International Journal of Research in Medical Sciences
Torres JAM et al. Int J Res Med Sci. 2024 May;12(5):1720-1723
www.msjonline.org
pISSN 2320-6071 | eISSN 2320-6012

DOI: https://dx.doi.org/10.18203/2320-6012.ijrms20241263

Case Report

CT findings of a patient with Hay-Wells syndrome: a case report

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Received: 10 March 2024
Accepted: 02 April 2024

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ABSTRACT

Hay-Wells syndrome is a rare genetic disorder caused by a heterozygous mutation in the TP63 gene that affects the development of ectodermal derivatives. While the exact prevalence of hearing loss in this condition is not well established, it has been suggested that approximately 50% of patients may present with varying degrees of hearing impairment. Here we present a case of a 6-year-old female patient diagnosed with Hay-Wells syndrome since birth who has been developing hearing loss predominantly on the right side for the past year. Considering the embryological origin of the external ear, an associated abnormality with the syndrome cannot be disregarded. Early recognition and diagnosis of this syndrome are crucial for appropriate management and potential interventions for hearing loss. Utilizing imaging methods can aid in demonstrating the characteristic alterations of the syndrome, which is important given its low incidence in the general population. This case report emphasizes the significance of prompt diagnosis, which is critical for implementing genetic counseling and effective disease prevention measures. We should be well-versed in the characteristic imaging findings that aid in early identification and genetic diagnosis of the disorder.

Keywords: Hay-Wells syndrome, CT imaging cleft lip/palate, Hearing loss, Ectodermal dysplasia

INTRODUCTION

Hay-Wells syndrome, also known as ankyloblepharon-ectodermal dysplasia-cleft lip/palate syndrome, is a rare genetic disorder caused by a heterozygous mutation in the tumor protein 63 (TP63) gene, located on chromosome 3q28.[1] While its exact prevalence is unknown, it affects both sexes equally and can be sporadic or inherited in an autosomal dominant manner.[2] The syndrome presents as congenital defects or alterations in the development of ectodermal derivatives (hair, teeth, sweat glands, skin, and nails), and is characterized by cleft lip and/or palate, alopecia, onychodystrophy, oligodontia, and hypohidrosis. One of the associated clinical features of Hay-Wells syndrome is hearing loss, the exact prevalence of this manifestation is not well established. Diagnosis is based on clinical signs and confirmed by genetic analysis.[3] The aim of this case report is to utilize imaging methods to demonstrate the characteristic alterations of the Hay-Wells syndrome, given its low incidence in our population, and to provide a comprehensive understanding of the syndrome's clinical presentation and diagnosis, including the potential for hearing loss which is not well known as a rare manifestation of the disorder.

CASE REPORT

A clinical case is presented of a 6-year-old female patient diagnosed with Hay-Wells syndrome since birth, who has undergone five surgeries for cleft lip and palate. She presented with bilateral hearing loss predominantly on the right side for the past year, with progressive deterioration in her auditory capacity. During the clinical evaluation, oligodontia was detected, characterized by the presence of only one central incisor and one canine, as well as maxillary protrusion treated with orthodontics. Alopecia was evidenced in the frontal region and hair insertion areas, as well as koilonychia in the hands and feet (Figure 1). Proper evaluation of the tympanic membrane could not be performed. Because a hypoacusis was suspected by the clinician, a CT scan of the ears was requested.

