from CMN.

In general, the stigma associated with a CMN is related to the size of the nevus and its location in visible areas, especially the face. The resulting effect is related to the self-perception of their image, anxiety secondary to the risk of complications, discomfort that the different treatments imply, and the resulting scars. Parents and other relatives frequently present psychological alterations regarding the acceptance of the congenital problem in their child and its implications. Decreased social performance and increased emotional problems are observed in children with giant CMN regardless of the visibility of the nevus, whether they have received treatment or

not, and age. All of these factors imply the need for comprehensive family support.[13]

Treatment must be individualized with a multidisciplinary approach that takes into account the particularities of each patient: age of the child, size, location, depth, appearance of the lesion, ease of monitoring its evolution, risk of malignancy and appearance of neurocutaneous melanosis, risks surgery, aesthetic consequences of the injury and surgery, psychological repercussions, its treatment and family anxiety.

In a case of CMN, the primary care pediatrician plays a central role both in clinically-based diagnosis and in monitoring the evolution of the disease in all its phases and in providing information, counseling and support to the patient and their family. [14]

Numerous therapeutic options, such as laser surgery, dermabrasion, and curettage are available. Surgery is the most appropriate therapy when possible, and the surgical approach dependson the depth and size of the nevus and taking into account the risks/benefits of the surgical intervention. Excision of the congenital CMN is recommended in order to reduce the possibility of malignancy and avoid cosmetic problems. Tissue expansion has emerged as the primary treatment since excellent texture, better color, and less deformity of the donor site are obtained. [15]

For small, benign-looking nevi, observation without surgery can be considered, provided that the location allows monitoring of their evolution.

There are no established management protocols for small nevi and medical monitoring seems reasonable. For the elderly, control must be frequent, especially during the first years of life. It is important to offer a correct explanation to the parents about the probability of the appearance of melanoma, the different therapeutic options and possible complications. They must be instructed in monthly self-assessments and explained that they will require continuous follow-up every 3 or 6 months.[16]

CLINICAL CASE

The case of a male preschool patient, aged 2 years and 5 months, of mixed race, residing in Ambato, Ecuador, with no significant personal and family pathological history is described. At birth, the skin was observed to have the presence of nevi of different diameter scattered throughout the skin area. The largest nevus was a dark one in the posterior thorax area along the mid-dorsal line.

On physical examination, a large mass of dark, bumpy color was found that arose from the skin, began from the occiput, and extended along the midline until it reached the sacral region and buttocks. It covered the shoulders in an inverse triangular shape (distribution in shawl) with a diameter of 27 by 25 cm with irregular edges, hyperlobulated with hypertrichosis at the edges (**Figure 1**), and accompanied by a large number of satellite nevi that varied in size from 3 mm to 15 cm (**Figure 2**). Nevic lesions between 0.03 and 15 mm are evidenced on the face, thorax, extremities, scalp, palms, soles, and perineal region (Figure 3). In addition, neurofibromas were found on the fourth and fifth right fingers measuring 1.5 and 0.5 cm, respectively, with a purplish color (**Figure 4**). Non-bleeding scaly areas could also be seen in a giant nevus, which, according to the patient's description, caused intense itching.

To approximate the calculation of the body surface area compromised by congenital melanocytic nevus, the Lund–Browder table designed to calculate the body surface area in burned patients was used as a reference and in this patient, showed a compromise of 19% of the body surface area.

The general condition and psychomotor development were apparently normal in addition to the rest of the physical examination, including the neurological examination.

