# The Importance of Clinical Nutrition in Patients with Genetic Disorders Diagnosed with 5000 Genetic Studies

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## Abstract

Mutations or chromosome aberrations are considered alterations in the chromosome number or structure. They are mainly considered due to gametogenesis inborn error (meiosis) or during the zygote first cellular divisions. All these alterations might be observed during metaphase from the cellular cycle, where DNA loses are seen (clastogenic processes) due to DNA repair processes deficiency o total absence, among others. 5000 chromosome studies were performed at Hospital Para El Niño Poblano (Pediatric Hospital) in Mexico. From 1992 to 2011, were 34.6% (1596 patients) showed different chromosome alterations. Among the studies population, male and female pediatric patients with different genetic diseases were chosen. These chromosome changes are classified as numeric or structural alterations, respectively. Another group of genetic alterations in this study were based on inborn metabolic diseases known as metabolic diseases which can be inherited among generations in a recessive inheritance trait mainly. A wide variety of pediatric patients with genetic diseases due to metabolic or chromosome alterations are described in this study analyzing their clinical characteristics, medical, surgical and nutrimental treatments and their clinical evolution according to the genetic disease.

**Keywords:** chromosome mutation, numerical and structural changes, karyotype metabolism, inborn errors of metabolism and DNA

## INTRODUCTION

**1. Chromosome Alterations.** Mutations and chromosome alterations are considered to be alterations in the number and structure and secondary to errors of the birth (meiosis) or during the first cell division of the Zygote. All these alterations must be observed during the metaphase of cell cycle where the losses are observed (clastogenic process) due to the deficiency

of the processes of repair of DNA or total absence. In order to have an incidence of chromosome alterations in the State of puebla 5000 chromosome studies in the Hospital for the Poblano child, carried out (1992 -2011) **Figure 1**.

The 34.6% (1596 patients) of the studied patients, showed different chromosome alterations. Another group of genetic alterations are known as mutations and are inherited in different generations. A wide range of pediatric patients with genetic diseases by chromosome alterations and are described in this chapter.

Clinical features as dental, nutritional, medical or surgical procedures are analyzed and their evolution according to the genetic condition evaluating its relationship in the health area.

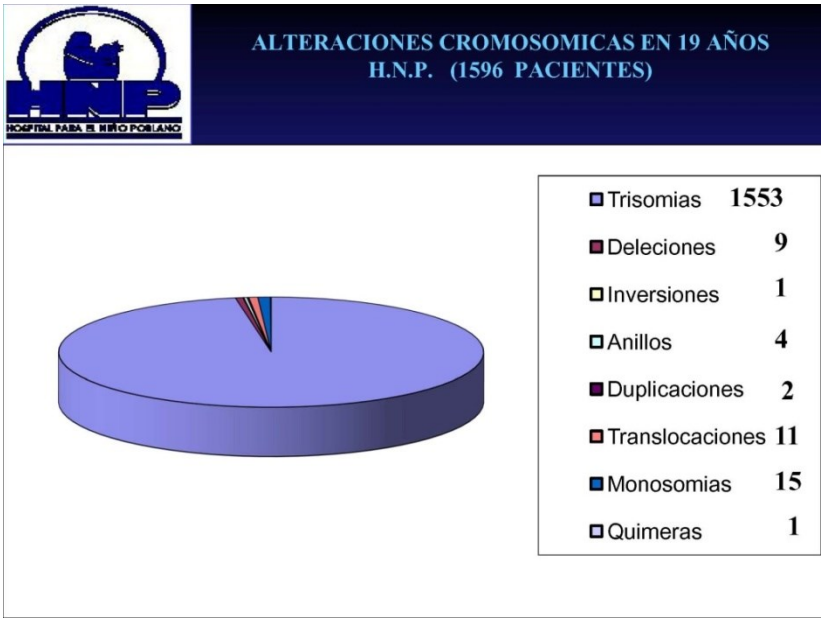



**Figure 1.** Chromosome alterations, shows 1596 (34.6%) patients with different aberrations. Of these, 1553 (33.6%) patients with Trisomy.

The word chromosome comes from the Greek (chroma, color) and (soma, body) for their property to be stained with specific staining particles. A chromosome is a structure composed of deoxyribonucleic acid (DNA) and protein within the cells. It is a simple piece of DNA with genes, regulatory elements and other nucleotide sequences. Chromosomes are also found attached to proteins, which serve as packages of DNA and controls its function Thanbichler et al., 2005; Sandman et al., 1998; Sandman K, Reeve, 2000; Pereira et al., 1997. The chromosomes are

different according to each variety of organisms. The DNA molecule may be circular or linear, and consist of 100,000 to 10,000,000,000 Paux et al., 2008. nucleotides in a long chain. Eucariotas, 1973 cells usually have linear chromosomes and cells procariotas Thanbichler M, Shapiro L, 2006; Nakabachi et al., 2006; Pradella et al., 2002, it has small groups of circular chromosomes. Also, cells contain more than one type of chromosomes; for example, mitochondria in most eukaryotes and chloroplasts in plants their chromosomes are small.

In eukaryotes, nuclear chromosomes are packaged by proteins into a condensed structure called chromatin. This allows the complete DNA molecule is found inside the nucleus. The structure of chromosomes and chromatin varies through the cell cycle. Chromosomes are considered essential in cell division unit must replicate, divide and pass in a satisfactory manner to the new cells to preserve their offspring.

The chromosomes exist as duplicates or not duplicate. Not duplicated chromosomes are single linear chains, different of duplicated chromosomes (copied during synthesis phase) contain two copies joined by a centromere. The compaction of the chromosomes during meiosis and mitosis. Chromosome recombination plays a vital role in the evolution and genetic diversity. If these structures initiated through processes known as chromosome instability and mutation, cells may die or avoid the onset of apoptosis, initiating a risk for the onset of cancer.

Human chromosomes are divided into two types; autosomes and sex chromosomes. Some genetic characteristics linked to the X chromosome and are passed through this chromosome. The autosomes contain hereditary information. Gametes are produced by meiosis of a cell line. During meiosis, homologous chromosomes of the parents can exchange chromosome material between them (crossover), and resulting in new chromosomes.

**DISCUSSION:** Chromosome aberrations are disruptions in the normal chromosome structures and is considered a major cause of genetic human conditions, known as genetic disease that can have or not a pattern of inheritance, such as Down syndrome, considered as the most frequent chromosome Trisomy observed in this study with a number of 1511 patients, **Table 1 & Figure 2.**

| Chromosome Aberrations | (%) patients |
|---|---|
| 1. Trisomy | 1553 |
| 2. Deletions | 9 |
| 3. Inversions | 1 |
| 4. Rings | 4 |
| 5. Duplications | 2 |
| 6. Translocations | 11 |
| 7. Monosomíes | 15 |
| 8. Chimeras | 1 |
| Chromosome Aberrations | (34.6%) 1596 |
| Total of Trisomíes | (33.6%) 1553 |
| Trisomy 21 | (32.8%) 1511 |
| 1. T21<br>2. T21;14<br>2. T21;21<br>3. Mosaic | 1127<br>260<br>43<br>81 |
| Varias Trisomías | (0.90%) 42 |
| Different Chromosome Aberrations: | (0.93%) 43 |
| Total (karyotipes) | (100%) 5000 |
| Total normal karyotipes | (65.4%) 3021 |
| Total Chromosóme Aberrations | (34.6%) 1596 |

**Table 1**. Different chromosome alterations in 19 years in the Hospital del niño Poblano, Mexico.

Research on the human karyotype took many years to resolve the most basic question. They questioned how many chromosomes contain a normal diploid human cell. In 10, reported 47 chromosomes in spermatogonia and 48 in oogonia, concluding an XX/XO von Winiwarter, 1912; Painter, 1922 sex determination mechanism was not sure whether the diploid number of man was 46 or 48 and subsequently insisted on humans have a XX/XY Painter, 1923; Tjio and Levan, 1956; Ford and Hamerton, 1956 system.

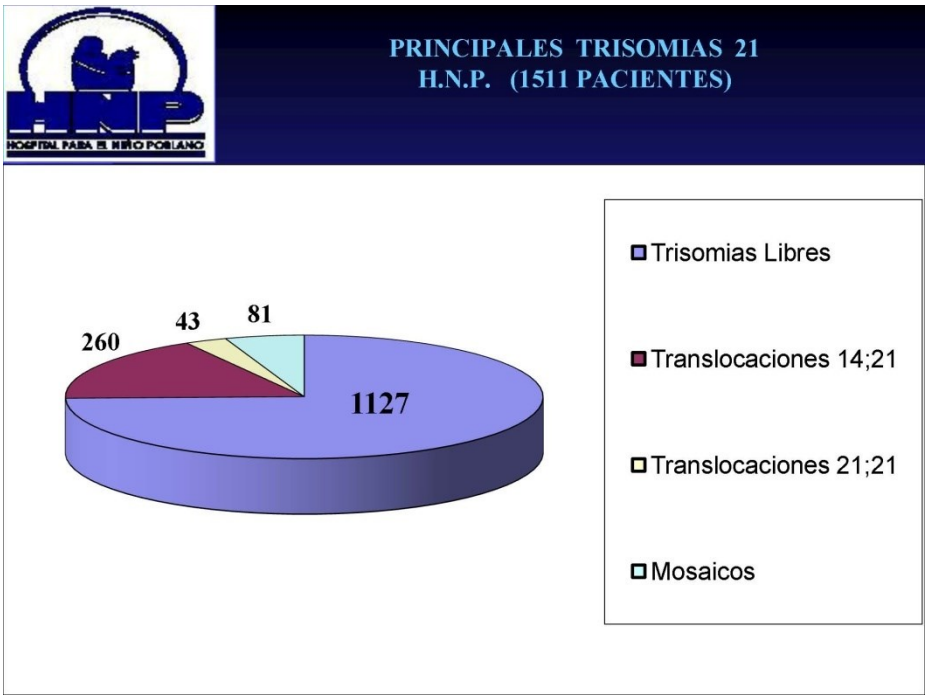


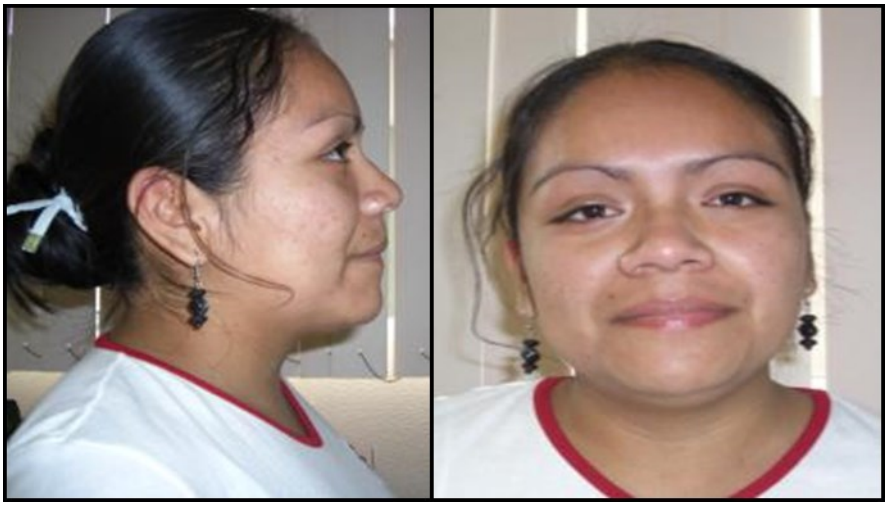

**Figure 2.** Of all the trisomy chromosomes, 1553 (33.6%), are observed 1511 (32.7%) were different types if compared to trisomy 21.

