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Miller Syndrome: A Case Report

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Abstract:- Miller syndrome, also known as post-axial acrofacial dysostosis, is a rare congenital genetic disorder, characterized by mandibular and malar hypoplasia, and symmetrical post-axial deficits members. The most frequent ophthalmologic involvement is an ectropion of the lower eyelids.

I. INTRODUCTION

Miller syndrome, also known as post-axial acrofacial dysostosis, is a rare congenital genetic disorder, characterized by mandibular and malar hypoplasia, and symmetrical post-axial deficits members. The most frequent ophthalmologic involvement is an ectropion of the lower eyelids.

II. CASE REPORT

We report a case of an 8 months-old girl, followed-up in the pediatric department for a global developmental delay, with multiple craniofacial abnormalities and congenital heart defect. She was referred to our department for lagophtalmy.

A full ophthalmological examination was carried out under general anesthesia. We noticed complexe orbito palpebral abnormalities regrouping: hypertelorism, bilateral and symetric lower eyelid ectropion and downslanted palpebral fissures. (Figure 1)

Figure 1:- image-showing ectropion of the lower eyelids, hypertelorism and convergent strabismus

Besides, the girl had convergent strabismus with normal anterior segment and fundus findings and a normotone globe.

The girl hase also a microcrania with a head circumference of 36 cm (-2DS), malar hypoplasia, micrognathia, a broad bulbous nose with a broad nasal tip, a thin upper lip and a high arched palate along with a clinodactyly of the 5th finger.

Cytogenetic analysis revealed a chromosomal formula at 46, XX with no chromosomal abnormalities on all mitoses observed

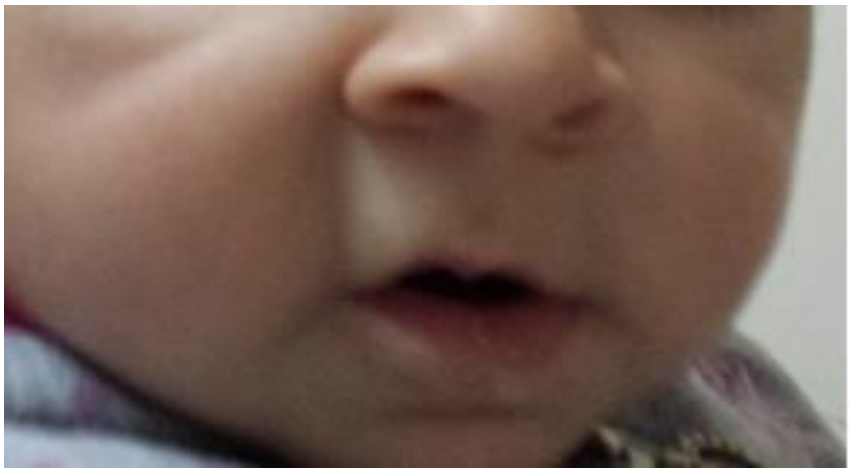

Figure 2:- Picture showing a broad bulbous nose, with a broad nasal tip, a thin upper lip and a high arched palate.

III. DISCUSSION

Miller syndrome is a rare disorder with an estimated prevalence of 1 case per 1 million newborns. The desease was first described in 1969 by Genée who speculated that it is a part form Treacher-Collins syndrome [1]. Wiedemann in 1975 described it as a separate entity. [2], other cases were reported by Miller et al in 1979. [3] [4], the syndrome was named Genée-Wiedemann syndrome in 1987. [5]

Miller syndrome is caused by a mutation in the DHODH (dihydro-orotate dehydrogenase) gene located on chromosome 16q22 [6] [7] with an autosomal recessive pattern of inheritance. To date, less than 75 cases have been described in the literature [8], both sexes are affected in equal numbers. Most abnormalities are noticeable at birth. The mean ophthalmologic symptoms are: ectropion of the lower eyelid, colobomas of the lower eyelids, downward sloping eyelids, total or partial absence of the eyelashes of the lower eyelids, and corneal opacities.

There is phenotypic variability in Miller syndrome, patients may present different subgroups in postaxial acrofacial dysostosis.

The other signs of the disease includes: the absence or anomaly of the fifth fingers of the hands and feet, radiocubital synostosis, developmental abnormalities of the cubital and / or fibula, casesyndactyly of the fingers, vesicoureteric urinary reflux in the child, abnormal positioning of the intestines and pyloric There are no mental disorders.

Specialized X-ray imaging will confirm the presence certain craniofacial or limb abnormalities observed.

Molecular genetic testing can confirm the diagnosis of Miller syndrome and can detect a mutation in the DHODH gene, but it is only available in specialized laboratories.

The main disorders that should be distinguished from Miller syndrome are Treacher Collins syndrome (no limb abnormalities) and Nager syndrome (paraxial cranial dysostosis). [10]

Ophtalmologic managment depends on the clinical presentation, form a simple follow up to complicated surgery like corneal transplant.

The treatment of Miller syndrome may require the coordinated efforts of a team of specialists. Pediatricians, maxillofacial surgeons; dental, plastic surgeons, ophthalmologist, ENT specialists, psychologists and other health professionals should systematically plan a personalized treatment.

IV. CONCLUSION

The diagnosis of Miller syndrome is based on a careful clinical evaluation, and identification of characteristic physical findings. Ophthalmological manifestations should be systematically searched because, in the absence of appropriate treatment, it can be blinding.

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