Ayalew Moges, Oumer Harun, Tinsae Alemayehu. *Ethiop Med J, 2019, Vol. 57, No. 1*

CASE REPORT

A CLUSTER OF SEIZURES IN A CHILD WITH HYPOMELANOSIS OF ITO: A RARE DISORDER OF MELANOBLAST MIGRATION

Ayalew Moges, MD[1], Oumer Harun, MD[1] ,Tinsae Alemayehu MD[1 *]

ABSTRACT

Hypomelanosis of Ito is a rare multisystem neuro-cutaneous disorder. There are no published reports from African children. We report a ten month old female infant who presented with a cluster of generalized tonic clonic seizures of three months. She also had visual defects and was responding to sound. Physical examination revealed linear and whorled hypo-pigmented lesions along the lines of Blaschko distributed over her right upper limb and trunk and marked hypotonia. Recognition of the characteristic hypopigmented skin lesions with associated neurologic complications should alert clinicians to diagnose Hypomelanosis of Ito and initiate early treatment.

Keywords: *Hypomelanosis of Ito, Blaschko lines, Neuro-cutaneous, Child, Ethiopia*

INTRODUCTION

The first description of Hypomelanosis of Ito (HI) was made in 1952, when M Ito described a woman with a distinctive pattern of whorls and streaks of hypopigmentation over the trunk and extremities and gave the disorder the name Incontinentia Pigmenti Achromicans (1). Hypomelanosis of Ito (HI), as it is now called, is the third commonly diagnosed multi-system neuro-cutaneous disorder with an incidence of 1 in 3000 children (2).

HI may be congenital or acquired. Its cutaneous lesions are characteristic. Various patterns of distribution, shapes and colors of lesions may be seen but they generally follow the lines of Blaschko. Most patients have linear unilateral or bilateral lesions, and occasional patchy lesions. Lesions are usually hypochromic. Mental retardation and epilepsy are seen in more than 50% of cases. One in ten patients have infantile spasms during infancy and another 10% exhibit autistic behaviors. Other complications includehypotonia, macro- or microcephaly and congenital cardiac, urological or genital malformations (3). Autosomal dominant, autosomal recessive and X -linked dominant modes of inheritance as well as sporadic occurrences have been reported (4).

There are few available data on the epidemiology of HI in Africa. We report an infant with Hypomelanosis of Ito manifesting with typical skin lesions, intractable seizures, developmental regression, blindness, and hearing loss.

CASE REPORT

A ten month old female infant presented to Tikur Anbessa Specialized Hospital, Addis Ababa, Ethiopia after being referred for evaluation of a cluster of seizures which started at the age of seven months occurring at a rate of 10 - 15 times per day. She had been evaluated at her local hospital where she was started on Phenytoin and Phenobarbitone, progressively escalated to 9.3 mg/kg and 7.5 mg/kg respectively over one month.

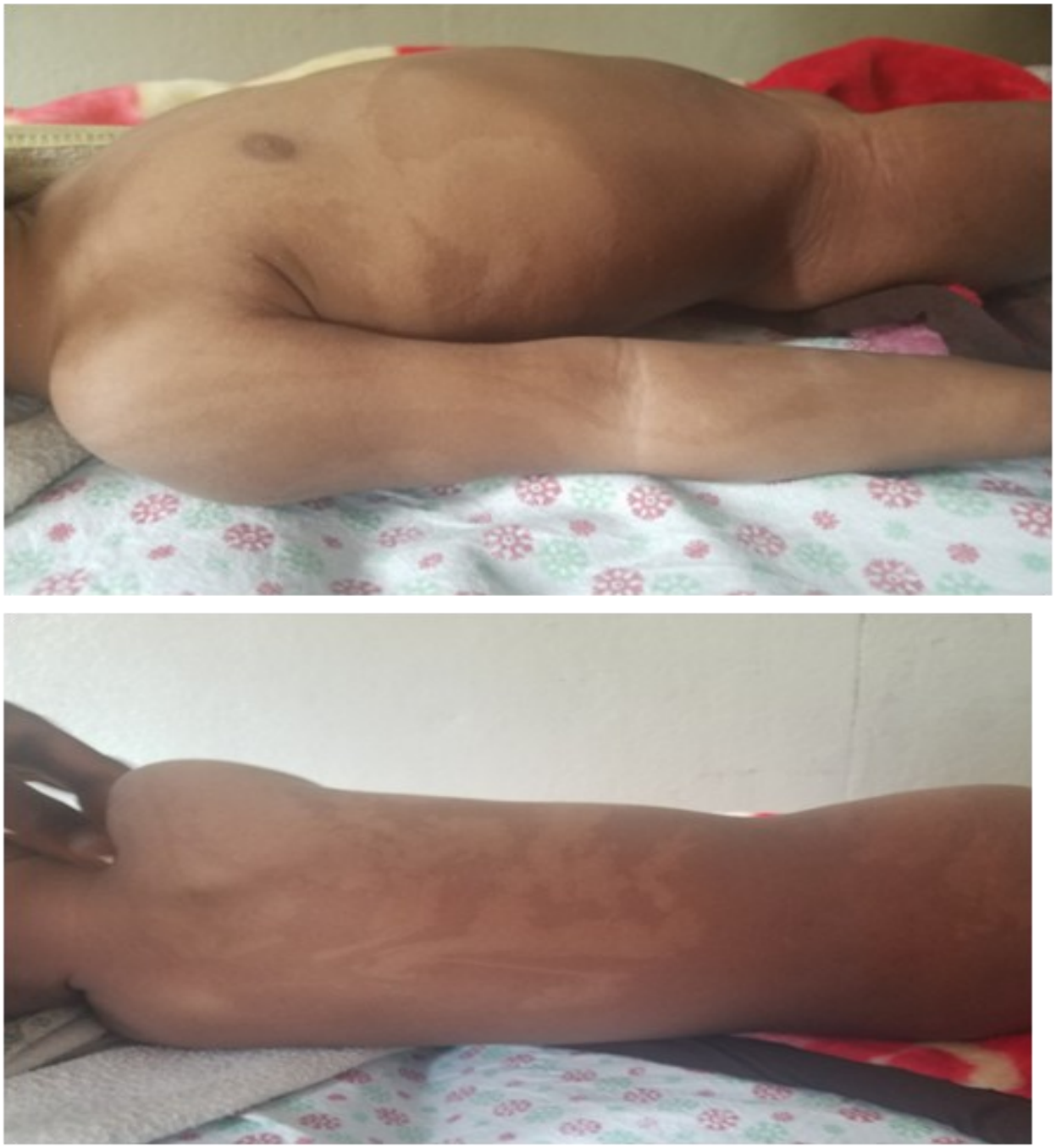
She was born to a non-consanguineous couple and she is the seventh child for her family. All other siblings are healthy. There was no history of mental retardation or epileptic disorders in her family. Her mother had an uneventful pregnancy and an uncomplicated delivery at term. She had supported her head at the age of three months, was rolling backwards on her fifth month, and started sitting without support at her seventh month before her illness started and she regressed developmentally. She had not yet started to crawl or say baba or mama and at presentation was not following follow objects with her eyes, recognizing her mother, or responding to sound stimuli.