Taking into account the reported techniques von Winiwarter, 1912; Painter, 1922; Hinnebusch and Tilly, 1993, 48 chromosomes in chimpanzees were showed (the closest to the current human beings living relative). Some chromosome rearrangements do not parents cause disease healthy carriers, such as translocations, or chromosome insersions, although it can lead to a higher probability of a child with a chromosome disorder as found in this study **Table 1.**

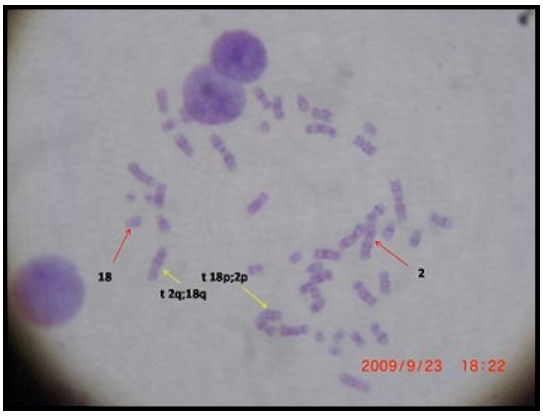
In this first study different chromosome aberrations were analyzed from 1 to chromosome 22 and sex chromosomes.

Patients as mentioned in **Figure 1**, are the result of 5000 karyotypes which were carried out from 1992 to 2011, showing different chromosome alterations 33.6% (1553) showed a chromosome Trisomy. Of these 33.6% of the population, trisomic, 32.8% (1511 patients) were diagnosed as Down syndrome (trisomy 21) **Figure 2** and 0.90% have other Trisomia **table 1**. Similar to the rest of the chromosome aberrations 0.93% (43 patients) **Table 1 & figure 1**.

In relation to chromosome translocations; a male patient with multiple oral tumors diagnosed by studies of oral histopathology as Cementoma Gigantiforme (being evaluated by oral histopathology and every 6 months for Oncology to avoid cell metastasis) with a translocation 46, XY, t(1;4) (q11q11) Aparicio et al., 2002; Aparicio et al., 2006 this case is different from a patient with multiple processes of abortion in their medical history without craniofacial alterations **figures 3 A, B & C,** and a chromosome translocation t(2;18).



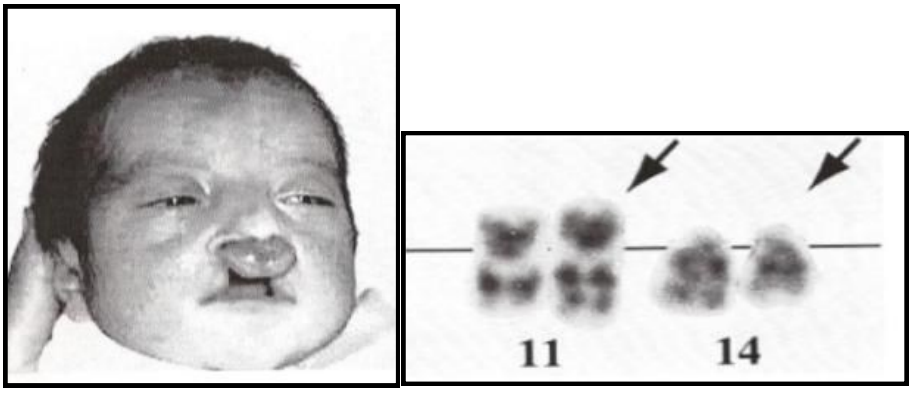
**A** **B**




**C**

**Figure 3 A.** Patient with multiple processes of abortion, **B.** without craniofacial dimorphism. **C.** the karyotype shows chromosome translocation t(2;18).

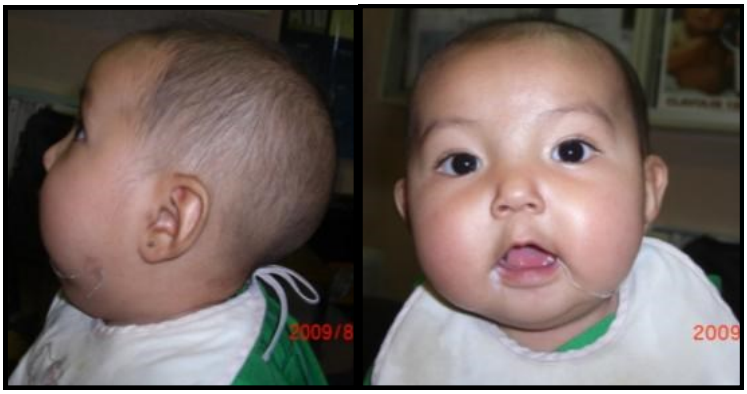
Another patient with altered craniofacial and oral cavity diagnosed as Opitz G/B.B.B syndrome, hypertelorism, unilateral cleft lip and oral and facial asymmetry with an unexpected translocation between the long arms of chromosomes 3 and 4, t(3q;4q) 46XX Aparicio et al., 2001. A male patient with hypertelorism, sinofris, small nose and hypoplasia of the nasal wings with a translocation of the short arm of chromosome 6 at the long arm of chromosome 9 t(6;9) was also analyzed, and was associated with cancer predisposition Aparicio et al, 2006 ; Aparicio et al., 2012**.** Two healthy carriers parents were observed in this study, one of them (mother) with 46XX, ins (10; 7) (q21; q23q35) translocation. However, her child with mental retardation revealed a translocation of the long arm of chromosome 10 t(10q+), giving rise to a partial Trisomy 7, Aparicio et al., 2010. This family was compared with a family where an affected mother as well as both son and daughter without phenotypic malformations. The mother was diagnosed with breast cancer having practiced mastectomy. The karyotype of the family members (mother, son and daughter) revealed a translocation between chromosomes 13 and 15 t(13;15), this type of cancer has also been associated with this chromosome aberration Aparicio et al, 2006. Several patients were diagnosed with lip and palate cleft bilateral, without craniofacial dimorphism. As well as the male patient who revealed a partial Trisomy of chromosome 14 (14q24-qter) due to a translocation t(11;14) (p25; 12)(p25.1;Q24.1) **figures 4 A & B**.



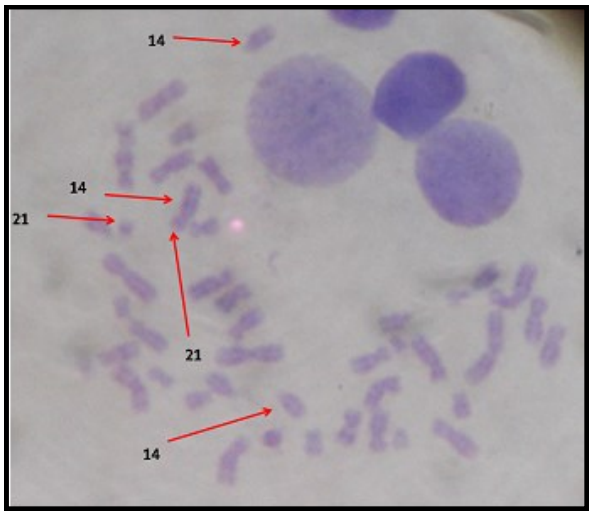

**A** **B**

**Figure 4 A.** Male patient with lip and bilateral cleft palate and Craniofacial dimorphism **B.** The karyotype revealed a partial Trisomy of chromosome 14 (14q24-qter) due to a t (11; 14), a balanced translocation (p25; 12) (p25.1;Q24.1).

Since the chromosome 11 is associated with several genetic syndromes Grossfeld et al., 2004. Similar case as the newborn patient with cleft palate and Dysplasia of ears with a 21; 14 chromosome translocation due to Trisomy 14, although the patient as shown does not show mayor craniofacial malformations associated **figure 5, A, B y C.**




**A** **B**

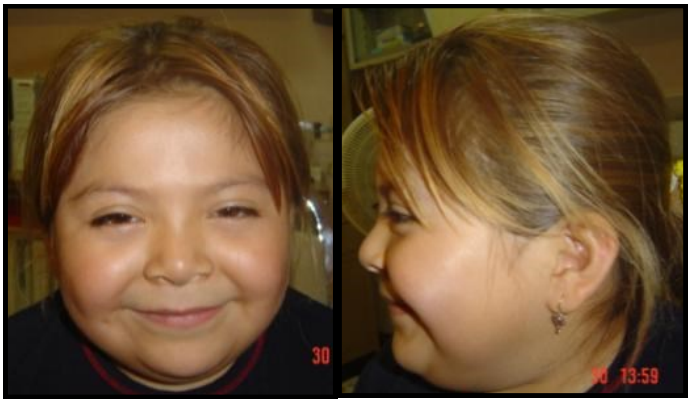



**C**

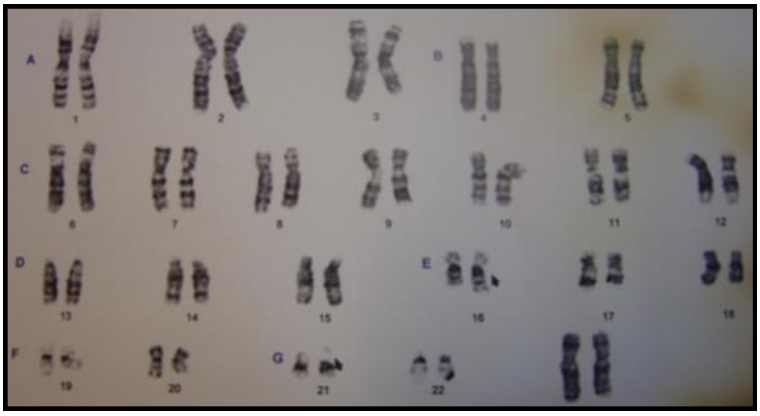
**Figure 5A.** Newborn with cleft palate B. patient And C. Dysplastic ears revealed a balanced chromosome translocation 21; 14 due to Trisomy 14.

However several patients with facial asymmetry associated with chromosome 16 were observed, for example, two cases where a male patient with craniofacial dimorphism was diagnosed as Parry Romber syndrome with duplication of the long arm of chromosome 16 (16q +) Aparicio et al., 2005, similar chromosome findings were observed in one patient.

Obesity and mental retardation were the main outcomes, as shown below in Figure 6A, B and c, where karyotyping revealed a doubling in the heterochromatin of chromosome 16, XX16qh region + chromosome 21 (46, XX, 16qh + 21PSTK +).



A B



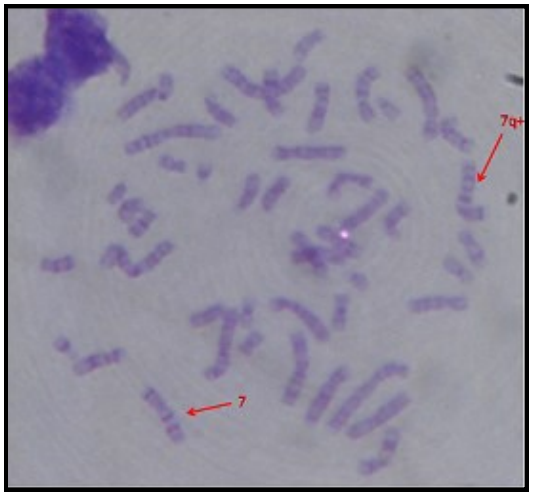
C

**Figure 6A**. Female patient with obesity and mental retardation **B.** Without phenotypic malformations**, C.** karyotyping revealed a doubling in the heterochromatin region of chromosome 16, 46, XX 16qh + chromosome 21 (46, XX, 16qh + 21PSTK +).

