

Ectopic Mongolian spot

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The Mongolian spot is a congenital, blue-gray color with a variety of shapes and sizes, mostly located in the lumbosacral area, commonly observed in Asian and African infants but rare in Caucasians. A diagnosis is based on the clinical morphology. A histopathological examination of the lesions is required when in doubt. Mongolian spots are often present at birth, usually disappear during childhood, and are generally unnoticeable after one to four years. On the other hand, these spots persist for a long time in approximately 3%–4% of Asian populations. Darker pigmented spots located in the extrasacral area are called ectopic Mongolian spots (previously known as aberrant Mongolian spots). These are more likely to persist than the typical sacral Mongolian spots. They have a benign course, but patients with ectopic Mongolian spots in exposed areas may experience psychological distress owing to their cosmetic appearance. Herein, ectopic Mongolian spots are described in terms of their epidemiology, pathogenesis, clinical features, pathological features, and treatment.

Key words: Ectopic Mongolian spot; Lasers; Treatment

INTRODUCTION

Mongolian spot is a macular blue-gray pigmentation presents at birth in the sacral area of normal infants of darker-skinned races. This lesion is present in more than 90% of infants of the Mongolic race, with the highest incidence in the Mediterranean region. Pigmentation is macular, diffuse, more or less uniform, slate blue to gray, and usually relatively faint. The patches are usually rounded or oval in shape and are usually single but occasionally multiple [1].

The lumbosacral region is a common site, and the buttocks and flanks may be affected by extensive lesions. Pigmentation develops during fetal life, increases in depth for a period after birth, and diminishes thereafter. It usually disappears during the first decade but occasionally persists into adulthood [2].

Ectopic Mongolian spots are variants of common Mongolian spots, a type of congenital dermal melanocytosis. These spots are characterized by their presence in atypical locations such as the arms, legs, and other parts of the body, unlike the typical lumbosacral region. Unlike typical Mongolian spots that often fade during childhood, ectopic spots can persist into adulthood. As results, extralumbosacral variants tend to be more persistent, and thus are easily confused with adult-onset dermal melanocytoses, nevus of Ota, nevus of Ito, and patch blue nevus. They have been associated with genetic disorders such as lysosomal storage diseases, including Hurler's disease and mucopolipidosis. Understanding the characteristics, implications, and treatment options is crucial for accurate diagnosis and management.

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EPIDEMIOLOGY

Mongolian spots are highly prevalent in certain ethnic groups, with >90% occurring in Mongolians and Native Americans, approximately 80% in Asians, and significantly lower in Caucasians (<10%). A study in Tehran found Mongolian spots in 37% of the neonates, with the sacral area being the most common site, followed by the gluteal area [3].

Ectopic Mongolian spots, which appear in nontraditional locations such as the extremities, are less frequently documented. In a study of 2,313 infants, the lower extremities were noted to be the most common extrasacral site for Mongolian spots [4].

PATHOGENESIS

The pathogenesis of ectopic Mongolian spots involves a complex interplay of genetic, embryological, and possibly metabolic factors and may be associated with various syndromes and metabolic disorders. This overview explores the pathogenesis of ectopic Mongolian spots and highlights their embryological origins and association with metabolic disorders. As embryological origins, Mongolian spots result from the migration of neural crest-derived melanocytes into the skin during embryo-

genesis. These melanocytes fail to reach the epidermis and remain in the dermis, indicating a possible variation in the migration pattern of melanocytes, which leads to a characteristic bluish-grey pigmentation [5].

In association with metabolic disorders, ectopic Mongolian spots have been related to lysosomal storage disorders such as GM1-gangliosidosis and mucopolysaccharidoses. These conditions are characterized by lysosomal storage issues and enzyme deficiencies that lead to the cellular accumulation of complex molecules, affecting various tissues including the skin, leading to the persistence and unusual distribution of Mongolian spots. In GM1-gangliosidosis, extensive Mongolian spots are diagnostic clues, often accompanied by other symptoms, such as developmental delay and hepatosplenomegaly.

In cases of Hurler's disease, a type of mucopolysaccharidosis, atypical Mongolian spots may serve as early indicators of the disorder, prompting further investigation and early intervention. Similarly, mucopolipidosis II, another lysosomal storage disorder, has been associated with extensive Mongolian spots, suggesting its potential as a diagnostic marker for these metabolic conditions [6].

Kleefstra syndrome, caused by *EHMT1* mutations, has been reported to be associated with multiple Mongolian spots, suggesting an association between genetic mutations and the persistence of these birthmarks [7].

CLINICAL FEATURES

While Mongolian spots are commonly found in the lumbosacral area (Fig. 1), ectopic Mongolian spots may appear in unusual locations, such as the ankles, abdomen, back, arms, and legs (Fig. 2). These atypical spots do not always fade in early childhood and may persist or even increase in number. Ectopic Mongolian spots vary significantly in size, with some extensive lesions covering large areas of the body. Pigmentation is typically bluish-grey, similar to typical Mongolian spots, but it can be darker and more pronounced in ectopic cases.



