Case Report

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# Midas syndrome (microphthalmia syndrome and linear defects of the skin): case report

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## Introduction

MIDAS syndrome (microphthalmia, dermal aplasia and sclerocornea), also known as microphthalmia and linear skin defects syndrome, is considered a rare disease, characterized by ocular defects (microphthalmia, orbital cysts, corneal opacities) and linear dysplasia of the skin of the neck, head and chin. 56 diagnosed cases have been described in the world. It is transmitted as an X-linked dominant trait with lethal effect on male offspring.[1] In 77% of cases; it is associated with a terminal deletion of the Xp22.3 region. It may also be due to iatrogenic mutations in the HCCS (mitochondrial type c holocytochrome synthetase) c-IV gene, located at Xp22.3, or to mutations in the COX7B gene (structural subunit of cytochrome-c-oxidase) c-IV, with locus Xq21.1, however, these patients present linear defects without macrophthalmia. Both genes are part of mitochondrial complex IV.[2] Clinically, it is characterized by unilateral or bilateral microphthalmia with regularly linear skin defects of the face and neck, which are present from birth and disappear with age, leaving minimal residual scarring. Other findings may include central nervous system involvement (e.g., structural abnormalities, infantile seizures), developmental delay, cardiac defects (hypertrophic cardiomyopathy, oncocytic cardiomyopathy, and arrhythmias), short stature, diaphragmatic hernia, nail dystrophy, and genitourinary malformations.[1]

## Objective

To report a clinical case of "rare diseases" in the Mexican population, diagnosed at the Mónica Pretelini Sáenz Maternal Perinatal Hospital in Toluca, State of Mexico (Figure 1).

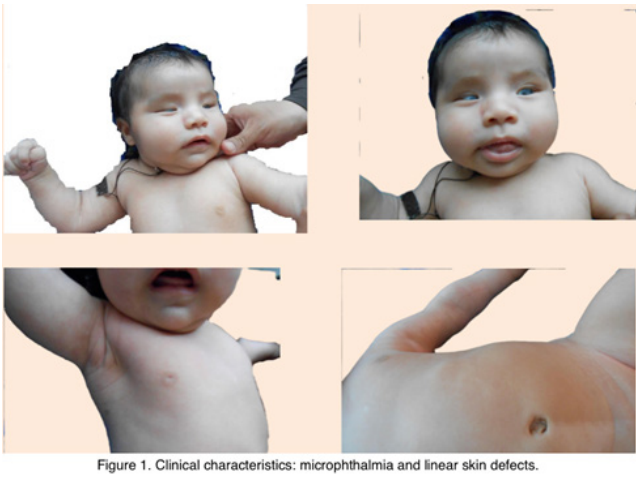

Figure 1. Clinical characteristics: microphthalmia and linear skin defects.

**Figure 1** Saenz Maternal Perinatal Hospital in Toluca, State of Mexico.

## Clinical case

Female newborn: Dad, 37 years, previously healthy. A 25-year-old mother diagnosed at birth with sclero-cornea and skin alterations, currently using a unilateral ocular prosthesis, normal intelligence, no consanguinity. Product of G1 prenatal control from the first trimester. It is obtained abdominally due to fetal hypo motility. He was considered eutrophic at term, without requiring specialized neonatal care. Lack of right eye opening, corneal opacity of both eyes, as well as the presence of "ecchymotic" spots on the right side of the face are observed (Figure 1).

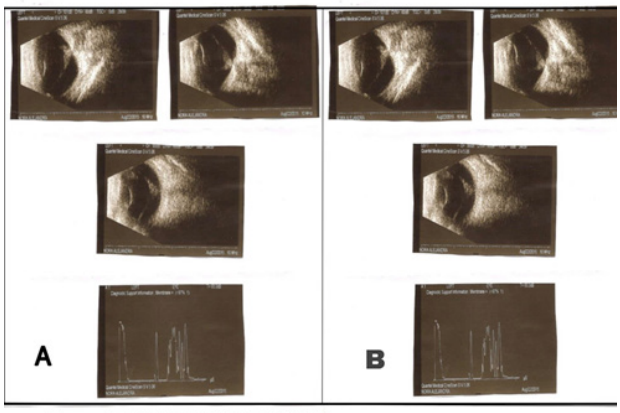


Figure 2. OCULAR ULTRASOUND.