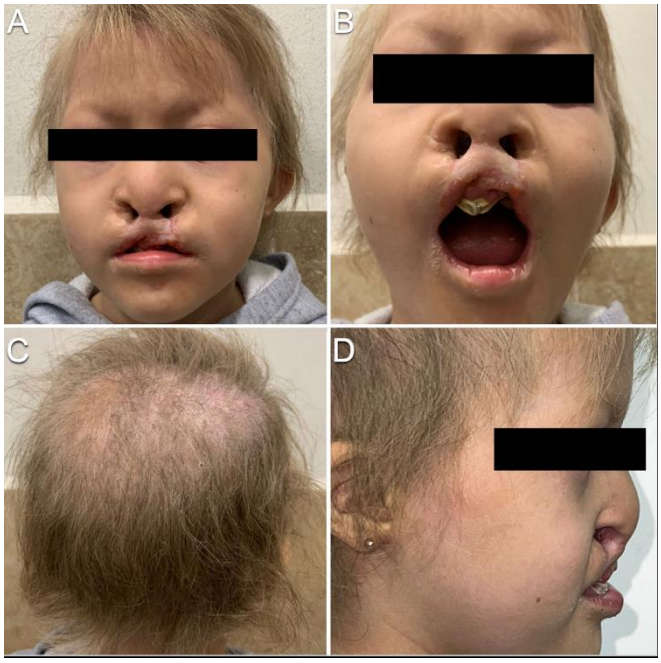

Figure 1: (A and D) The images presented herein depict the stigmata of Hay-Wells syndrome. (B) Post-operative changes resulting from surgical intervention for cleft lip and palate as well as nasal retraction are evident at the superior labial level. (C) Oligodontia with orthodontic intervention for central and canine teeth is also apparent. Additionally, central-predominant alopecia is present.

Imaging findings

High-resolution tomography with axial sections and multiplanar reconstructions was performed, using both simple phase and bone window techniques, from the base of the cochlea to the superior semicircular canal and from the anterior margin of the attic to the posterior semicircular canal. The following findings were identified:

The external auditory canals showed only the cartilaginous segment, with a complete absence of the bony portion. The tympanic membrane was not clearly identifiable. Soft tissue densities (76-92 HU) surrounded the left malleus and incus and occupied the bilateral Prussak space, extending towards the epitympanum and mesotympanum on both sides and only the epitympanum on the right side. The mastoid air cells displayed an absence of pneumatization and were occupied by soft tissue densities. The right aditus ad antrum measured 2.3 mm, while the left measured 1.6 mm. The hypotympanum, ossicular chains, oval and round windows, otic capsule, semicircular canals, vestibule, cochlea, promontory, internal auditory canals, and sickle ridges exhibited normal tomographic appearances (Figure 2 and 3).

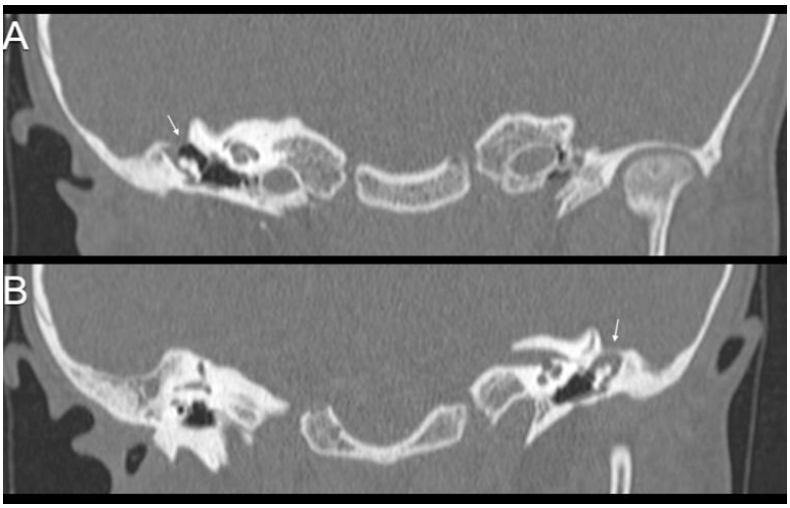

Figure 2: NECT of the ears. (A) Right and (B) left view. The coronal section of the patient in the bone window reveals both external auditory canals with the absence of the osseous portion but preservation of the cartilaginous portion. The arrow indicates bilateral soft-tissue densities occupying the middle ear cavity.