1553 patients were diagnosed with chromosome trisomy, where 32.8% (1511 patients) in **Figure 2** were diagnosed as Down syndrome (trisomy 21). This alteration is caused by an extra copy of chromosome 21 (trisomy 21). Characteristics include decreased muscle tone, skull asymmetrical and mild to moderate development disability Aparicio et al, 2009. An observed association was reported to chromosome translocations; 260 patients 46XY t (14;21)o 46XXt(14;21q) between the chromosome 21 long and chromosome 14, 43 patients 46XY t (21; 21) or 46XX t (21; 21) and 81 patients with mosaicism. In addition, 0.90% has other trisomies in different chromosomes **Table 1** as in the case of trisomy of chromosome 6 in a female patient diagnosed without phenotypic or craniofacial malformations, considering that Trisomy 6 is an extremely rare event and has been associated with aplastic anemia Geraedts and Haak, 1976 and secondary malnutrition.

Facial hypoplasia has been associated with a partial Trisomy of chromosome 7, 46XY, 7q +, it was diagnosed in a male patient and hypotelorism, sinofris, hypoplasic maxilla with restricted movement and limited mouth opening as shown below in **Figure 7 A, B and C,** important issue to the department of nutrition from the Hospital due to malnutrition, secondary to a bad feeding technique.

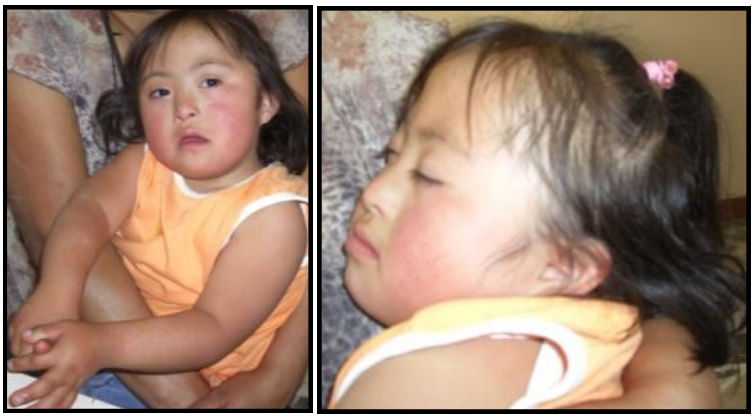
**A** **B**



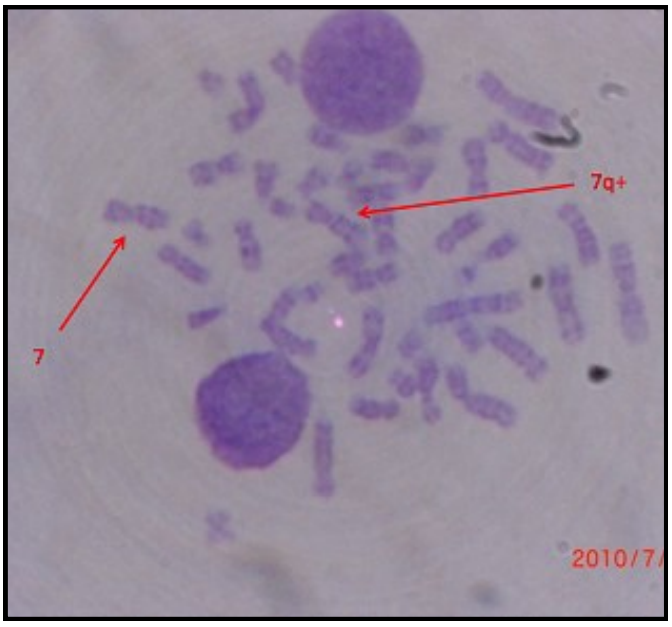

C

**Figure 7A.** Patient men with facial hypoplasia, B. maxillary have hypotelorism, sinofris and hypoplasia with restricted movement and oral limitation, C. karyotype revealed a partial Trisomy of chromosome 7, 46XY, 7q +.

Instead, facial hypoplasia was associated with facial asymmetry have been associated with partial Trisomy of chromosome 7,46XX, 7q +. **Figure 8 A, B & C.**



A B




C

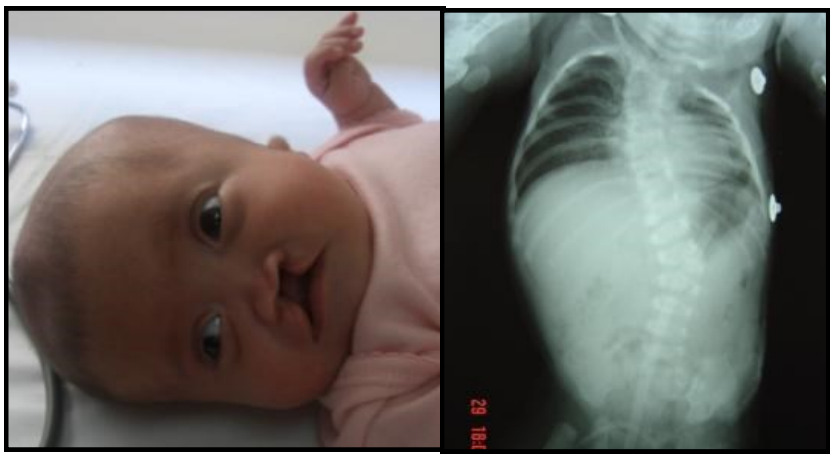
**Figure 8 A.** Female patient with facial hypoplasia and deviation of both eyes, small mouth **B.** facial and maxillary hypoplasia **C.** karyotype revealed a partial Trisomy of chromosome 7, 46XX, 7q +.

Edwards and Patau syndromes are rarely diagnosed at the Clinic of Pediatrics, taking into account that both syndromes share similar symptoms. A patient with Trisomy of chromosome 13 or Patau syndrome was evaluated with hypoplastic face, lip and bilateral cleft palate and small nose, craniofacial dimorphism, flexion of the fingers of both hands. Trisomy 13, is the least common autosomal trisomies, after the Down syndrome (trisomy 21) and Edwards (Trisomy 18) syndrome. The extra copy of chromosome 13 in Patau syndrome causes serious cardiovascular, neurological and facial defects that make it difficult for the infants to survive. However clinical data such as hirsutism, microcephaly, sinofris, mandibular hypoplasia, flexion of the fingers of both hands is observed in Edwards syndrome.

If compared to mental retardation, microcephaly hypoplasias and epileptic seizures reported in the patient with 22 trisomy, (47 XY + 22). The deletions of chromosomes have been observed in patients with neurologic damage and craniofacial alteration, like the male patient diagnosed as WolfHirshhorn syndrome, with typical phenotype: "Greek helmet" facial dimorphism: prominent glabella, ocular hypertelorism, epicanthal folds and broad nose with marked peak and microphthalmia associated to loss of

genetic material in the short arm of chromosome 4. A probable case of mutation of novo with suppression of the gene WHSC1 and others linked to nearby genes Aparicio et al, 1997; Aviña et al., 2008.

Other rare, chromosome alteration is the 13 ring which can be seen below in **Figure 9 A, B and C**, a patient with cleft lip and palate bilateral, dimorphism craniofacial and hypertelorism characterized by progressive spinal hemivertebrae deformity and 13 ring chromosome malformation Aparicio et al, 2000.



A B


C

**Figure 9 A**. Female patient with cleft lip and palate bilateral, B. hypertelorism and craniofacial dimorphism Characterized by progressive deformity of the spine with hemivertebrae C. karyotype revealed a malformation of the 13 ring chromosome.

Several malformations of the central nervous system associated with changes in the structure were observed with regard to the sex chromosomes, Craniofacial, as it is the case of monosomy as Turner syndrome, chimera, polyploidy XXX and Kllinefelter, among others. This study shows one of the patients, male diagnosed as syndrome of Opitz G/ B.B.B, hypertelorism and lip and cleft palate and craniofacial dimorphism. However the karyotype reveals a duplication of the sex chromosomes X 46, XXY (Klinefelter's syndrome). Men with this syndrome are usually sterile and has a higher incidence of retardation and dyslexia. During puberty, without testosterone treatment, some of them may develop gynecomastia. This patient is evaluated periodically by dentistry, maxillofacial surgery, endocrinology, nutriology and pediatric surgery. On the other hand few oral alterations were observed, in a female patient diagnosed as syndrome of Turner, with wide neck and short stature. Their karyotype revealed only one chromosome X (45XO) Turner syndrome (X instead of XX). In Turner syndrome, female sexual characteristics are present but underdeveloped, unlike this patient, another female patient with small eyes, Craniofacial dimorphism with hypotelorism, turricefalia and hypoplasia associated had a sex chromosomes trisomy 47, XXX (triple X syndrome). The patient with XXX tends to be tall and thin. They have a higher incidence of dyslexia and it is more likely to have learning problems and bad nutrition.

This type of sex chromosomes alteration is important in medical areas as nutrition, estomatology, psychology also important in chromosome events such as the chimera chromosome where two different cells that there are lines 46, XX/46, XY in the same patient associated with ambiguous genitalia. In relation to this patient it has been discussed among a medical group, if the patient should keep their sexual identity as a male or female, since half the information of the sex chromosomes are XX and half XY. It is important to decide the future sexual behavior, psychological, physical function, and external and internal genital structure of the patient. A decision shall be taken by the multidisciplinary group of physicians and the family of the patient for a better future of the patient Aparicio et al, 2010. Some genetic mutations are neutral and have little or no effect on nutrition and estomatology. However, other chromosome aberrations change the life of the patient and play a great role in the patient craniofacial and mental evolution.

Therefore, international associations and institutions such as the Institute de Vega offer information in relation to the human genome data. This version of Vega is information obtained from 2008 to 2011 and the structures of genes are presented in **Table 2.**

| Cromosoma | Genes | Total de bases | Secuencia de bases |
|---|---|---|---|
| 1 | 4,220 | 247,199,719 | 224,999 ,719 |
| 2 | 1,491 | 242,751,149 | 237,712 ,649 |
| 3 | 1,550 | 199,446,827 | 194,704 ,827 |
| 4 | 446 | 191,263,063 | 187,297 ,063 |
| 5 | 609 | 180,837,866 | 177,702 ,766 |
| 6 | 2,281 | 170,896,993 | 167,273 ,993 |
| 7 | 2,135 | 158,821,424 | 154,952 ,424 |
| 8 | 1,106 | 146,274,826 | 142,612 ,826 |
| 9 | 1,920 | 140,442,298 | 120,312 ,298 |
| 10 | 1,793 | 135,374,737 | 131,624 ,737 |
| 11 | 379 | 134,452,384 | 131,130 ,853 |
| 12 | 1,430 | 132,289,534 | 130,303 ,534 |
| 13 | 924 | 114,127,980 | 95,559 ,980 |
| 14 | 1,347 | 106,360,585 | 88,290 ,585 |
| 15 | 921 | 100,338,915 | 81,341 ,915 |
| 16 | 909 | 88,822,254 | 78,884 ,754 |
| 17 | 1,672 | 78,654,742 | 77,800 ,220 |
| 18 | 519 | 76,117,153 | 74,656 ,155 |
| 19 | 1,555 | 63,806,651 | 55,785 ,651 |
| 20 | 1,008 | 62,435,965 | 59,505 ,254 |
| 21 | 578 | 46,944,323 | 34,171 ,998 |
| 22 | 1,092 | 49,528,953 | 34,893 ,953 |
| X (cromosoma sexual) | 1,846 | 154,913,754 | 151,058 ,754 |
| Y (cromosoma sexual) | 454 | 57,741,652 | 25,121 ,652 |
| Total | 32,185 | 3,079,843,747 | 2,857,698,560 |

**Table 2.** (http://www.ncbi.nlm.nih.gov/genome/seq/) sequencing of the human genome has provided a lot of information about each of the chromosomes. This table contains statistics for the chromosomes, based on information about the human genome of the Sanger Institute in the annotation of vertebrate genome (VEGA) database. Vega.Sanger.ad.uk, 2008.

