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# Clinical presentation of monosomy 1P36 in the Mexican population

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## Introduction

Monosomy 1p36 syndrome is part of the group of diseases known as "low prevalence diseases" or "rare diseases" (RD). RDs have been defined within the framework of the European Community as those with a life-threatening or chronic debilitating character, with a prevalence of less than 5 cases per 10,000 inhabitants.[1]

Microdeletions consist of the loss of chromosomal material, in most cases between 1 and 3 million base pairs of DNA, which cannot be detected by conventional chromosomal analysis, so specific techniques such as FISH (Fluorescent In Situ Hybridization) are required.[2] Monosomy 1p36 is the most frequent terminal microdeletion syndrome and therefore best characterized.[2]

### History

The first case was described in 1981. It was a 4-year-old girl with severe mental retardation and congenital anomalies (wide fontanelles, generalized hypotonia and grade III/IV systolic murmur). Banding analysis showed that her karyotype was balanced (45 XX) (1;21 ) (p36;p13), and it was suggested that a deletion Submicroscopic imaging of the short arm of chromosome 1 could account for the patient's clinical features as a result of missegregation of a parental balanced translocation.[1]

However, it was not until 1997 that this new clinical entity caught the attention of clinical geneticists.[3] We now know that it includes within its clinical spectrum multiple congenital anomalies and intellectual disability, and that it occurs in approximately 1 in 5,000 to 10,000 live births and is the microdeletion subtelomeric most commonly observed in humans.[3–6]

### Epidemiology

It can be observed in up to 1.2% of cases of mental retardation of unknown or idiopathic cause, with an approximate report of 100 cases in the international literature and 2 cases reported until 2011 in Mexico.[7] With a recent work, carried out at the National Institute of Pediatrics (pediatric reference center at the national level), there is a report of 8 patients without sex predilection. The cases described in Mexico are reported by Dr. Villarroel,[5,8,9] which will be included in this work.

### Genetics

Four types of rearrangements have been described in monosomy 1p36:

a) Unbalanced translocations

b) Interstitial deletions

c) Simple terminal deletions

d) Complex rearrangements.

Unbalanced translocations result in chromosome 1 with a deleted segment of 1p36 (partial monosomy) and a region of another chromosome attached to the distal end of 1p (partial trisomy).

Deletions results from rearrangements after two telomere - proximal breaks occur, resulting in retention of the 1p36 telomere and removal of material proximal to this region. Complex rearrangements include deletions with duplications, triplications, inversions, and/or insertions.[10] More than 60% of patients have interstitial deletions de novo in the subtelomeric region inherited from maternal chromosome 1.[11]

Approximately 40% of deletion or breakpoints occur between 3.0 – 5.0 Mb of the 1p telomere and are clustered between 4.0 – 4.5 Mb. However, deletion size and breakpoints are varied and are not specifically correlated with the severity of clinical expression.[11]

Several genes are involved in the phenotypic variation of the syndrome, among which the most relevant are:

1. **MMP23B** which provides the critical region for a wide, late-closing anterior fontanelle[12]
2. **GABRD** has been implicated in the neurodevelopmental abnormalities, neuropsychiatric problems and seizures that occur in this syndrome, associated with GABA haploinsufficiency[12]
3. Proto -oncogene haploinsufficiency **SKI** is thought to be involved in intellectual disability, seizures and heart defects[12]
4. PRDM16 haploinsufficiency **has been associated with specific defects in left** ventricular growth and alterations in its morphology. The alteration in potassium channels and their voltage associated with haploinsufficiency of the gene **KCNAB2** has been linked to developmental delay, intellectual disability, and seizures.[12]
5. One of the genes whose clinical involvement in humans has not been proven is the **RERE gene**, which in mice has suggested alterations in the phenotype, including short stature, brain abnormalities, with its involvement in neurodevelopmental delay and cardiac disorders.[12]