A. RIGHT EYE(RE) IS PHAKIC, CLEAR VITREUS, RETINA, CHOROID AND EXCAVATION OF NORMAL OPTIC NERVE. B. LEFT EYE(LE) IS PHAKIC, CLEAR VITREOUS, MEMBRANE EMERGING OF OPTIC NERVE IS NOT MOBILE AND THICKENED

**Figure 2**

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Hematic biometry without alterations, not data of obstetric trauma. At one month of age, renal ultrasound and transfontanelar ultrasound were performed normal. A multidisciplinary study protocol was initiated without structural neurological or cardiac alterations. He went to a center specialized in Ophthalmology with a diagnosis of sclerocornea versus bilateral leukoma (Figure 2).

Clinical genetic study reports phenotype: right microphthalmia, left predominantly bilateral sclerocornea, vertical nystagmus, short nose with longitudinal Blaschko-type lesions on the nose and trunk, short neck, hypo pigmented spots on the face with a diagnosis of MIDAS syndrome.[3] Chromosomal complement 46, XX in 25 metaphases resolution 425 bands. At 4 months of age, mild psychomotor retardation was reported in follow-up by early stimulation. Brainstem Auditory Evoked Potentials showed data consistent with bilateral normal hearing for high tones (Figure 3).

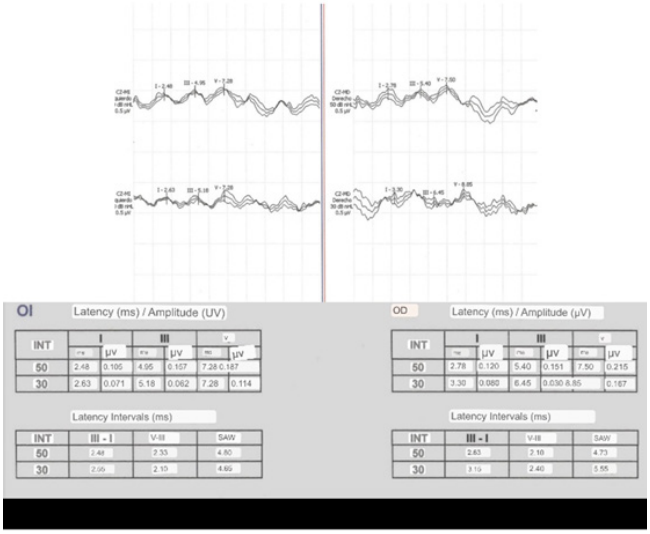


**Figure 3. Brainstem Auditory Evoked Potentials. Bilateral normal hearing for high tones.**

**Figure 3**

The mother presents data compatible with the same entity; a genealogy is performed, not finding another affected relative (Figure 4).

## Discussion

Comparing this clinical case with the literature, our patient presented the 2 major criteria of this disease, which is why the diagnosis was suspected, of the minor criteria only psychomotor retardation was reported and structural neurological, cardio logical and genitourinary alterations were ruled out. Since the mother is affected, it is considered a familial case. Its molecular genetic study is proposed.

## Conclusion

The clinical data are compatible with the MIDAS syndrome, with dominant inheritance linked to the X chromosome. Since no chromosomal alteration was found, an intragenic mutation in the HCCS gene is suspected, which encodes for the mitochondrial type c holocytochrome synthetase enzyme, which belongs to complex IV of the mitochondrial chain.

## Acknowledgments

None.

## Conflicts of interest

The author declares no conflicts of interest.

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