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A RARE CHROMOSOMAL DISORDER: CASE REPORT OF MONOSOMY 18P WITH LITERATURE REVIEW

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ABSTRACT

Monosomy 18p is a rare genetic disorder caused by partial or complete deletion of the short arm of chromosome 18. Clinically, it is characterized by cognitive impairment associated with a dysmorphic syndrome. It can be associated with cerebral malformations, which determine the prognosis. The definitive diagnosis is based on cytogenetic analysis which identifies a partial or total absence of the short arm of chromosome 18p. There is currently no specific treatment. Management care is multidisciplinary and is mainly based on speech therapy sessions and early educational programs, in order to help improve the performance of affected children.

KEYWORDS: Monosomy 18p, deletion 18p syndrome, partial monosomy 18p.

INTRODUCTION

Monosomy 18p refers to a chromosomal abnormality resulting from the absence of all or part of the short arm of chromosome 18. First reported in 1963 by the french geneticist Jean de Grouchy, it was considered the first example of a partial monosomy compatible with life. Clinical features of monosomy 18 P typically include mild to moderate intellectual disability, short stature, a round face with a short and proeminent philtrum, eyelid ptosis, and large ears with detached pines. Cytogenetic analysis is required to establish a definitive diagnosis. There is no specific treatment, management is multidisciplinary and mainly relies on speech therapy sessions and early educational programs, which aim to improve the developmental outcomes in affected children.

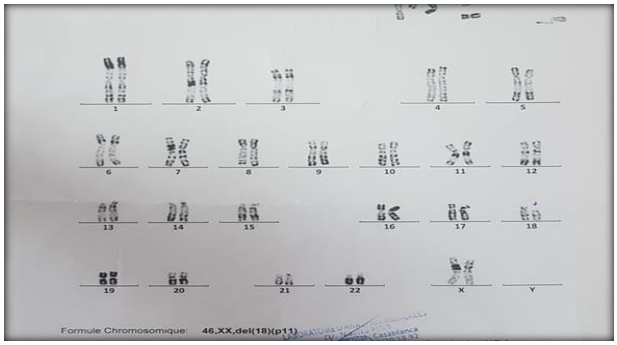
We report the case of an 11 year old girl admitted to the Pediatrics Department of Military Hospital Mohamed V of Rabat for intellectual deficiency and language disorder.

CASE REPORT

The patient, M.H., is an 11 year old female, the youngest of two siblings, with a healthy older brother and non-consanguineous parents. She presented with a psychomotor delay with delayed language acquisition. The clinical examination revealed a child with short stature below 2 standard deviations, with short fingers and hypertelorism. There was no nipple spacing anomaly. She had difficulty walking and presented global cognitive development disorder.

Initial investigations included a brain CT scan which was unremarkable. The brain MRI did not show any abnormalities, except a small simple cyst in the frontal horn of the left lateral ventricle.

A genetic analysis was requested, revealing a partial monosomy of chromosome 18 in all the mitoses observed.



As part of the malformation assessment, a transthoracic ultrasound and an abdominal ultrasound were performed to screen for cardiac and abdominal anomalies, respectively, but no abnormalities were found.

As part of the screening, a genetic analysis was requested for the parents.

Therapeutically, the patient benefited from multidisciplinary care with speech therapy sessions.

DISCUSSION

Although monosomy 18p is a rare disorder, its incidence is estimated at approximmately 1 in 50 000 live birth, with a female to male ratio of 3/2.

In the most common form of monosomy 18P, the dysmorphic syndrome is usually mild and nonspecific, and the intellectual disability ranges from mild to moderate.

A small subgroup of patients, approximately 10 to 15% of cases, presents with severe brain and/or facial malformations, suggestive of holoprosencephalic spectrum disorders.

In the majority of cases (about two thirds of cases), the syndrome is due to a simple de novo terminal deletion. In the remaining one third, several mechanisms may be involved, including : de novo translocation with loss of 18p, a partial separation of a parental translocation or inversion, or a chr18 ring. Parental transmission of 18p syndrome has also been reported.

Cytogenetic analysis is necessary to make an accurate diagnosis.

The risk of recurrence for siblings is low in de novo deletions and translocations, but is significant in cases of parental rearrangement.

Monosomy 18p can be detected prenatally, through amniocentesis or chorionic villus sampling and cytogenetic testing.

The differential diagnosis may include a large number of syndromes associated with short stature and mild intellectual disability. In young children, 18p deletion syndrome can vaguely suggest Turner syndrome or Trisomy 21. In all cases, cytogenetic analysis allows the correct diagnosis to be made.

There is no specific treatment. However, speech therapy and early educational programs may help improve children's performance. Physical therapy is also recommended for patients with hypotonia.

Except for patients with severe brain malformations, life expectancy does not appear to be significantly reduced.

CONCLUSION

Monosomy 18p is a rare entity. The challenge is to recognize this diagnosis, make it accurately, and distinguish it from other syndromes associated with short stature and mental retardation. The prognosis is generally good, except the forms associated with severe cerebral malformations.

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