Fig. 1. Typical Mongolian spot that appeared on the lumbosacral region and buttock from birth in a 2-year-old girl.

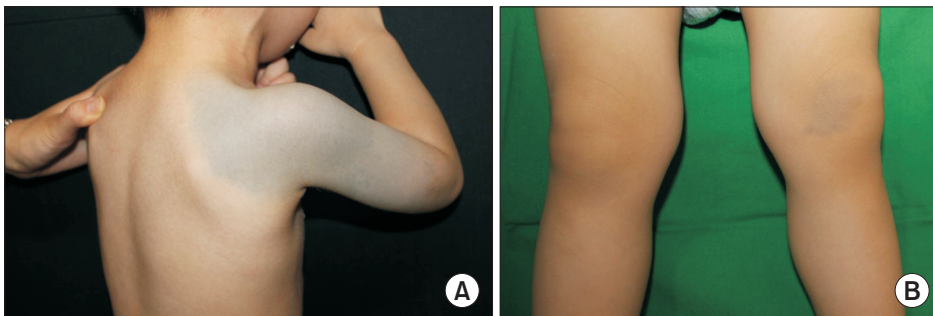


Fig. 2. Ectopic Mongolian spots occurring in atypical areas such as (A) shoulders, arms, and (B) legs in infant age.

Owing to their appearance, ectopic Mongolian spots can be mistaken for bruises or signs of physical abuse, leading to unnecessary investigations and stress for families. Healthcare professionals must differentiate these spots from injuries to avoid misdiagnosis. Differential diagnoses should consider other causes of pigmentation, such as acquired pseudo-Mongolian spots from medications such as minocycline, which can cause similar pigmentation changes.

Adult-onset Mongolian spots, also known as persistent Mongolian spots, are rare phenomena in which congenital dermal melanocytoses do not fade during childhood but persist into adulthood. Although typically benign, their persistence or unusual presentations can be associated with underlying genetic or metabolic disorders. A study on Chinese adults reported cases of persistent Mongolian spots, highlighting that such occurrences, although rare, do exist and can be found on various body parts like arms and shoulders [8]. In a Japanese study, persistent Mongolian spots were observed in approximately 4% of adults, indicating a potential ethnic or genetic predisposition [9].

PATHOLOGY

Mongolian spots are histologically characterized by the presence of spindle-shaped melanocytes in the lower dermis. These melanocytes fail to migrate to the dermal-epidermal junction during fetal development [6]. Dermal melanocytosis is a key histological feature distinguishing Mongolian spots from other pigmented lesions. Dermo-

scopic examination of the lesion reveals a homogeneous pattern without local structures. These lesions consist of widely scattered, inconspicuous, and dispersed melanocytes among collagen fibers and around the neurovascular bundles in the dermis. They run parallel to the skin surface and contain fine granules of melanin [10].

TREATMENT

Table 1 summarizes various treatment methods for Mongolian spots. Natural resolution is the most commonly used approach for this purpose. Laser therapy is the most promising treatment option for patients with persistent condition. Combination therapies and topical agents are less commonly used, but can be considered in specific cases.

LASER TREATMENT

The treatment of Mongolian spots, particularly those that persist beyond infancy or appear at atypical locations, involves various laser therapies. These treatments aim to reduce pigmentation and improve the cosmetic appearance. The most commonly used lasers include the Q-switched Nd:YAG, ruby, and alexandrite, each with varying degrees of efficacy and side effects. Intense pulsed light (IPL) therapy has been explored as an alternative treatment option. We discuss the specifics of these treatments and their outcomes, as shown below.

The Q-switched Nd:YAG laser, particularly at a wavelength of 1,064 nm, has shown significant efficacy in

Table 1. Treatment methods for Mongolian spots

Treatment method	Description	Reference
Observation and natural resolution	Most Mongolian spots naturally fade over time without any intervention. This is the most common and recommended strategy for benign and asymptomatic spots	[2]
Laser therapy	Laser therapy, specifically with the Q-switched Nd:YAG laser, targets the melanin within the dermal layer, breaking down the pigmented cells and allowing the body to naturally eliminate the pigment Q-switched alexandrite laser and other laser modalities have been used to treat persistent Mongolian spots. These therapies target pigmentation and help lighten the lesions. Effectiveness varies and multiple sessions may be required An important consideration is the treatment of ectopic Mongolian spots in childhood, when the skin is more resilient and the spots have a higher likelihood of responding positively to laser therapy Early intervention with Q-switched lasers (ruby, alexandrite, Nd:YAG) has been effective, though they require careful management to prevent long-term complications	See Table 2 for more details
Topical agents	Though not commonly used, topical agents such as hydroquinone or other skin-lightening creams have been explored. However, their effectiveness is limited, and they are generally not recommended for young children owing to potential side effects. Bleaching creams are recommended in the treatment of persistent sacral Mongolian spots in adults	[17]
Combination therapies	In some cases, combination therapies involving both laser treatments and topical agents may be considered for more resistant lesions. The combination aims to enhance overall effectiveness	[18]