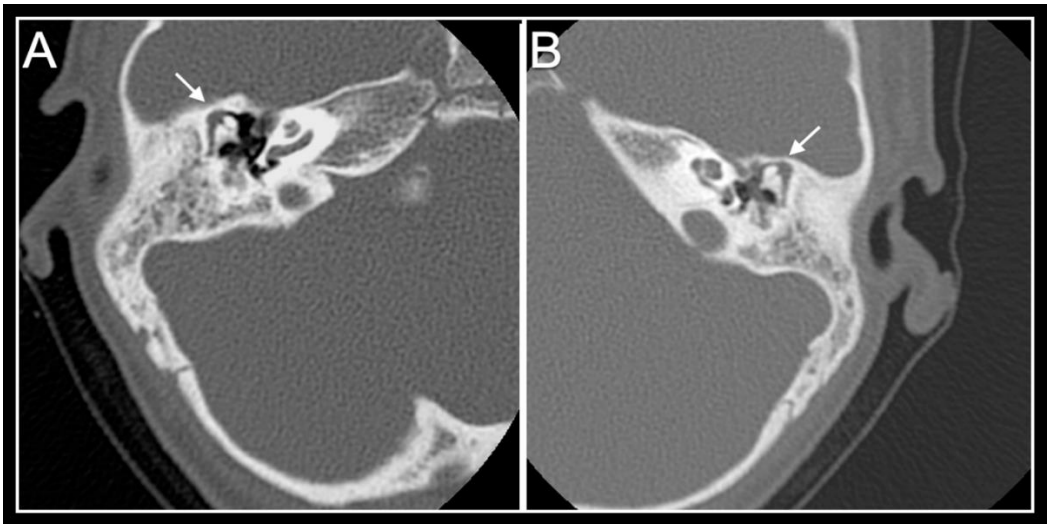

Figure 3: NECT of the ears. (A) Right and (B) left view. Axial section of the patient in bone window provides a more detailed visualization of the soft tissue occupation in the bilateral Prussak's space indicated by the arrows (B), enveloping the left malleus and incus, and extending towards the epitympanum and mesotympanum, while only occupying the epitympanum on the right side (A) with no evidence of adjacent bone remodeling. The bilateral mastoid air cells show a complete absence of pneumatization and soft tissue occupation.

DISCUSSION

Ectodermal dysplasias constitute a heterogeneous group of conditions characterized by alterations in the development of ectodermal structures (hair, salivary glands, skin, and nails), with the most evident being filiform ankyloblepharon adnatum, cleft lip/palate, and the classic presentation of erythroderma with skin scaling and superficial erosions, predominantly on the scalp, with a potential risk for the appearance of infections, scarring alopecia, and hypotrichosis.[4] Other findings often present are agenesis or atresia of the lacrimal duct, which often produces chronic conjunctivitis or blepharitis, nail dystrophy, hypodontia, and conical teeth. Although rare, atresia of the auditory canal, supernumerary nipples, hypospadias, and cardiac defects may also occur.[1]

The diagnosis of Hay-Wells syndrome is confirmed by TP63 sequencing analysis. It results from a heterozygous "nonsense mutation in the TP63 gene" involved in embryonic ectoderm development. It has an autosomal dominant pattern, with 70% of cases being de novo mutations.[5,6]

Approximately 50% of individuals with Hay-Wells syndrome may have some degree of hearing impairment, although the exact prevalence of hearing loss in this condition is not well known. Conductive hearing loss (CHL) may indirectly result from a cleft palate due to various factors, including missing cilia, inflammation, abnormal Eustachian tube structure, and recurrent infections due to communication between these structures. The etiology of CHL in these patients is likely multifactorial. Given the importance of hearing in speech development, it is crucial to evaluate hearing in these at-risk patients.[7-9]

The management of these patients should involve a multidisciplinary approach to prevent associated morbidities and complications, including infections. Furthermore, the growth, development, and integration of these patients must be assessed and promoted. Individualized and multidisciplinary follow-up is necessary for affected individuals and their families. The prognosis of ectodermal dysplasia is influenced by the phenotype of the dysplastic syndrome and any associated complications.[6-9]

CONCLUSION

Performing a computed tomography (CT) scan to assess for anatomical anomalies in the ear can aid in the early detection and prevention of neurological development in patients diagnosed with Hay-Wells syndrome. To ensure timely detection of hearing loss in this condition, it is important to utilize all possible approaches, including imaging techniques. Delayed-onset hearing loss may occur due to preservation of bone conduction combined with missing cilia, inflammation, abnormal Eustachian tube structure, and recurrent infections, along with a lack of screening. Therefore, identifying potential abnormalities in the ear through CT scans can be crucial for early diagnosis and effective management of hearing loss in patients with Hay-Wells syndrome. CT is the method of choice to confirm anatomic problems of the ear in these patients. This case report emphasizes the significance of prompt diagnosis, which is critical for implementing genetic counseling and effective disease prevention measures such as hearing loss. Therefore, it is essential for us to be well-versed with the characteristic imaging findings that aid in early identification and genetic diagnosis of the disorder.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

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Cite this article as: Torres JAM, Escobar BF, García FJJ. CT findings of a patient with Hay-Wells syndrome: a case report. Int J Res Med Sci 2024;12:1720-3.