6. **PDPN gene and the UBE4B** gene which has been associated with alterations in Purkinje fibers.[12]
7. The **SPEN gene** It is related to alterations in the formation of the septum and the cardiac muscle and at the same time it has been associated with pancreatic alterations, with alterations in the differentiation of β cells and with hypoplasia of hepatocytes.[12]

## Phenotype

Phenotypes are described, both share dysmorphisms, but are differentiated by growth characteristics, some with growth retardation and others with macrosomia. We can see some examples of them in Figure 1.[15] Most patients have craniofacial characteristics such as: straight eyebrows 84%, sunken eyes 93%, wide nasal bridge 97%, dysplastic earlobes 88% and pointed chin 89%; the combination of these characteristics make the characteristic facial pattern of the 1p36 deletion syndrome.[16]

**Figure 1** Taken from: Kurosawa K, Kawame H, Okamoto N, et al. Epilepsy and neurological findings in 11 individuals with 1p36 deletion syndrome.[15]

Postnatal microcephaly has been reported in 38%, brachycephaly in 43%, elongated anterior fontanelle in 100%, low hairline in 38%, asymmetrical and low-set auricles in 58%, oblique palpebral fissures upward in 41%, hypertelorism in 31%, prognathism in 44%, short fifth finger or clinodactyly in 64%, and estrogen hypoplasia in 14%.[17] The most frequent symptoms in patients with this syndrome are detailed below; however, other reported symptoms are hearing loss in 39%, strabismus in 33%, obesity in 11%, and hypothyroidism in 20%.[15]

## Neurologic symptoms

The clinical picture includes intellectual disability (98%) and is frequently associated with epilepsy (70%) without family history.[15,16] The spectrum of cognitive impairment ranges from severe (87%) to moderate (13%).[16] The onset of seizures is in the first 6 months (79%), with a mean age of 2.75 months, with the most common being generalized seizures, with tonic, tonic-clonic, clonic- myoclonic seizures also being found, with findings in interictal periods in the electroencephalogram of alterations in the rolandic zone, posterior temporo -occipital, multifocal or generalized waves, polyspikes and spike-wave discharges. Axial hypotonia 92%, weak sucking 70%, infantile spasms (16%), hypotonia (85% in the first years of life) are other neurological manifestations described.[14]

The most common structural finding in patients with epilepsy is cortical atrophy with elongation of the ventricles, white matter abnormalities with predominance of myelination deficiency and non-specific alterations.[16,5] In this regard, it is described that in brain tomography and magnetic resonance imaging it is common to find brain abnormalities at birth, such as ventricular dilatation, hydrocephalus, brain atrophy, leukoencephalopathy and abnormalities or agenesis of the corpus callosum.

## Cardiac symptoms

Structural congenital defects are found in 69% and functional in 22%, with patent ductus arteriosus being the most frequent 37% and ventricular septal defects 37%.[14] Ebstein 's anomaly (attachment of the posterior and septal tricuspid valve leaflets to the right ventricular endocardium) being the most serious cardiac manifestation associated with the clinical spectrum, with a probable association with the SKI gene located in a distal locus and other loci corresponding to the RERE and UBE48 genes as probable contributing factors to the etiology of this entity.[17]

## Genitourinary

The literature reports that 22% of patients have alterations at this level, among the most frequently found are unilateral renal pelvis with hydronephrosis of the upper pole, renal ectopia with cyst in the right kidney and pelvic ectasia. In a minority of men, cryptorchidism, hypospadias, scrotal hypoplasia and micropenis have been described. In women, small labia minora and clitoris, hypertrophic labia majora and uterine hypoplasia have been described.[1]

Skeletal or delayed bone growth, brachydactyly (short hands and feet), scoliosis, rib abnormalities, and asymmetric lower limbs have been described in 40% of patients. Congenital spinal stenosis and clinodactyly have also been observed in some patients, and polydactyly has been described in a few cases.[1]

## Cytogenetic diagnosis

Diagnosis requires molecular cytogenetic analysis. Conventional cytogenetic techniques cannot detect these different rearrangements. Most visible deletions involve the telomeric bands of chromosomes. Rearrangements of these regions are difficult to identify by routine banding techniques.[1]

Fluorescence in situ hybridization technique (FISH) and the comparative genomic hybridization technique (array -CGH)[1] or its variants.