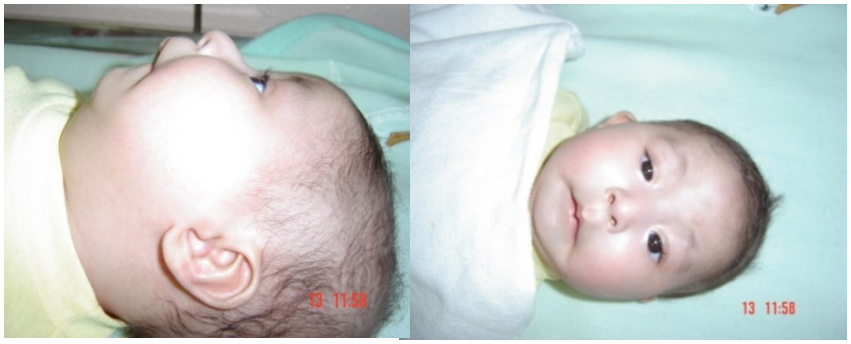
An abnormal number of chromosomes or chromosome sets, aneuploidy, may be lethal Huret et al., 2000; Huret et al., 2000; Vega.sanger.ad.uk, 2011; Hsu TC, 1979; Kelman and Kelman, 2004 Some of the main chromosome alterations in this study are seen in **table 1 and Figure 1**. Genetic counseling can be provided in multidisciplinary form with the necessary knowledge, which is important for families carrying this type of chromosome rearrangements. The gain or loss of DNA from chromosomes can lead to a variety of genetic diseases, as it is possible to be observed in this study. It could be important if chromosome aberrations could be diagnosed, not only in the practice of genetics but also nutrition, dentistry and pediatrics, providing to the patient an early management, which will contribute to an accurate diagnosis, early treatment and a better genetic counseling and a better quality of life for the patient and family.

## AUTOSÓMIC CHROMOSOMES

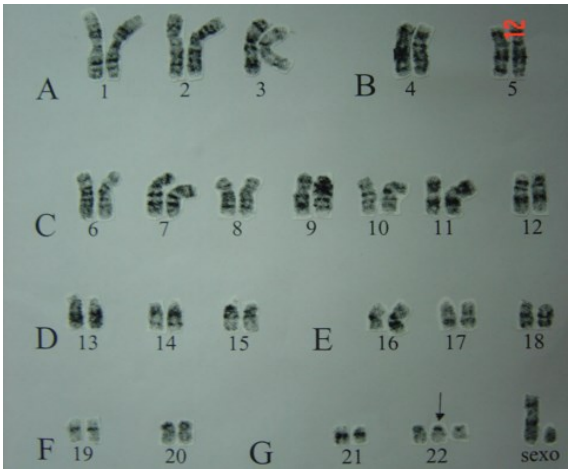
Chromosomes as already mentioned, are found in the nucleus of all cells of the body. They carry the genetic characteristics of each individual. Each chromosome has one short arm designated as "p" and a long arm designated as "q" Denver Conference 1960. The pairs of human chromosomes are numbered from 1 to 22, with a 23 pair, one X chromosomes for men, and two X chromosomes for women. Persons with a copy have an extra chromosome added to one of the normal pairs Crawfurd MDA, 1961. The syndrome of Down according to the (SD) Ibero-American Down 21 Foundation, 2007 is the set of characteristics that are manifested in a person by the presence in cells of three chromosomes 21 instead of a pair. For this reason it is also called trisomy 21. In contrast to the chromosome 22, The genetic material of both chromosomes (21,22) represents only the 1.53.0% of the total genomic DNA. Genes are the smallest structures of human chromosomes. The sequences obtained from the 22 contain a minimum of 545 genes and the estimated number of genes containing chromosome 21 is located between 200 and 400 genes. In relation to the human genome, chromosome 21 was the second human chromosome to be fully sequenced, while the knowledge of chromosome 22 is going to be very useful to show other major units from the rest of the genome. The clinical interest of nutrition and dental is by morphological and numerical alterations of both chromosomes 21 and 22 with data on malnutrition.

In relation to SD or trisomy 21, the clinical features Carnevale A., 1973 that have been affected are: stature, brachycephaly, flattened and high forehead, epicanthal folds, in 50% of cases are congenital cardiopathies Freemam et al., 1998; McElhinney et al., 2002 mainly atrial septal defect (ASD), communication Ventricular septal defect (VSD), and patent ductus arteriosus (PCA) persistence. The cognitive level varies from 20 to 60 (an average intelligence reaches a value of 100) but higher values can be achieved with a good nutritional therapy and early rehabilitation. There is a higher incidence of congenital hypothyroidism Grozinsky Glasberg et al., 2006 and organic, nutritional and dental alterations in patients with syndrome of Down Sindoor SD, 1997. The oral cavity is small, macroglossia with frequent habits of digital suction, scrotal tongue, fissured or lobulate, mouth breathing that in addition to inappropriate development of the palate, produces xerostomia in mucous membranes, being frequent infection by opportunistic germs stomatitis and angular cheilitis in labial commissures, disturbances in tooth eruption. A change in the pH of the saliva as well as an increase in the content of sodium, calcium, uric acid and bicarbonate secretion, decreased speed is observed to suffer from tooth decay and periodontal problems [41] influencing secondary malnutrition. They have, in addition, a risk in the development of medical and pediatric diseases as leukemia (acute myeloid leukemia). In comparison with the trisomy 22. Dunham et al., 1999 investigations began 40 years when Martín Municio in 1999 Nowell in 1960, studying the karyotype of a patient with chronic granulocytic leukemia, described a tiny chromosome 22 Daley and Van Etten 1990; Rowley JD 1996, since then called chromosome (Ph1) Philadelphia, recalling that the work was carried out in that city. Chronic granulocytic leukemia is a malignant disease. The Philadelphia chromosome is currently an important prognostic marker and therapeutic this pathology. It was Hsu et al., 1971 associated with chromosome 22 pathologies. This chromosome aberration, Bueno M. Bueno et al., 1982; Bueno et al.,1990; Bueno M del Amo 1969 **,** has variable mental deficiency, disorders of language and dysmorphic phenotype characterized by mental weakness, pondoestatural, micrognathia, Craniofacial dysmorphia hypotrophy, large rear rotating ears, tubers or sinus or pits, long and deep philtrum and congenital heart disease. Trisomies are described in this study, as frequent chromosome alterations, in which an extra chromosome 21 or 22, is present in some of the cells of the body. The severity of symptoms is variable. A Trisomy in chromosome 22 additional **figures 1 A, B and C,** Pediatric Clinical features; hemidistrofia, webbed neck and microcephaly, psychomotor and seizures retardation. This disease is detected at birth, and can be diagnosed during pregnancy or after birth, the karyotype is definitive for

diagnosis.



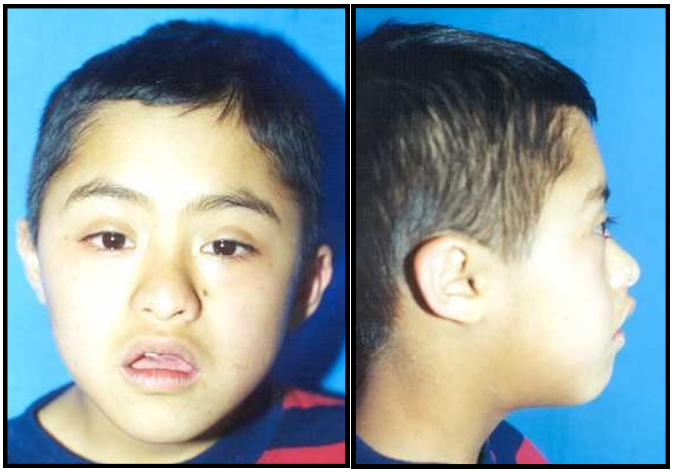
**A** **B**



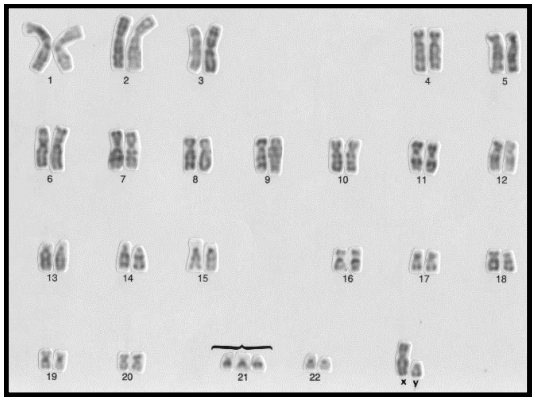

**C**

**Figure 1 A.** Male patient with Trisomy 22, with mental retardation, microcephaly **B.** epilepsy and hypoplasia, hypoglycemia **C.** karyotype reveals Trisomy 22, 47 XY + 22.

This differs to the Pediatric Clinical features in trisomy 21 **figures 2 and 3 A, B and C**; stature, high and flattened face, language and dry lips, epicanthal folds, have been diagnosed in 50% of cases with congenital heart disease, mainly atrial septal defect (ASD), Ventricular septal defect (VSD), and patent ductus arteriosus (PCA) persistence, which causes them a secondary malnutrition.

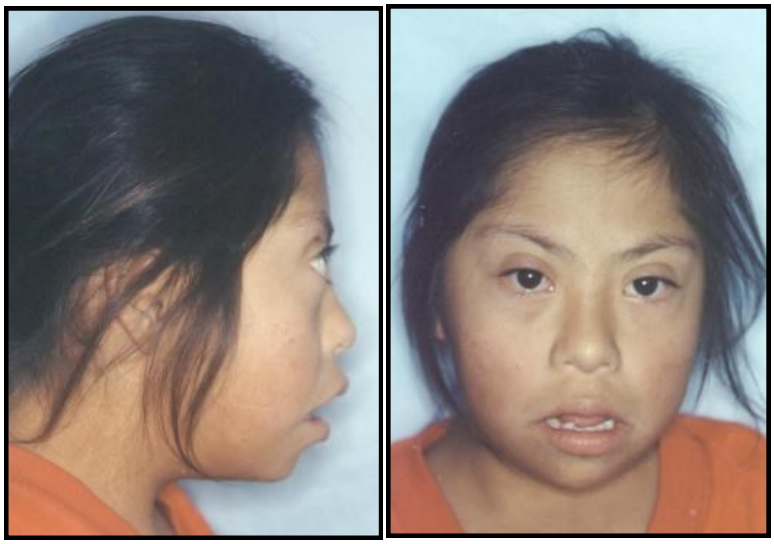


A B

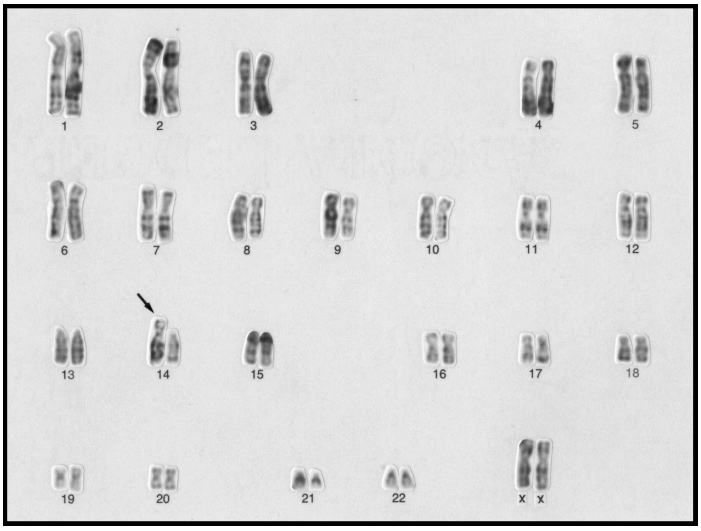


C

**Figure 2 A & B**. A male patient with Down syndrome and mental retardation**, C.** karyotype reveals a trisomy 21 (47, XX, + 21) caused by an event of nondisjunction.



