عنوان مقاله:

H Syndrome Masquerade as Rheumatologic Disease

محل انتشار:

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نویسندگان:

Mohammad Razmyar - *Assistant of Dermatology, Cutaneous Leishmaniasis Research Center, Mashhad University of Medical Sciences, Mashhad, Iran.*

Zahra Rezaieyazdi - *Professor of Rheumatology, Rheumatic Diseases Research Center, Mashhad University of Medical Sciences, Mashhad, Iran.*

Naser Tayebi Meibodi - *Professor of Pathology, Fellowship in Dermatopathology, Cutaneous Leishmaniasis Research Center, Mashhad University of Medical Sciences, Mashhad, Iran.*

Zahra Fazel - *Dermatologist, Mashhad, Iran.*

خلاصه مقاله:

Background H syndrome is an autosomal recessive genodermatosis with a low prevalence which is caused by a mutation in SLC29A3 gene. This disorder is characterized by sclerotic, hyperpigmented, hypertrichotic cutaneous plaques with systemic involvement including: hepatosplenomegaly, heart anomalies, hearing loss, hypogonadism, low height, and hyperglycemia. Case Presentation Here we have presented two cases of H syndrome that have been misdiagnosed and mismanaged as rheumatologic disease. The first case had been represented with sclerotic skin lesions and diagnosed as morphea, and second one with chronic and recalcitrant to treatment arthritis as juvenile idiopathic arthritis. Conclusion H syndrome is an autosomal recessive genodermatosis that has been recently recognized with a variety of manifestations and overlapping features with other diseases. Increase the knowledge of physicians for wide spectrum manifestations of this syndrome along with reporting the misdiagnosis of this condition can increase the accuracy of physicians for its better identification. This time our cases masquerade as rheumatologic diseases.

کلمات کلیدی:

Children, H syndrome, Genodermatosis, SLC29A3 gene mutation