Figure 1. Congenital melanocytic nevus in shawl distribution

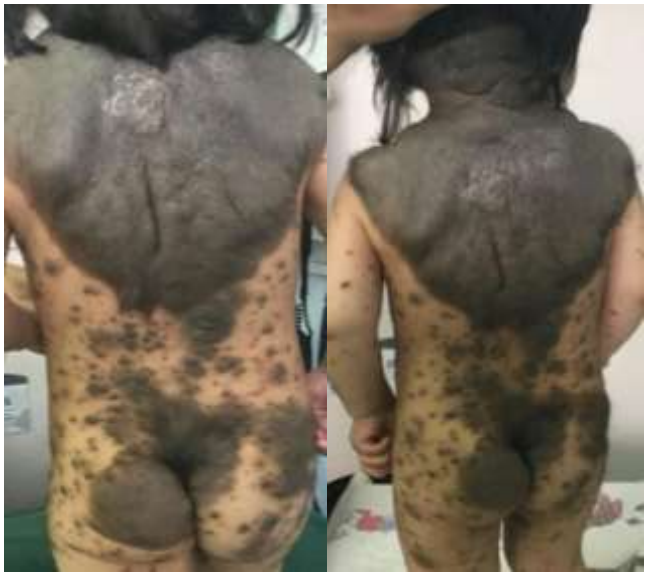


Figure 2. Patient with multiple satellite nevi of different diameter

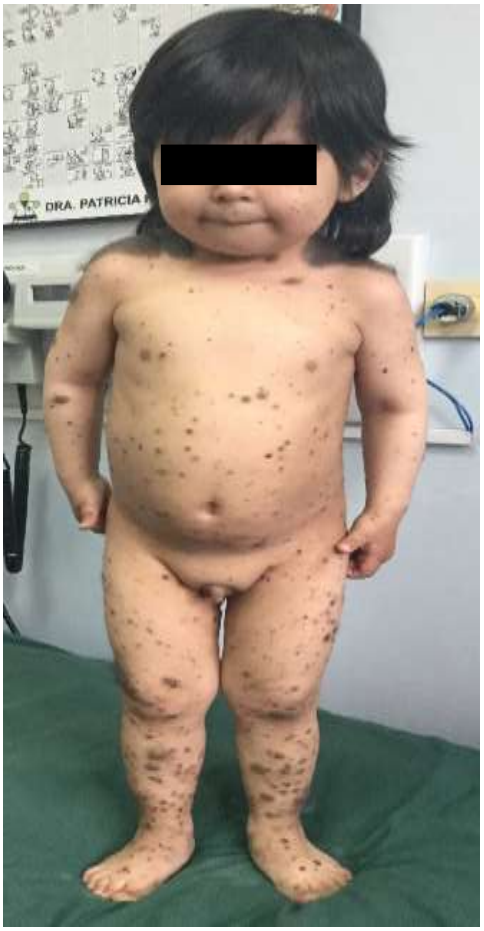
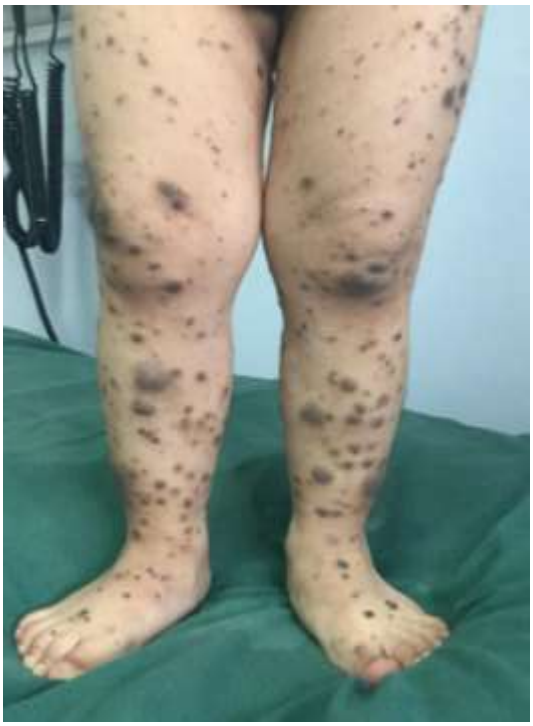


Figure 3. Satellite nevi in lower limbs of different diameter with hypertrichosis.



Evolution:

A consultation with pediatric neurology recommended a neurological examination without alteration, and the neuropediatrician physician requested a brain nuclear magnetic resonance of the spinal canal, which was normal (**Figure 5**). Complementary tests, such as hematic biometry, blood chemistry, and liver and thyroid profiles were performed and all values were within normal parameters. An abdominal echo was also performed, which was also reported normal.

In the dermatology department, a biopsy of the most representative area from the dorsal region was performed. The findings demonstrated a histological pattern with architecture of conglomerates of melanocytic cells, circumscribed intradermally, hyperpigmented and within a dense superficial reticular dermis (**Figure 6**).

Due to the extent of the injury, observation was decided for a review period. Pediatric controls have been normal so far and have been followed for six months. The pruritus was treated symptomatically and also with moisturizing creams.