The FISH technique uses probes specific to centromeres or specific regions of chromosomes marked with a fluorochrome. After hybridization with nuclei in interphase or metaphase, it is possible to observe the presence of gains, losses or fusions between genes.[21]

The possibility of performing prenatal diagnosis of this entity has been described, both by noninvasive and invasive methods. Ultrasonography is used as a noninvasive method, in which intrauterine growth retardation, congenital abnormalities already described and mainly brain abnormalities can be observed, among which the most common are ventriculomegaly and hydrocephalus; an increase in alpha-fetoprotein in maternal serum has been reported. From invasive tests we found an increase in alpha-fetoprotein in amniotic fluid obtained by amniocentesis and cytogenetic study in amniotic fluid.

## Differential diagnosis

Dysmorphological characteristics with various syndromes, among which Rett Syndrome stands out, with which they share particular hand movements (squeezing, tremors, involuntary clapping, bringing the hands to the mouth and light hitting among others) alterations in intellectual development, motor development, language, autistic traits, hypotonia and seizures, the differential characteristic being that patients with Rett Syndrome have normal development the first 6 - 18 months of age, with subsequent loss of motor ability.[1]

Angelman syndrome they share intellectual disability, poor communicative intent, severe language impairment, alterations in motor development, in feeding and presence of seizures. The difference is made cytogenetically, as Angelman syndrome is found deletion of the maternal chromosome in the 15q12 region.[1,20]

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Finally, monosomy 1p36 also shares characteristics with Prader-Willi syndrome, such as overeating, obesity, hypotonia, cognitive impairment, psychomotor retardation, so those patients in whom Prader-Willi syndrome was suspected and had a negative cytogenetic result should be approached for probable monosomy 1p36.[19] In addition, both manifest other less visible data; difficulties feeding in early childhood, growth retardation, visual (strabismus) and genitourinary disorders, with the differential diagnosis being confirmed by means of molecular cytogenetics with a deletion in the 15q11-q13 region.[1]

### Treatment

It must be multidisciplinary; the management of neurodevelopmental difficulties involves physiotherapy for motor, muscular and skeletal disorders, with surgical treatments becoming necessary in the case of severe skeletal disorders. Language therapy will work on the deficiency in verbal communication, aspects related to swallowing-ingesting and facial hypotonia.[1]

Neuropsychological management plays an important role in supporting cognitive difficulties in attention, memory, perception, executive functions and learning, with emphasis on the control of seizures, for which there is still no antiepileptic drug of choice, with the use of monotherapy and combination therapy, where the most commonly used drugs are carbamazepine, valproate, phenobarbital, topiramate, with carbamazepine being the drug that has given the best control in seizures. However, it has been found that the development of infantile spasms is common, even with the use of carbamazepine, so the use of steroids has been added to the therapeutic scheme, which have helped to have adequate control of the seizures; but the development of Lennox Syndrome at an older age has been described Gastaut.

### Justification

Since 1p36 deletion is a genetic syndrome with a low prevalence and a diverse clinical spectrum, it is necessary to analyze the most common clinical presentation in the Mexican population, as well as analyze the diagnostic route of reference centers, such as the National Institute of Pediatrics, where this condition has been diagnosed, in order to suspect this clinical entity early, diagnose it and initiate the relevant interventions as soon as possible.