Table 2. Details of various treatment methods for ectopic nevi

Treatment method	Description and efficacy	Side effects	Reference
High-fluence 1,064 nm Q-switched Nd:YAG	Applied without tissue whitening to treat ectopic Mongolian spots Significant improvement with statistically better results compared with the observation group	Well-tolerated with minimal side effects	[1]
Q-switched ruby laser	Fifty-three patients (16 males and 37 females) were treated in a previous study Treatment in the exposed regions that cannot be concealed by clothing is more effective than AMS in the non-exposed regions that can be concealed by clothing	Post-treatment hyperpigmentation in one patient	[12]
Q-switched alexandrite laser	Treatment of Mongolian spots; results compared by age and spot location Overall good outcomes; sacral spots are more resistant with better results in younger patients (Mongolian spots of 16 Japanese patients aged ≥ 14 years are studied)	Post-inflammatory hyperpigmentation and hypopigmentation were observed in two patients	[14]
Q-switched laser (various types)	Used for ectopic Mongolian spots in childhood Effective, especially early in life; complications include hyperpigmentation and hypopigmentation	Special care needed to prevent long-term complications	[13]
Intense pulsed light	Seven patients (four males and three females) with a mean age of 2.0 years (range, 0-7 years) were studied Evaluation of the effect of treatment by a physician was less favorable, with excellent, good, and improvements in one (14%), two (29%), and four (57%) patients, respectively	No comment	[15]
755 nm picosecond laser	Lesions almost disappeared after 1-2 sessions in three ectopic Mongolian spots and one Mongolian spots	Reported minimal	[16]

AMS, aberrant Mongolian spots.

treating ectopic Mongolian spots [1,11]. A study involving 61 patients demonstrated that high-fluence Nd:YAG laser treatment resulted in a significant reduction in pigmentation (mean 5-quintile grading scale: treatment group 2.85 ± 1.00 vs. observation group 0.49 ± 0.73 , $p < 0.001$ vs. $p < 0.001$), as measured by a melanin index (significant difference observed in the treatment group, 156.7 ± 78.4 vs. observation group, 7.1 ± 62.7 , $p < 0.001$), compared with untreated lesions [1].

The Q-switched ruby laser is another effective treatment for ectopic Mongolian spots, particularly those located in exposed areas, where its color improves with fewer complications than in non-exposed areas. However, treatment efficacy can vary based on spot location, with sacral spots being more resistant to treatment than extrasacral spots [12].

The Q-switched alexandrite laser has been successfully used to treat persistent Mongolian spots and has achieved good therapeutic outcomes. However, it is associated with risks of post-inflammatory hyperpigmentation, particularly in adult patients. The treatment is more effective when initiated before the age of 20 years, and careful consideration of treatment intervals and adjunctive therapies, such as bleaching creams, is advised to minimize side effects [13,14].

IPL therapy has been explored as a treatment for aberrant Mongolian spots with varying degrees of success. In

a study of seven patients, IPL treatment resulted in good-to-excellent improvements in pigmentation in the majority of cases, although physician evaluations were less favorable. Histopathological examination of the pigmented region revealed the typical features of a Mongolian spot in the hematoxylin-eosin-stained section. No changes were observed in the dermis immediately after IPL treatment. However, 6 months after treatment, the number of melanocytes in the middle and upper dermis decreased [15].

The efficacy of picosecond lasers, particularly those with a 755 nm wavelength, has been recently reported with significant advantages for the treatment of dermal melanocytosis, including ectopic Mongolian nevi. Rapid clearance is a marked improvement over traditional Q-switched laser therapies, which typically require longer treatment [16].

Although laser treatments are effective in reducing the pigmentation of Mongolian spots, they are associated with risks. Post-inflammatory hyperpigmentation is a common side effect, particularly in adults, and careful management of treatment intervals and adjunctive therapies is necessary to mitigate these risks. Additionally, the persistence of Mongolian spots can sometimes be indicative of underlying conditions such as mucopolysaccharidosis, highlighting the importance of a comprehensive clinical evaluation.

Table 2 summarizes the various treatment methods for ectopic Mongolian nevi, including their descriptions, efficacies, side effects, and relevant references.

NON-LASER TREATMENTS

Non-laser treatments, such as topical retinoids and traditional Mongolian medicine, have also been explored, although their efficacy, specifically for Mongolian spots, is less documented. Although not commonly used, topical agents such as hydroquinone or other skin-lightening creams have been explored. However, their effectiveness is limited, and they are generally not recommended for young children because of their potential side effects [17,18].

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AUTHOR CONTRIBUTIONS

All work was done by KY.

CONFLICT OF INTEREST

Kwangho Yoo is the Editor-in-Chief of the journal, but was not involved in the review process of this manuscript. Otherwise, there is no conflict of interest to declare.

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DATA AVAILABILITY

None.

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SUPPLEMENTARY MATERIALS

None.

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