A B



C

**Figure 3 A & B.** Female patient with Down syndrome, mental retardation and epilepsy. **C.** the karyotype reveals a robertsonian translocation in the long arm of chromosome 14 (46, XX, t(14;21q).

There are three cytogenetic types in relation to trisomy described and widely known:

**REGULAR:** characterized because there is one chromosome 21 extra free **figure 2 C**, in other words, there are 3 chromosomes 21 instead of a pair. This type of alteration is the most frequent: 94% of the cases.

**TRANSLOCATION**: After free Trisomy, chromosome translocation is observed. In this variant the extra 21 chromosome (or a fragment thereof) is joined to another chromosome (often to one of the two chromosomes pair 13 or 14), by which the chromosome formula is of 46 chromosomes. In this case there is a problem with the chromosome disjunction, but one of them carries a "extra" fragment with the fused chromosome genes. For the purpose of genetic information still being a trisomy 21 since it duplicates the genetic information. The rate is 3% of all patients with Down syndrome and should be ruled out if there are healthy carriers parents. Approximately 1% of all cases of Down syndrome, one of the parents (father or mother) is a carrier, i.e., does not have Down syndrome, but by having an abnormality in the structure of their chromosomes can pass it on to their offspring.

**MOSAICISM:** Appears in 1.5% of children with Down syndrome and 1.8% of the trisomy 22. It corresponds to the situation in which egg and sperm have normal 23 chromosomes, but along the first divisions of that cell and his daughters emerged in some of them the phenomenon of the nodisyuncion of the pair of chromosomes 21 or 22 so that a cell will have 47 chromosomes, three of whom shall be of the pair 21 or 22 from there, all the millions of cells that arise from that abnormal cell will have 47 chromosomes (will be trisomic) and the normal rest. The age of parents (under 20 or over 40 years of age) is very important to have a baby with trisomy 21 or 22, although the frequency in the population is greater compared to the Down syndrome compared with Trisomy 22.

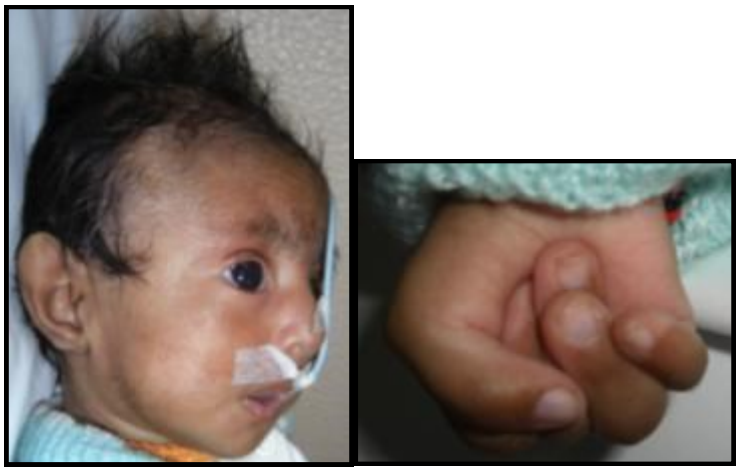
There is no specific pediatric, nutritional or estomatologic treatment for both trisomies. However some newborns may need surgery as in trisomy 21. Nutritional therapy is a great help for these patients with different capacity, to achieve their full potential. With regard to the oral rehabilitation, done a wide range of dental studies and management mainly in Down syndrome, reporting that these patients can be treated in the clinic of Stomatology Deassai NN, 1997; Bianchi et al., 1991**.** Assessed the rate of dental caries, loss of teeth, oral hygiene, nutritional therapy and pediatric. Emphasized in the oral health of patients with physical disabilities and psychological Santoro et al., 1991 and there are programs of oral hygiene and dental and periodontal disease prevention Ulseth et al., 1991. In different studies has been documented that patients with Down syndrome

and diminished skills, dental treatment and regular dental care as well as secondary malnutrition have an inadequate oral health. Some of them require a more specialized estomatologic, medical and nutritional management Allison et al., 2000. The dental needs of these patients, a study in patients with trisomy 21 showed the importance of elderly care in comparison with a normal population. Since they have increased susceptibility to diseases periodontal Randell et al., 1992. 30 people studied with SD by comparing them with another Control Group, there was an increase in the frequency of periodontitis (gingivitis) and lack of teeth (edentulism) was more frequent in the group with SD.

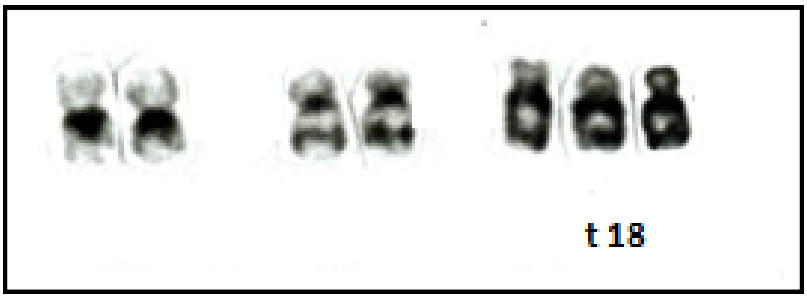
The fact that 50 maternal and paternal errors contribute in a similar percentage to the phenomena of no disjunction of the gametes which contrasts with the increased frequency of maternal as trisomies 21 and 22 errors. This is important to be able to offer genetic counseling to assess whether the affected family has more likely to file a new case than the general population Gabre P., 2000.

In conclusion both trisomy 21 and 22 presented an increase in susceptibility and a greater risk to the general population, the development of pediatric diseases as leukemia already found in patients with Down syndrome. This type of chronic granulocytic leukemia have been associated with chromosome 22 that is called a chromosome (Ph1). Philadelphia as mentioned before. It is important to provide timely and accurate genetic counseling parents and assess the risk of recurrence in order to prevent another genetic alteration in a future pregnancy.

**Edwards Syndrome (t18).** It's a polymalformative syndrome, known as Edwards syndrome (SE) Edwards et al., 1960**, Figure 1. A, B and C** secondary to a chromosome alteration by the presence of three 18 chromosomes. Its frequency is calculated approximately 1/13000. It occurs in all races and geographical areas.



A B




C

**Figure 1 A.** Male patient with hirsutism, microcephaly, sinofris, generalized hypoplasia **B.** over position of the fingers of both hands **C.** The karyotype reveals a Trisomy 18 regular free non-hereditary.

95% of cases correspond to regular free Trisomy product of nondisjunction, being the remaining Trisomy **Patau et al., 1960** by translocation. Partial Trisomy and in this Trisomy mosaicism, shows an incomplete phenotype, with absence of some of the typical anomalies. A unique chromosome region, critical and responsible for the syndrome has not been identified. Seems to be necessary duplication of two zones, 18q1221 and 18q23 to allow the typical phenotype of, with an area, 18q12.3q21.1 with strong influence in mental retardation and facial hypoplasia, mainly of the first and second branchial arch, upper jaw and lower and sometimes with micrognathia or retromicrognacia which gives a secondary malnutrition this genetic alteration Campusano Carlos, 1972; Yunis et al., 1964.

Main clinical features.

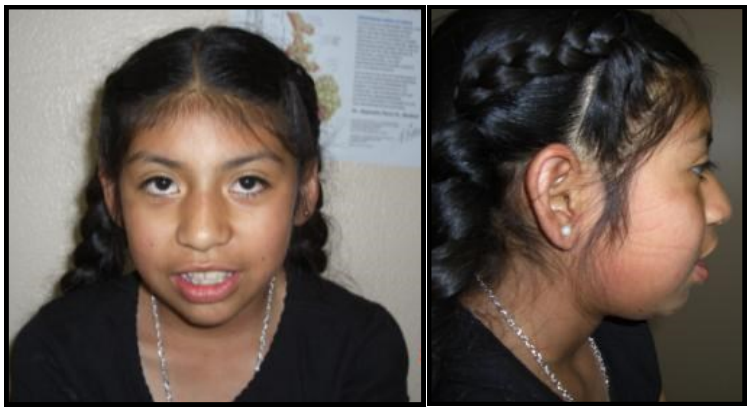
1. Prenatal slow growth with malnutrition
2. Hypoplastic facial features.
3. Position of the 4 phalanges on the Middle phalanx.
4. Congenital heart (cardiac malformations 95%).
5. Diaphragmatic hernia
6. Moderate to severe mental retardation, generalized hypertonia, dolichocephaly with microcephaly, micrognathia.

Other secondary clinical features in these patients include: narrow Pelvis, equinovarus foot uni or bilateral, low ears, short sternum, folds of hands pointed, prominent heel, cryptorchidism or hypertrophy of clitoris, kidney and digestive malformations (among other multiple internal alterations). In relation to survival is 50% mortality to 1 month and 10% 1 year alive. It is very important to maintain adequate nutrition if possible with the intervention of Gastrostomy tube to ensure good nutrition. The relationship of gender is a male for every 4 women. Patients die in early childhood and only 14 cases with prolonged survival have been described.
It is more frequent in older mothers. From age 35 to 40, the frequency increases progressively. 1/2500 live births at age 36 to 1/500 to 43. In women age > 35 years old, or with previous child with Trisomy 18 or SD should suggest a prenatal diagnosis by amniocentesis in following pregnancies especially if there is a chromosome translocation to discard unless one of the parents is healthy carrier and be able to assess the risk of recurrence in a future pregnancy.

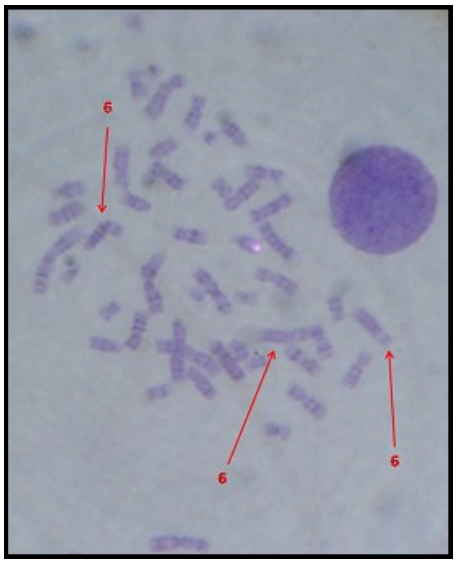
**6 Chromosome trisomy.** Chromosome 6, partial Trisomy 6q is an extremely rare chromosome disease, in which the portion distal end of the long (q) arm of chromosome 6 (6q) appears three times Hahnemann and Vejerslev 1997; Hsu et al., 1997**.** This disease is characterized by craniofacial malformations (head and face), unusually short and winged collar, Contracture in flexion of the fingers, the growth retardation, severe psychomotor retardation (acquisition of skills requiring coordination of muscular and mental activity), and mental retardation. The level of nutrition is good without clinical data for malnutrition.