DISCUSSION

Giant CMNs are characterized by pigmented skin lesions that are > 20 cm in diameter. The present case exceeded this diameter. The incidence of giant congenital melanocytic nevus is < 1 case per 500,000 live births.[1] The prevalence according to gender is three women for two men.[4] The present case describes a male patient.

According to the classification, the present case corresponds to a giant CMN G3 due to the diameter of the nevus and the existence of multiple satellite melanomas.[6-7].

In addition to the size of giant CMN, some clinical characteristics that facilitate their diagnosis, such as the almost constant presence of hypertrichosis in the terminal borders on their surface, the different shades of brown and black in their coloration, and their distribution in order decreasing on the trunk, limbs, and head are found. In addition, they frequently present round satellite melanocytic nevi [7-11]; these signs were present in this patient.

It is not uncommon to observe the development of nodules of a few millimeters to several centimeters on the surface of a nevus as in this case. The slow growth and lack of ulceration of these nodules can help guide us towards the benignity of the process. However, the development of a melanoma on the nevus can start in a similar way. Giant nevi in the head and neck area can be associated with neurological involvement due to infiltration of the leptomeningeal and other structures of the central nervous system with the appearance of hydrocephalus, mental retardation and melanomas. In this location, it is called neurocutaneous melanosis. The patient described in this report did not show any central nervous system disorders so far; however, these can appear in up to 25% of neurologically asymptomatic patients.[12] Nevi that affect the mid-dorsal area can coexist with spina bifida and other vertebral and spinal anomalies that were not present in this case.[11]

Figure 4. Neurofibromas in the fourth and fifth fingers.

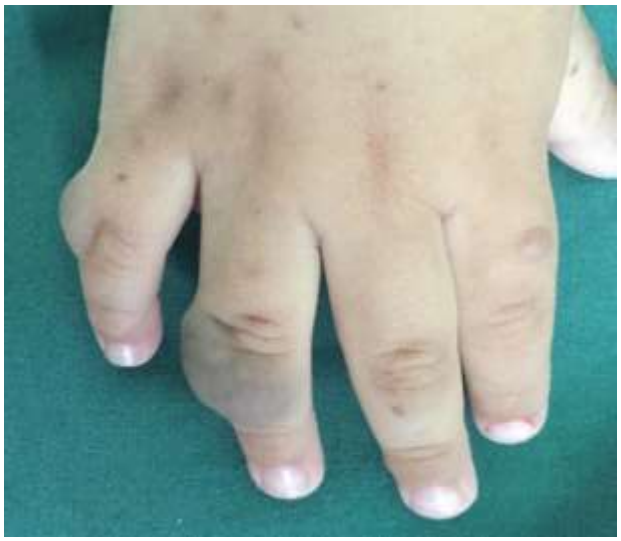
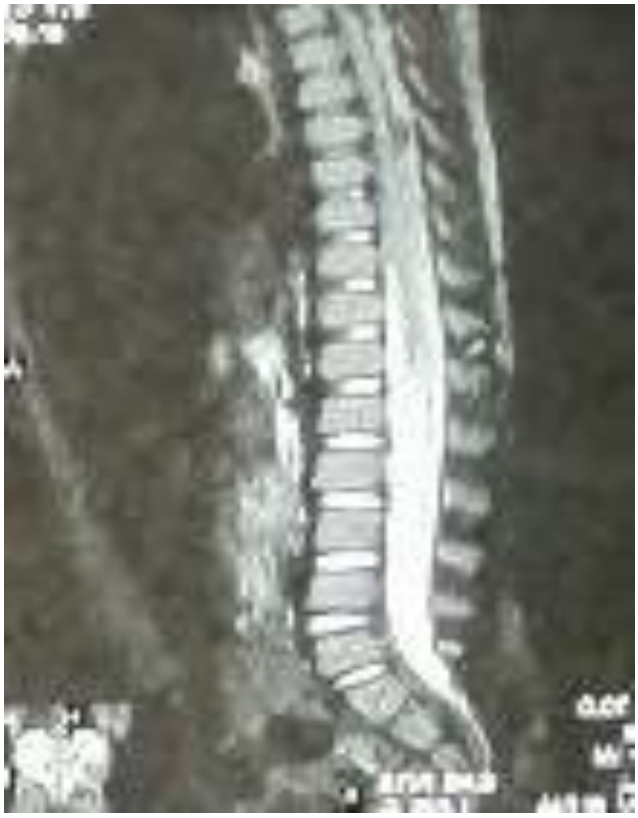


Figure 5. Nuclear magnetic resonance of the medullary canal.