## Materials and methods

An observational, retrospective, descriptive study was carried out on patients at the National Institute of Pediatrics with dysmorphia and malformations with a suspected diagnosis of 1p36 deletion syndrome. A convenience sampling was carried out with the clinical file records between January 1, 2008, and January 1, 2018, with a diagnosis of 1p36 deletion. From which a database was created in Excel 2019 that included the variables (clinical spectrum described in the international literature on 1p36 deletion), it was exported to the SPSS statistical program version 21 where the data analysis was carried out.

A description of the clinical characteristics of the patients was made using frequencies and percentages for the categorical variables.

Patients were stratified according to the positivity or negativity of FISH for the detection of the 1p36 deletion, taking as cases the patients who had the confirmed diagnosis and as non-cases the patients who did not have it. The differences between the variables studied were demonstrated by comparing the two groups using the chi- square test or Fisher's exact test. A p value of 0.05 was considered statistically significant $\leq$ .

The chi- square test loses validity due to the number of boxes that have expected a count less than five, so in cases where it was less than five, Fisher's exact test was used.

## Results

As shown in Table 1, of the 31 patients in whom the syndrome was clinically suspected, eight of them had positive FISH, while the remaining 23 were negative, so the results are described according to this classification.

**Table 1** Fish positivity for Monosomy 1p36

| | Monosomy 1p36 n= 8 (%) | No monosomy 1p36 n=23 (%) | Chi or fisher | p-value |
|---|---|---|---|---|
| **Sex** | | | | |
| Male<br>Female | 4 (50)<br>4 (50) | 11 (47.8)<br>12 (52.2) | 0.6 | 0.6 |
| Middle Ages +- DE | 4.56 (+-4.49) | 4.13 (+-2.9) | 0.2 | 0.2 |
| **Clinical features** | | | | |
| Facials | | | | |
| Straight eyebrows | 7 (87.5) | 10 (43.5) | 4.644 | 0.031* |
| Sunken eyes | 6 (75) | 8 (34.8) | 3.876 | 0.049* |
| Wide nose bridge | 6 (75) | 12 (52.2) | 1,270 | 0.260 |
| Sunken Ears | 2 (25) | 2 (8.7) | 1.404 | 0.268 |
| Pointed chin | 5 ( 62.5) | 3 (13) | 7.582 | 0.013* |
| Postnatal microcephaly | 6 (75) | 9 (39.1) | 3.058 | 0.08 |
| Brachycephaly | 3 (37.5) | 4 (17.4) | 1.373 | 0.335 |
| Elongated anterior fontanelle | 2 (25) | 3 (13) | 0.627 | 0.583 |
| Low hair implantation | 2 (25) | 4 (76) | 0.220 | 0.634 |
| Asymmetrical earlobes | 3 (37.5) | 5 (21.7) | 0.770 | 0.393 |
| Upward palpebral fissures | 3 (37.5) | 5 (21.7) | 0.770 | 0.393 |
| Hypertelorism | 1 (12.5) | 1 (4.3) | 0.654 | 0.456 |
| Prognathism | 3 (37.5) | 2 (8.7) | 3.640 | 0.093 |
| Neurological | | | | |
| Mental retardation | 8 (100) | 22 (95.7) | 0.359 | 0.549 |
| Epilepsy | 3 (37.5) | 3 (13) | 2.274 | 0.161 |
| Cortical atrophy | 3 (37.5) | 6 (26.1) | 0.375 | 0.660 |
| Axial hypotonia | 6 (75) | 8 (34.8) | 3.876 | 0.049* |
| Suction weak | 0 | 0 | 0 | 0 |
| Infantile spasms | 0 | 0 | 0 | 0 |
| Cardiac | | | | |
| PCA | 2 (25) | 2 (8.7) | 1.404 | 0.268 |
| Septal defects | 1 (12.5) | 1 (4.3) | 0.654 | 0.456 |
| Ebstein 's anomaly | 1 (12.5) | 1 (4.3) | 0.654 | 0.456 |
| Genitourinary | | | | |
| Unilateral renal pelvis | 1 (12.5) | 0 | 2,971 | 0.258 |
| Hydronephrosis | 1 (12.5) | 0 | 2,971 | 0.258 |
| Renal ectopia | 1 (12.5) | 9 | 2,971 | 0.258 |