The range and severity of symptoms and physical signs vary from one case to another, depending on the length and location exact duplicate portion of chromosome 6q. In the majority of cases, the chromosome 6, partial Trisomy 6q may is due to a chromosome in Ledbetter and Engel, 1995 parental translocation. In relation to chromosome 6, was described total Trisomy

Geraedts and Haak, 1976; Moormeier et al, 1991; Jonveaux et al., 1994; La Starza et al., 1998; Mohamed et al., 1998; Onodera et al., 1998; Wong KF. 2004 , Dellacasa et al, 1993; BrondumNielsen et al., 1993; , Uhrich et al., 1991; Bartalena et al. 1990; Chase et al., 1983. The symptoms that occur vary depending on each case however the **figure 1 A, B and C** shows a female patient with a complete 6 Trisomy, with delay in growth without data on malnutrition and mental development, without added congenital craniofaciales malformations, short neck and normal hematological studies up to the present time.



**A** **B**




**C**

**Figure 1. A & B.** Patient diagnosed without phenotypic malformations **C.** Complete trisomy of chromosome 6 is revealed.

**Deletions: Síndrome De Wolf hirshornn (4P).** WolfHirschhorn (WH) Syndrome Zankl et al., 2001; Hirschhorn et al., 1965; Lurie et al., 1980, is a rare genetic disorder secondary to a chromosome abnormality by a deletion or distal microdeletion of the short arm of chromosome 4. Also known as partial deletion of chromosome 4, 4 p partial monosomy of chromosome 4, 4 p chromosome region, Wolf Hirschhorn syndrome partial 4 p Battaglia et al., 2001**,** syndrome of distal microdeletion of the short arm of chromosome 4.

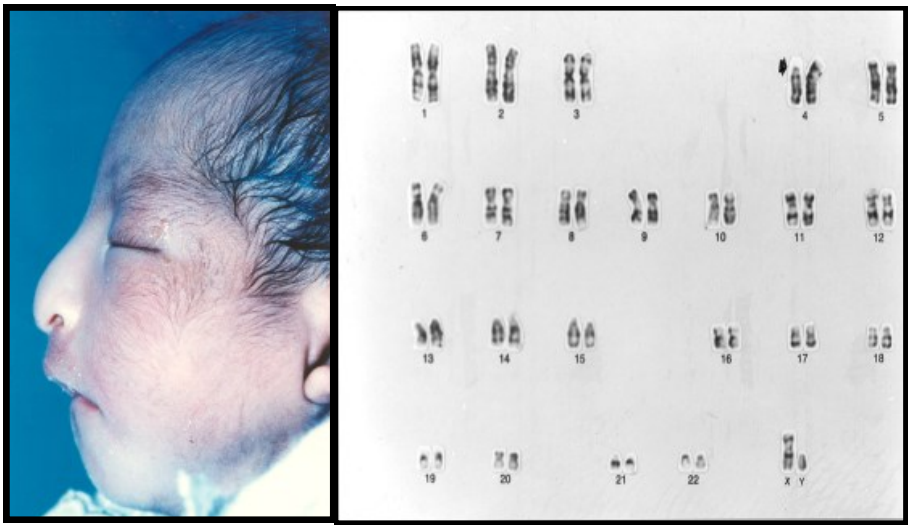
In relation to the history of this syndrome; 1961 Cooper and Hirschhorn: report first case 1965 Ulrich Wolf, Kart Hirschhorn Wolf et al., 1965 described as syndrome Zollino et al., 2000, full delineation of the syndrome.

Etiologic factors Bergemann and Cole, 2005 such as any chromosome alteration are considered; 87% Novo mutation, 13% descendant of a carrier of a translocation Tupler et al., 1992, 12-% unbalanced chromosome 4 ring, mosaicism, sporadic translocation.

Its main clinical manifestations are: malnutrition the growth retardation, malformation craniofacial, severe psychomotor retardation, and mental retardation as well as neurological alterations different Moretti et al., 2001;

Aparicio et al., 1997. Taking into account the complex facial skull **I**wanowski et al., 2005. Skull; Microcephaly, dolichocephaly, defects of the scalp. Face; Facial appearance in Greek helmet, broad forehead, prominent glabella, Microretrognatia. Eyes; Orbital flat flange, palpebral ptosis, Hypertelorism, palpebral fissures of tilt downward oblique, internal epicanthal folds, bilateral Coloboma, divergent strabismus, cataracts, sparse eyebrows. Nose; Very broad nasal bridge, large rectangular nose, anomalies of the lobe. Mouth; Cleft lip and cleft palate, mouth in carp, Cupid bow marked, downstream labial Commissures.

These alterations are associated with patients with a variable degree of malnutrition. **Figure 1 A, B, C and D** notes the case of WH Aparicio et al., 1997, with emphasis on craniofacial Pediatric alterations. Hypotonic patient with microcephaly and peculiar face of "Greek Warrior helmet": broad forehead, prominent glabella, hypertelorism, epicanthus internal and flat nose; patient delay in growth pre and postnatal, psychomotor retardation, and seizures. Confirmation of the diagnosis was carried out with G-band karyotype, **figure 1 D** where it can be seen a deletion of the short arm of chromosome 4.



**A** **B**

**Figure 1 A**. Male patient diagnosed as WolfHirshhorn syndrome, with a typical phenotype, as "Greek helmet" facial dimorphism: prominent glabella, ocular hypertelorism, epicanthal, nose beak and microphthalmia folds **B.** Karyotyping revealed a loss of genetic material in the short arm of chromosome 4, a case de Novo mutation likely suppression of the gene WHSC1 and other contiguous genes linked with.

**Translocations: 6;9, 13;15, 7;10 Y 1;4 Human Cancer Etiologic Factors.** Chromosome translocations have been related to the presentation of cell growth or oncogenic processes. There are agents that damage the normal structure of the chromosome producing alterations such as breaks and loss of the hereditary material, these agents can be physical in nature (radiation), chemical (drugs) and biological (virus). This brings as consequence a structural damage if cellular repair mechanisms do not exist or are inefficient. However, in evolution, there are mechanisms such as translocations, chromosome structure-changing naturally.

The chromosome translocation, can give a loss of hereditary material and as a result a change in the position of a chromosome segment, but without changing the total number of genes. A non-reciprocal translocation, is an aberration of two breaks that result in an inaccurately exchange of segments of non-homologous chromosomes in some cases are Etiologic Agent of partial Trisomy while a reciprocal translocation, is an aberration of two breaks that result in an accurate exchange of segments of non-homologous chromosomes resulting in single translocations. It has been reported a wide variety of studies with respect to translocations of chromosomes different with other non-counterparts, in cytogenetic studies, 2, 9, 11, 19 and 20 chromosomes have been observed most frequently involved in comparison with the chromosomes Y, 10, 13, 15, 16 y 21. We have analyzed the points of rupture, suggesting that the loss of specific chromosome regions may be important in the pathogenesis of syndromes or chronic diseases in the medical and eestomatologic area Dohner et al., 1992**.**

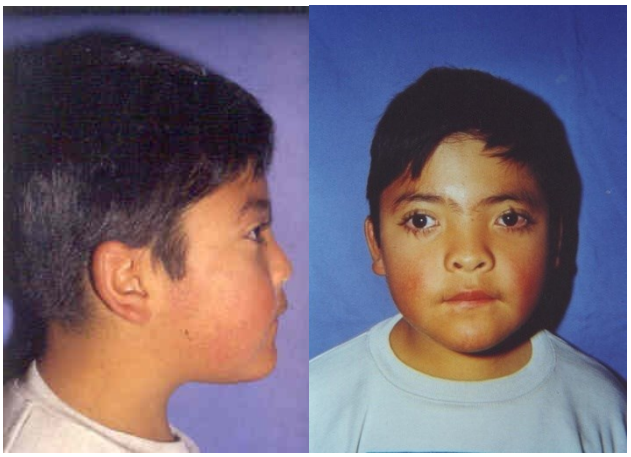
Between the balanced translocations observed in the general population, the chromosomes of the D group (D; (D), are the most frequent Francisca and Antonio, 1971**.** However, there are reports of benign and malignant cellular processes with some structural and functional features in specific regions of human chromosomes of groups D and G Wang et al., 1992**,** causing epithelial benign tumors of the mammary gland in some robertsonian translocations Hamerton JL., 1968 and fibrocystic mastopathy in case t (13: 15) **Figure 2 A, B and C,** without phenotypic alterations Aparicio et al., 1995), and also with malformations without data on malnutrition and mental retardation Mangelschots et al., 1992.

The objective of this chapter is the analysis of two patient male one of them with a dimorphic Phenotype and a chromosome aberration

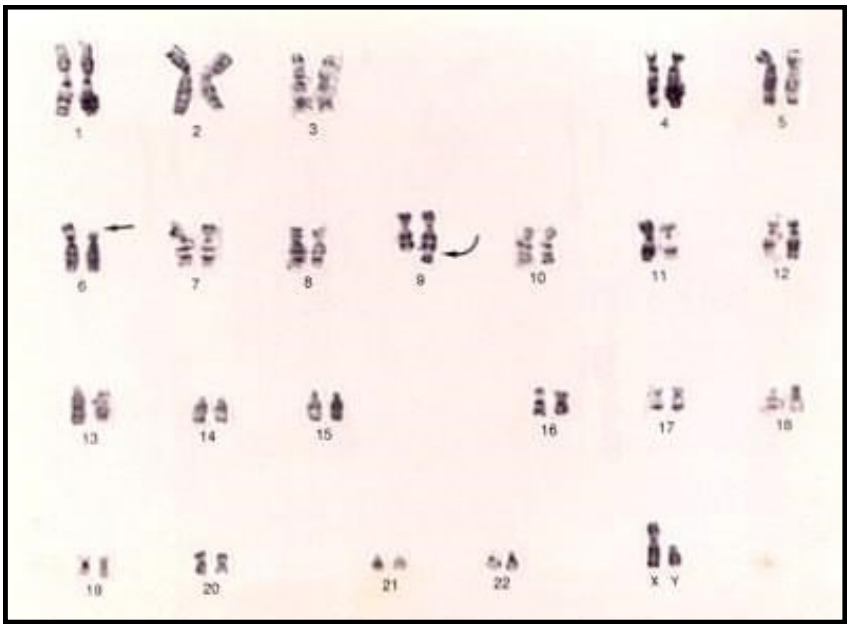
characterized by a translocation 46, XY, t(6;9) (6p:9q) **Figure 1 A, B and C,** which has a latent risk of acquiring a myeloproliferative syndrome; and other case, associated with oncogenic predisposition with another type of translocation, t (13,15-)(q13:q15) **Figure 2 A, B and C.** It is important a periodic oncohematológical assessment, to provide early therapy and thus obtain a better prognosis for the patient, as well as to offer a better nutrimental and estomatologic rehabilitation therapy and avoid the side effects of cancer or after management of antioncologic treatment such as the application of chemotherapy and/or radiation therapy. This is important to look after the patient in case of osteoradionecrosis, immunosuppression or alteration in the healing post stomatologic treatment, so a good parameter of nutrition in this type of patients is basic.