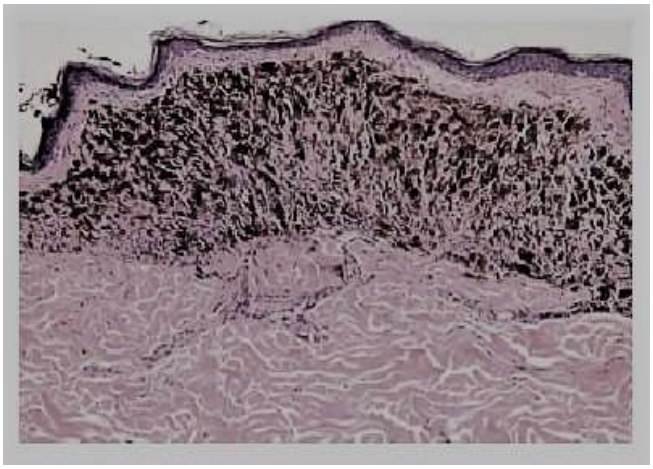
Melanoma can develop from any type of congenital nevus; however, it has been shown that the risk is directly proportional to the size of the nevus and the degree of depth as described above. So far, no malignancy has been found in our patient.[9,11]

The imaging test of choice for the detection of lesions in the central nervous system is brain magnetic resonance imaging, which is recommended in the first six months of life, especially in the presence of two or more CMN. In the present case, brain and spinal canal resonance imaging was performed, and no lesions in the central nervous system were observed (**Figure 5**).

Complementary tests, such as hematic biometry, blood chemistry, and liver and thyroid profiles were performed and all values were within normal parameters. An abdominal echo was also performed, which was also normal.

Histopathologically, a pattern with infiltrations of intradermal melanocytic cells was observed, but no malignant cells were found.[8] (**Figure 6**).

Figure 6. Histology of the lesion.



Skin fragment taken from the nevus of the dorsal region; histological pattern with architecture of conglomerates of melanocytic cells, circumscribed intradermally, hyperpigmented, within a dense superficial reticular dermis.

At present, the patient described in this report is under multidisciplinary control in which the therapeutic options will be defined according to the findings. Strict monitoring and surveillance is recommended since surgical resolution is not recommended due to the extent of skin that the nevus occupies. Monthly reviews and updating of laboratory tests, imaging studies, in addition to general, neurological and psychosocial state of the patient should be considered. [11]. A thorough physical examination will be performed to find out if there are suspicious signs of evolution to melanoma and educate the family about this disease and provide psychological support.[13]. Careful observation and reviews should be maintained to decide the best therapeutic option. A nevus recession could be carried out using the skin expander method since the evidence shows a decrease in the new melanoma formation from the melanocytic nevus with use of the option.[15] Multidisciplinary follow-up is important for the early detection of melanoma or neurocutaneous melanocytosis due to the high risk of developing these pathologies. [9,10,12]

CONCLUSIONS

CMN Syndrome is associated with multiple classic phenotypic findings, among which are pigmentation patterns that occupy Blaschko's lines, neurofibromas, and multiple satellite melanomas. Its diagnosis is based on clinical features, and its treatment requires surgical procedures after taking into account the extent of the injury. Comprehensive management in an interdisciplinary manner is essential in its treatment.

ARTICLE ADMINISTRATIVE INFORMATION

Abbreviations
CMN: Congenital Melanocytic Nevus.

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Authors' contributions
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Ethics approval and consent to participate

Not Applicable.

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The authors have the authorization for the publication of the data by the tutors of the patient.

Protection of people:
The authors have applied the components of the Singapore Declaration.

Confidentiality of the data:
The authors have technically concealed the identification data of each patient for the analysis and publication process.

Competing interests
The authors declare that they have no competing interests.

Originality of the article

The Ecuadorian Journal of Pediatrics guarantees that the article is original and without redundancy. The anti-plagiarism system of our journal reported less than 3 % similarity. The analysis is available at: https://secure.urkund.com/view/76853790-131468-596572

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