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Table 1 Continued..

| | | | | |
|---|---|---|---|---|
| CryptoQuidia | 1 (12.5) | 1 (8.3) | 0.762 | 0.450 |
| Hypospadias | 0 | 0 | 0 | 0 |
| Small labia minora | 1 (25) | 2 (18.2) | 0.085 | 1 |
| Small clitoris | 0 | 0 | 0 | 0 |
| Hypertrophic labia majora | 0 | 0 | 0 | 0 |
| Uterine hypoplasia | 0 | 1 (9.1) | 0.390 | 1.09 |
| Orthopedic | | | | |
| Growth retardation | 5 (62.5) | 15 (65.2) | 0.019 | 0.890 |
| Brachydactyly | 3 (37.5) | 3 (13) | 2.274 | 0.161 |
| Scoliosis | 1 (12.5) | 1 (4.3) | 0.654 | 0.456 |
| Asymmetry of limbs | 0 | 0 | 0 | 0 |
| Clinodactyly | 3 (37.5) | 3 (12) | 2.274 | 0.161 |

We can highlight that some facial characteristics are more frequent in patients in whom the syndrome was confirmed by FISH, such as straight eyebrows, which showed significant differences in cases and non-cases with an $X^2$ 4.644 and a p = 0.031 when this characteristic was present in 87% of cases compared to non-cases in which it was only present in 43%.

Another feature in which the analysis showed significant differences was sunken eyes, which is present in 75% of the cases that presented an $X^2$ of 3,876 with a p value = 0.049, as well as a pointed chin in 62% of the cases and in 13% of the non-cases, obtaining an $X^2$ 7.582 and p = 0.013.

Regarding neurological manifestations, axial hypotonia was more frequent in patients with FISH-confirmed 1p36 deletion syndrome, occurring in 75% of cases and 43% of non-cases, with a p value = 0.049; however, no statistically significant differences were found regarding cardiac, genitourinary or orthopedic characteristics of both groups.

By following up the cases, we found that only four of them required any surgical treatment, the most common being surgical closure of the ductus arteriosus, required in two of our cases, while right orchidopexy was only performed in one case, with surgical correction of the epiblepharon being equally frequent.

## Discussion

monosomy 1p36 syndrome is a "rare disease", it is the most frequent and best characterized terminal microdeletion syndrome, but it is rarely diagnosed in our setting; therefore, it is essential to have literature that describes and brings together the cases treated in concentration centers, such as the INP, since the experience of having 8 of these cases confirmed with FISH is a valuable opportunity that allows us to contribute to the international knowledge about this entity and its dissemination in our region.

This syndrome has been described to be more frequent in the female sex;[5] however, in the cases reported in the present work, we found the same frequency of the syndrome in men and women, 50% in each of them. In this study, the characteristic facial pattern of this syndrome was confirmed in features such as straight eyebrows (87% frequency in our series and 84%[16] reported in the literature), sunken eyes (75% reported in our series and 93%[16] reported in the literature) and pointed chin (62.5% reported in our series and 89%[16] in the literature).

The broad nasal bridge in this study was not shown to be more frequent in patients with monosomy 1p36 syndrome; however, it is important to note that out of the total of 8 patients, 6 had this characteristic and therefore, if the number of patients studied were increased we could confirm a statistically significant difference in this characteristic between groups, since this trait is reported in the literature in up to 97%[16] of patients. On the other hand, although sunken ears are reported in up to 88% [16] of cases in the literature, in this study this characteristic was presented in only 2 patients in each group.