The first mentionated case is a male patient. He was removed at 4 years of age an accessory toe of the left foot (polydactyl). With a healthy 10-year-old sister. He has obesity, idiocia face, hypertrichosis, external shunt from the right eye, broad and flattened nasal bridge **figure 1 A and B**, long eyelashes, with psychomotor retardation and partial anodontia. Thorax and abdomen with abundant adipose panniculus secondary to a bad diet. External genitalia with a shorter penis, being the scrotal pouch with testicles and testicular volume of 1 cm., less than appropriate for this patient with cytogenetic study **figure 1 C** was held with bands G technique, found in 20 metaphases observed (100%), a translocation between the short arm of chromosome 6 and the long arm of chromosome 9 giving a chromosome formula 46XY, t(6:9) (6p9P; 6q9q:6p). Cited at the services of Pediatrics, nutrition, pediatric dentistry and oncohematology with laboratory exams as hematic, blood chemistry, and bone marrow aspirate.

Syndromes and diseases in relation to not homologous chromosome translocations, as in the case of leukemia (LLA) in humans which has been observed in t(1;19) Uckun FD., 1998 have been reported. Genetic translocation with clinical case reports exist t(6;9) (p23; q34) associated with leukemia chronic myeloid (CML)



A B

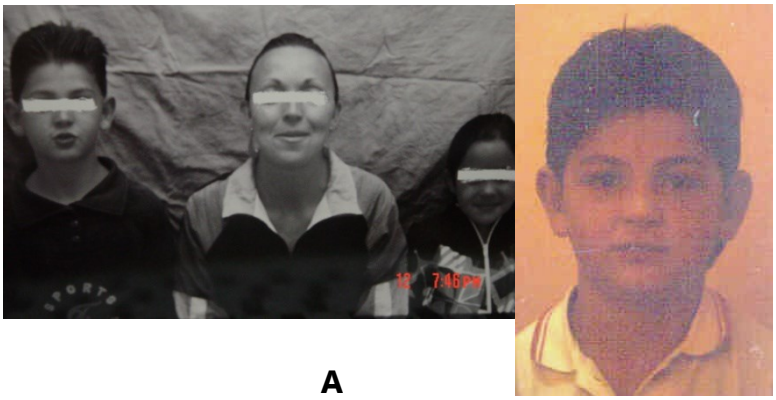


C

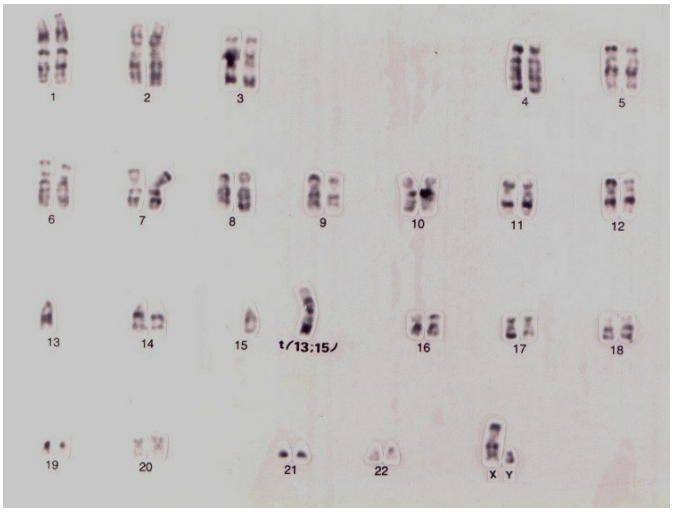
**Figure 1 A.** Male patient with hypertelorism, sinofris B. nose small, hypoplasia of the nasal wings. C. karyotype revealed a chromosome translocation of the short arm of chromosome 6 of the long arm of chromosome 9 t(6;9).

Jadayel et al., 1995; Awaya et al., 1995, and acute (AML) Alsabeh et al., 1997; Toyosawa et al., 1997; Hollings et al., 1995, in patients where the disease begins after 18 years of age, taking into account that the patient in this study has just 8 years, and although he has a genetic predisposition, by their chromosome aberration would be considered as it is still in early stages to start oncologic alteration, which is very important for early detection and a full estomatologic, pediatric, and nutritional management until he starts any kind of cancer condition. (AML) in patients with t(6;9) are reported in general with a poor prognosis, basophilia, myelodysplasia with sideroblastos**,** and with little chance of healing treatment with conventional and even a short period of survival has been reported, similar to the case of the patient with the translocation 13; 15, where the mother has a total mastectomy. In the case of his son and daughter who inherited the same translocation without malnutrition, it is very important to monitor them every six months. However early treatment, could at any given time to provide a better prognosis, where services as oncology, internal medicine, pediatrics, nutrition, genetics and dentistry are directly involved.

Like chromosomes 6/9 t (6; 9) and 13/15 t (13: 15), has been observed by studies of fluorescence (FISH) than other chromosomes, such as chromosome number 22 (q11.2), is also involved in cases of leukemia Gonzalez and Notohamiprosjo, 1996. It can be appreciated the importance of some translocations in ooncogenes activation due to the loss or change of the genetic material, as an etiological factor not only of hyperplasia in solid tumors has been observed in the translocation t (13: 15), but also of Leukemic syndromes, secondary to the t(6;9) as reported in this study which is important to know it by the medical doctor. This suggests that structural alterations in some chromosomes are important initiators of tumor processes. It has been reported that different animal species have differentiated among themselves by constant genetic changes through chromosome rearrangements like translocations being direct mechanisms in the evolution of the genome human Byatt et al., 1997 or natural recombination observed between chromosomes of some varieties of plants and/or animals Gobbi et al., 1997, concluding that the chromosome translocation occurs more often than it is imaginable, as part of the normal process of evolution of living beings; Although this type of chromosome abnormalities can be de novo, heredity plays a central role as is the case with the t (13: 15), where as mentioned previously, two of three children have such translocation, **figure 2 A, B & C.**



A




B

**Figure 2 A.** A family consisting of a mother, son and daughter without phenotypic malformations. Where the mother was diagnosed with breast cancer and mastectomy, secondary to a chromosome translocation. **B**. between chromosomes 13 and 15 t (13: 15). Same as the translocation inherited by his son.

This supports the theory of a relationship between the loss of chromosome material that results in deletions of oncogenic, important sites in the control of development and cell growth Awaya et al., 1995 as it is the case with this patient who is currently asymptomatic, with a good nutrimental state, probably by his young age and the importance of continuing with a surveillance by oncology, pediatrics and nutrition,

because of the high risk that presents due to translocation of chromosomes 6, 9, 13 and 15 among others as a likely trigger for an oncogenic activation Schneider et al., 1997; Johannesson et al., 1997; Sawyer et al., 1995; Yeo et al., 1996 to provide early handling and treatment, more accurate, and thus offer a better quality of life to patients with genetic risk for this specific chromosome aberration.

An analysis an explanation of the association between phenotypic alterations and the chromosome, chromosomes 13 and 15 Aparicio et al., 1995, who can be coincidental or be related to translocation was reported;

a. There is a microdeletion undetected at the chromosome level.

b. That a functional gene in one or both of the chromosomes was divided in two chromosomes and in these cases can be an abnormal hybrid gene.

c. That active genes are in the vicinity of heterocromatic regions.

d. In cases where there are more individuals carriers in the family, most likely is that the association is coincidental, however in cases 1 and 3 where the translocation is de Novo. This genetic mutation causes an alteration in these tissues and causes that they start to produce an abnormal number of white blood cells, alterations in these cells producing organs.

e. Which can give as a result systemic diseases such as: chronic leukemia or acute malignant (AML) considered a disease, although many times it is called "blood cancer", actually affecting the tissues involved in the formation of blood cells, i.e., to the bone marrow, spleen, and lymph nodes.

**Translocation 7;10.** Balanced translocations should not present phenotypic malformations there is no loss or gain of genetic material, as mentioned. However there are reports that associated them with clinical alterations as malnutrition, mental retardation, congenital malformations, or neoplastic processes as mentioned Aparicio et al., 1995) The present study also shows a patient with a balanced translocation and phenotypic or clinical alterations. As shown in **Figure 3 A,** where the mother is healthy carrier of chromosome 7 long arm deletion **Figure 3 B** this fragment through translocation joined the long arm of chromosome 10, 46xx, ins (10; 7) chromosome translocation (q21; q23q35). However the **figure 3 C** shows her child with several malformations and mental retardation, who inherited the chromosome translocation from his mother, the long arm of

chromosome 10 t (10q +) **Figure 3 D,** causing a partial Trisomy 7 in the patient and giving as a result the above clinical alterations.

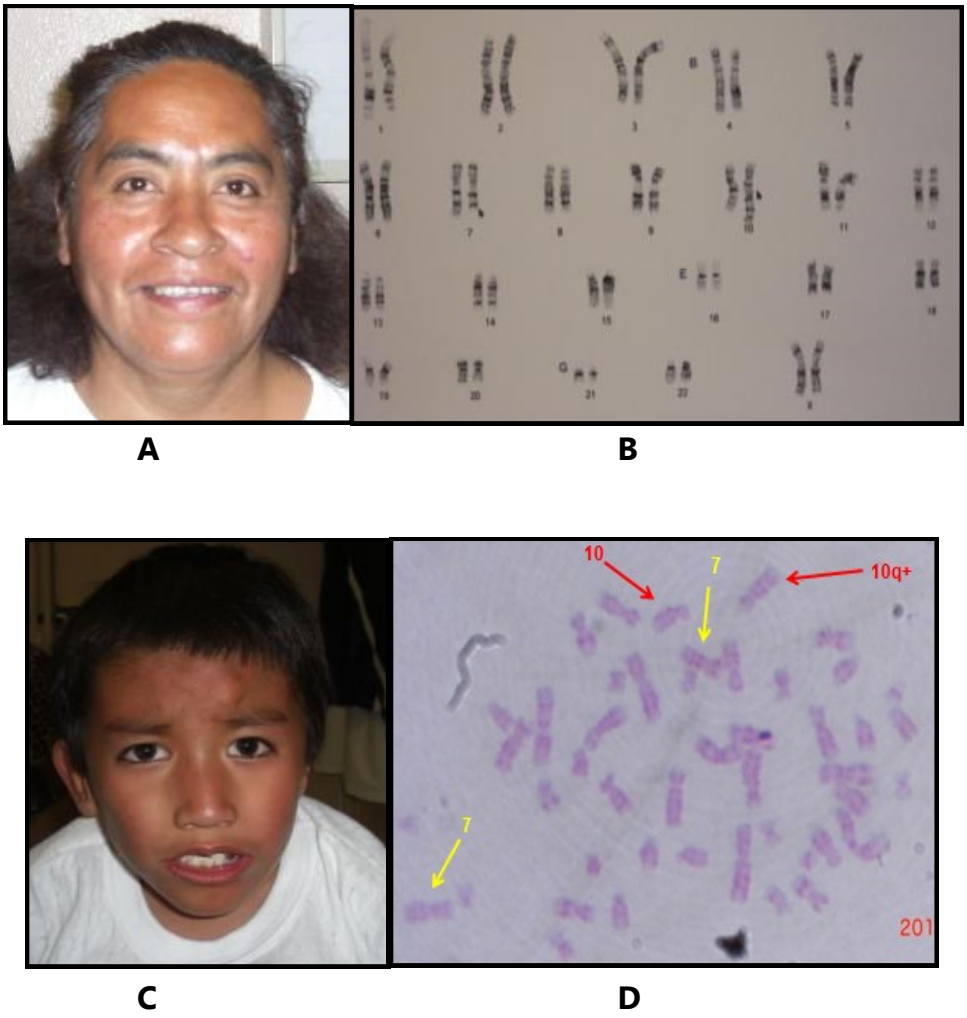



**Figure 3 A & B.** Healthy mother carrier of a 46XX, ins (10; 7) chromosome translocation (q21; q23q35) **C.** However his son with mental retardation **D.** who revealed a chromosome translocation balanced of the long arm of chromosome 10 t(10q+), causing a partial Trisomy 7.