Following her delivery, her parents noticed localized hypo-pigmented skin lesions on her trunk and right arm but had not sought medical attention as it had not shown any changes in size. She never had any bullous, verrucous or hyper-pigmented lesions at any age prior to her presentation which preceded the hypopigmentation.

[1]Department of Pediatrics and Child health, Addis Ababa University.
* Corresponding Author. E-mail: tinsae.alemayehu@aau.edu.et

On examination, she had no dysmorphism and anthropometric measurements were normal. There was no scoliosis or visible thoracic and limb deformities.

She was fair skinned and her cutaneous examination showed whorled, macular, hypo pigmented areas along the lines of Blaschko on the right side of the body involving the trunk both anterior and posterior aspect extending to the right upper limb and also over buttock and lower right limb (Figure 1 and 2).



Figures 1 and 2. Skin lesions of the patient on the anterior and posterior sides of the body

She had marked muscular hypotonia with head lag and she didn't follow movements nor respond to sounds. Baseline hematologic tests were within normal limits. An electroencephalograph (EEG) showed an abnormal wake tracing with persistent primary generalized epileptiform discharges noted throughout the recording time with generalized background slowing which was suggestive of epileptic encephalopathy (Figure 3).

MRI scan of head was normal. Parents couldn't afford chromosomal studies. A skin biopsy was not performed.