None of the neurological characteristics of the patients in this study showed a statistically significant difference between the groups studied. This contrasts with what is reported in the literature where up to 70% of the patients have epilepsy; however, something that is consistent both in the literature and in our study is the intellectual disability, present in all the positive cases. It is striking that one of the negative patients with suspected 1p36 deletion syndrome did not present it, therefore, the clinical suspicion of the syndrome was probably erroneous.

Of the eight cases reported at our Institute, the record shows that two patients did not attend the mental health service for assessment of severity, while five cases were reported with moderate intellectual disability, one of these cases with an additional diagnosis of autism spectrum disorder and one case with severe presentation. Regarding dependency, the five moderate cases attended Multiple Care Centers, in addition to having support from the Institute's psychology.

None of the cardiac or genitourinary characteristics showed a statistically significant difference between the groups studied; in fact, the frequency of each of the characteristics studied in each of the groups was the same. In the literature, the most frequent characteristics of this syndrome are reported to be patent ductus arteriosus (37%) and ventricular septal defects 37%,[14] as well as unilateral renal pelvis with hydronephrosis of the upper pole and labia minora and small clitoris in some cases.[1]

Likewise, growth retardation was found with the same frequency in both groups; it is reported in the international literature as the most frequent somatic developmental delay, occurring in up to 62.5% of cases, so if a larger number of patients were included in this study, the reported frequency of this characteristic would probably increase.

Finally, it is interesting to note that in this work, 28 of the patients in whom the syndrome was clinically suspected had a negative FISH result, and therefore, these patients are subject to continuing a study protocol that rules out numerous differential diagnoses, to mention a few examples, those in which dysmorphological characteristics are shared such as Rett, Angelman and Prader Syndromes Willi.

The treatment of our patients was aimed at controlling epilepsy, with one patient being reported to have difficult management of seizures, while the other two cases with epilepsy achieved adequate seizure control with the use of two antiepileptic drugs.

The most commonly used surgical treatment in our cases was closure of the ductus arteriosus, which was performed in two cases, while only one patient required right orchidopexy and only one case required epiblepharon correction, all patients without postoperative complications.

The follow-up by the genetics service after the diagnosis was confirmed was directed towards the risk of recurrence in subsequent pregnancies, so a FISH test was requested for both parents, reporting one case in which the father had died and the mother reported satisfied parity; one case confirmed by FISH of paternal origin due to a balanced translocation,[1,7] so a study was performed on four paternal uncles, all of which were reported without alterations. Three cases presented negative FISH in both parents, so counseling was given on de novo cases and the low risk of recurrence in subsequent pregnancies; while the rest of the cases lost follow-up with the genetics service.

This work provides for the first time to the literature valuable information about the clinical characteristics of Mexican patients with

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this "rare disease" confirmed by FISH. The importance of knowing this syndrome lies in the fact that it can be observed in up to 1.2% of cases of mental retardation of unknown or idiopathic cause,[7] which is a frequent cause of consultation at the third level of care.

On the other hand, this study has the disadvantage of having a limited number of patients. However, it is important to consider that this is the total number of patients in whom this syndrome has been suspected or confirmed in 10 years of experience; and therefore, considering that this is a disease of which there are only 100 cases in the international literature, our cases are part of a valuable sample.[7] This collection of cases opens the door to new studies in the future, which, by increasing the knowledge we have about this syndrome and the accessibility to the FISH test to make the diagnosis, can increase the number of cases studied and thus elucidate characteristics of the phenotype that in this study did not prove to be significantly different from the control group.

## Conclusion

This study describes and compiles the clinical characteristics of 31 patients with clinical suspicion of 1p36 deletion syndrome in the last 10 years; of which, only 8 were confirmed by FISH. Based on this work, it is concluded that straight eyebrows, sunken eyes, pointed chin and axial hypotonia are the clinical characteristics most frequently found in Mexican patients with this disease, thus allowing a valuable contribution to the international literature about the clinical characteristics that may be useful when suspecting this entity.

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## Conflicts of interest

The authors declare no conflicts of interest.

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