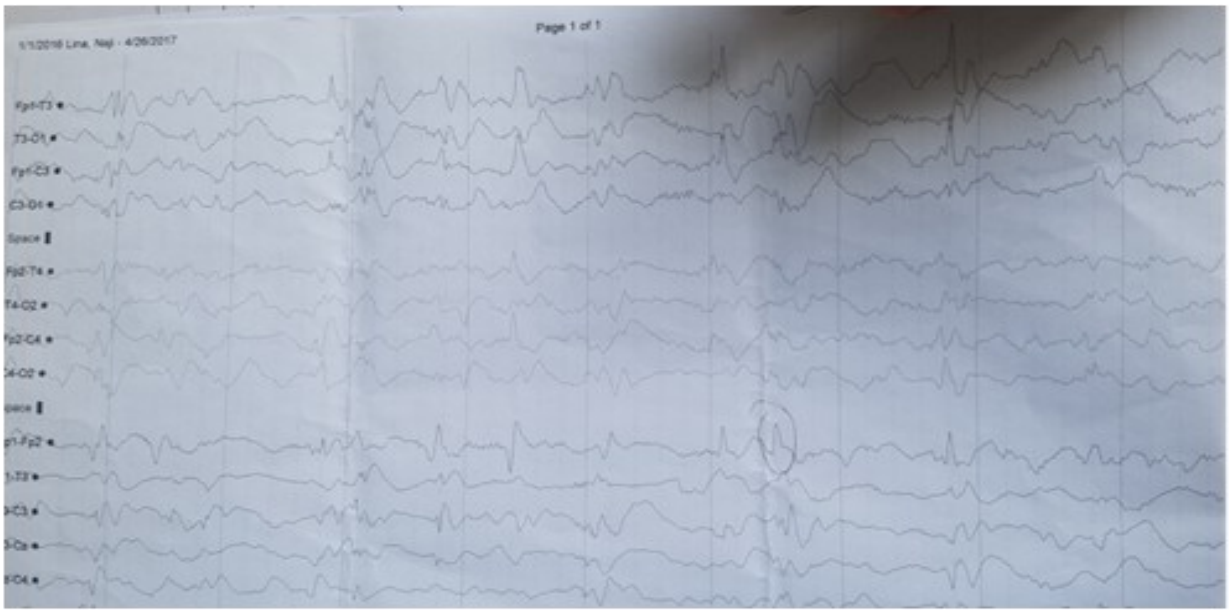


Figure 3: EEG of our patient

A diagnosis of Hypomelanosis of Ito was made and she was loaded with Phenytoin 10 mg/kg followed by a maintenance dose of 8 mg/kg/day. Phenobarbital was slowly tapered and discontinued. But the frequency of her seizures markedly reduced after the initiation of Valproate 15 mg/kg/day. Referrals for ophthalmologic and otolaryngologic assessments were made and she was appointed for follow-up at the pediatric neurologic clinic.

DISCUSSION

Hypomelanosis of Ito is a neurocutaneous disorder of melanoblast migration. Diagnostic criteria for HI include a congenital or early acquired non-hereditary cutaneous hypopigmentation in linear streaks or patches involving more than two body segments; one or more nervous system or musculoskeletal anomalies from the major criteria and two or more congenital malformations other than nervous system or musculoskeletal system or chromosomal anomalies constituting the minor criteria (5).

Patients usually have unilateral skin lesions which are contralateral to the side of the brain malformation. They are easily discernable in the black population (6). Neurological disorders are the most frequently associated extra-cutaneous anomalies, seen in three-fourth of patients. These consist of ataxia, intellectual disability and seizures (most frequent findings); microcephaly, mental retardation, hypotonia, hyperkinesia, hydrocephaly, ataxia, speech defects, motor retardation, cerebral asymmetry, breath holding, myelo-meningocele, cerebral atrophy, hypertonia, and sensorineural hearing loss (3,7).

Seizures appear commonly early in the first year of life with the most frequent types being generalized tonic or tonic-clonic, complex partial, myoclonic seizures and infantile spasm (8). While some patients suffer from generalized seizures that are well controlled with anti-convulsant therapy, many have severe, pharmaco-resistant focal seizures, and some benefit from surgery (9).

This is the first report of Hypomelanosis of Ito in an Ethiopian child. Any child with characteristic hypopigmented cutaneous lesions early in life and neurologic disorders needs an evaluation for HIinclusive of neurologic, ophthalmologic, auditory and developmental assessments.

REFERENCES

1. Ito M. A singular case of naevus depigmentosus systematicus bilateralis. Jpn J Dermatol 1951; 61: 31 - 2.
2. Pascual-Castroviejo I, López-Rodriguez L, de la Cruz Medina M, Salamanca-Maesso C, Roche Herrero C. Hypomelanosis of Ito. Neurological complications in 34 cases. Can J NeurolSci 1988; 15:124 – 9.
3. Kentab AY, Hassan HH, Hamad MHA, Alhumidi A. The neurologic aspects of hypomelanosis of Ito: Case report and review of the literature. Sudan J Paediatr 2014; 14(2):61 -70.
4. Sybert VP, Pagon RA, Donlan M, Bradley CM. Pigmentary abnormalities and mosaicism for chromosomal aberration: association with clinical features similar to Hypomelanosis of Ito. Journal of Pediatrics 1990; 116 (4), 581 - 6.
5. Ruiz-Maldonado R, Toussaint S, Tamayo L, Laterza A, del Castillo V.Hypomelanosis of Ito: diagnostic criteria and report of 41 cases. Pediatr Dermatol 1992; 9 (1): 1 - 10.
6. Okanari K, Miyahara H, Itoh M, Takahashi A, Aizaki K, Nakagawa E et al. Hemimegaloencephalopathy in a patient with coexisting Trisomy 21 and Hypomelanosis of Ito. J child Neurol 2012; 29 (3): 415 - 20.
7. Shobha N, Taly AB, Sinha S, Arunodaya GR. Hypomelanosis of Ito. J Neurol Neurosurg Psychiatry 2006; 77: 873.
8. Assogba K, Ferlazzo E, Striano P, Calarese T, Villeneuve N, Ivanov I et al. Heterogeneous seizure manifestations in Hypomelanosis of Ito: report of four new cases and review of the literature. Neurol Sci 2010; 31:9 – 16.
9. Placantonakis DG, Ney G, Edgar M, Souweidane M, Hosain S, Schwartz TH. Neurosurgical management of medically intractable epilepsy associated with Hypomelanosis of Ito. Epilepsia 2005; 46:329 – 31.
10. Ogino T, Hata H, Minakuchi E, Iyoda K, Narahara K, Ohtahara S. Neurophysiologic dysfunction in hypomelanosis of Ito: EEG and evoked potential studies. Brain Dev 1994; 16:407 – 12.