The association between phenotypic alterations and chromosome translocation was analyzed. In cases where there are more carriers in the family, (perhaps the robertsonian), the more likely it is that the Association is not at random. As in the case of reported family where the translocation is secondary to the healthy carrier mother. Phenotypic alterations could be related to chromosome rearrangement.

**Translocation 1;4.** The present clinical case aims to present concepts basic to pediatricians, nutritionists and odontoestomatologic general practice and postgraduate and General so they can expand their knowledge in the field of oral pathology, its relationship with genetics, injuries that affect the jaw and may in addition be applied these same to your daily clinical activities. I was to present a clinical case of an adolescent patient who was diagnosed as familiar giant cementoma **figure 1 A, B, C and D.** Aparicio et al., 2002; Young et al., 1989. In relation to the definition and classification of lesions benign Fibro-osseous (LFOB), of the jaw, these, include a few groups of diverse, interesting and complex disorders, which hinders diagnosis and treatment. The common process among them is the replacement of the normal bone by a tissue composed of fibroblasts and Collagen fibers, which contains variable amounts of bone and/or a calcified material reminiscent of the cementum

Waldron C.A., 1985; Neville W., 2002. Over the past years, several researchers in the field have tried to classify the LFOB. Some of them have included injuries that they originate from the periodontal ligament Hamner et al., 1968**,** or the Medullar portion of bone; others have considered that contain giant cells and lesions which do not contain them (pure LFO) Brannon et al., 2001**.** Within these classifications, there is the proposal of Waldron C.A., 1993, which has been one of the most useful and well-known; However, Brannon et al., 2001. In relation to the cementoma multiple (CGnF) is a tumor process benign fibroseo, affecting both maxillaries. Lesions fibroseas (LFO), as mentioned above, have in common that from the hystopathologic point of view there is replacement of the normal bone architecture by a tissue composed of fibroblasts and Collagen fibers that contain varying amounts of mineralized substance, which can be similar to cement bone appearance. The LFO include a diverse group of entities, which have difficulties in classification and management. One of the most recognized classifications is the proposal by Waldron C.A., 1993, which allows us to have a general idea of the entities that make up the Group:

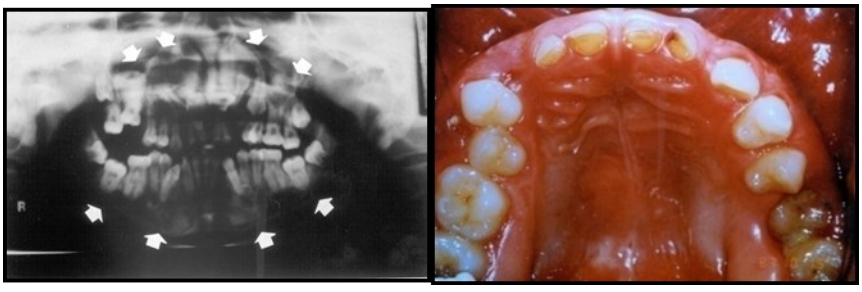
I. Fibro-osseous dysplasia;

II. Reactive lesions (Dysplastic), originating in the area that holds the teeth and is divided into three types: a). periapical dysplasia cement bone b) focal dysplasia cement florida bone c) Dysplasia of bone cement.

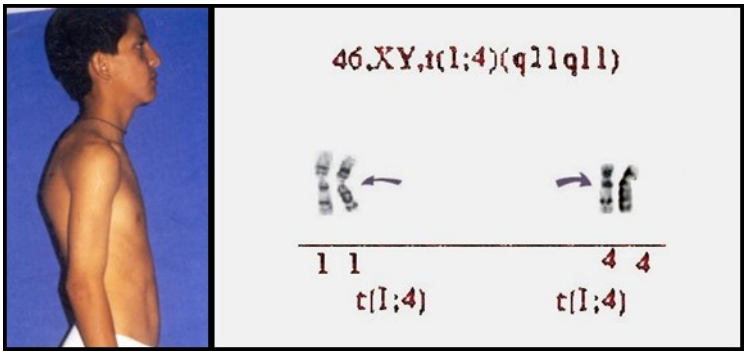
III. Neoplasms Fibro-osseous, designated as fibroma cemento-ossifying, or ossifying cement, to this group belong the CGnF.

El CGnF It may occur sporadically, but has also been reported a family variety, although the genetic transmission of the disease has not been fully confirmed; very few cases of CGnF with inheritance history have been published Kyosti et al., 1991 where an affected family, with an autosomal dominant inheritance pattern. Young in 1989 also reported a family with members affected in five generations with inheritance autosomal dominant, complete penetrance and variable phenotypic expression**.** In general, an autosomal dominant pattern has been found in cases of affected families and arises mainly related to multifocal lesions in all four quadrants of the maxilla Stevan et al., 1989. Far as we know, there is still jaw LFO, associated cytogenetic alteration, report or has been located a gene involved with this disease.

Another objective of this study is to present the case of a male adolescent, **Figure 1 A and B** changes clinical, radiological and histopathological in both compatible with CGnF jaws and report for the first time its association with a balanced reciprocal translocation 46,XY, t(1;4) (q21; q13), which we consider important for the professional in both undergraduate and graduate dentistry so it could be associated the direct relationship that exists of the oral histopathology with genetics.



**A** **B**




**C** **D**

**Figure 1 A, B & C.** Male patient diagnosed histologically as Cementoma Gigantiforme. **D.** karyotype shows a balanced translocation 46, XY, t(1;4) (q11q11).

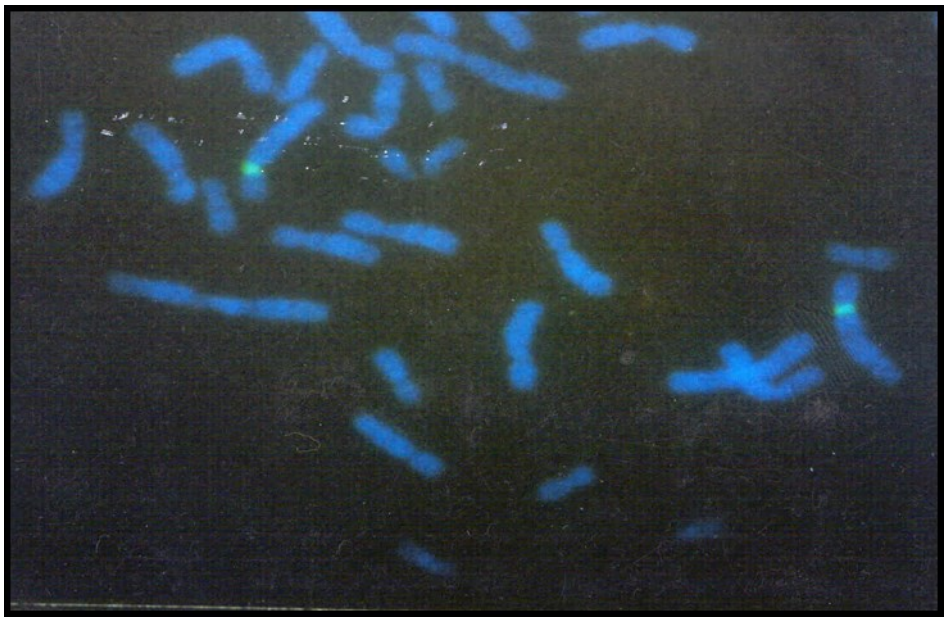
The patient presented convulsive crisis at three years of age treated with multiple anticonvulsants for one year. Referred to prolonged retention of deciduous teeth, malnutrition, infections, occasional oral breathing by nasal obstruction. Without a history of drug addiction or positive data from family congenital malformations. It was studied with bone fibro lesions compatible with fibroma cemento and minor dismorfias.

On physical examination flat facies, broad forehead, antimongoloides palpebral commissures, Dysplastic, flattened nasal bridge, elongated nose, oral commissure with deviation **figure 1 C,** normal nasal mucosa. Cardiopulmonary, abdomen and extremities without pathology. Valuation was conducted for Neurology services and Mental health, intelligence tests (WISCR and RAVEN progressive matrices). applied without reporting changes

Dental examination **figure 1 B** small **t**eeth, in upper incisive region, hypertrophic gums, well hydrated oral mucosa with moderate gingivitis in the former sectors, normal soft palate; the hard palate presents palatal gingiva and the tuberosities hard consistency, oval-shaped volume gains. It presents permanent dentition, with retention of temporary upper central incisors, which show marked attrition (functional wear).

Panoramic x-ray of the jaw **figure 1 A** shows permanent dentition, with temporary upper central incisors. At the level of canines and premolars jaw on both sides, circumscribed, Radiopaque, lesions with well-defined sclerotic borders, causing the divergence of the associated tooth roots. In the maxilla, on both sides, from the area of molars and continuing towards the anterior, in a close relationship of periodontal, presence of multiple masses lobed radiopaque appearance, well defined, some of which occupy the maxillary sinuses. Another important radiographic finding is the presence in both jaws of multiple impacted teeth, in association with radiopaque mass. The chest x-ray is reported to be normal.

Cytogenetic study was conducted in lymphocytes of peripheral blood of the patient, mother and brother; chromosomes in metaphase with the conventional technique were obtained and analyzed with GTG bands **figure 1 D.** To identify the involved centromeres became the technique of in situ hybridization fluorescent (FISH) with alpha DNA probes satellite to the centromeres of the chromosome 1 and 4 (Vysis, USA) in green color **figure 2 A.**



**A**

**Figure 2 A.** Hybridization in situ with fluorescent (FISH) with alpha DNA probes satellite for the centromeres of the chromosome 1 and 4 (Vysis, USA) in green.

It is important for the professional in health to analyze this important finding in this particular case since the final diagnosis of CGnF held by the radiological findings, histopathological and clinical. Radiologically, the patient presented features radiopaque lobulated lesions, with a component of dense non-cellular cement that can be associated with a simple bone cyst as the case that is presented in this study figure 1. The histopathological study showed a benign lesion fibro bone that affects both maxillaries and presented multiple formations of hard tissue (cementum) of acellular type. Cortical bone is represented by a thin layer of mature bone trabeculae, Compact type fibro cemetery. In the CGnF cement-like calcifications often merge to form Coalescent masses as seen in this patient, that supported our diagnostic Harrison DF., 1984. While inheritance in the family history of the patient, mother, father and brother are negative, in this case a pattern of multifocal lesions is located in the maxilla which has been associated with the familial cases related to monogenic inheritance**.**

Cytogenetic bands GTG in the patient study resulted in 46, XY, t(1;4) (q21; q13) **Figure 1 D**; This chromosome translocation was balanced and possibly originated de novo, since mother and brother karyotypes were normal, however, the karyotype in the father was not possible to do so. However, the brother of the patient was normal and has been reported that 80% of structural chromosome alterations are originated de Novo Shelby et al., 1993. The study of FISH showed the adequate presence of centromeres in the normal chromosomes and derivatives 1 y 4 **figure 1 A.**

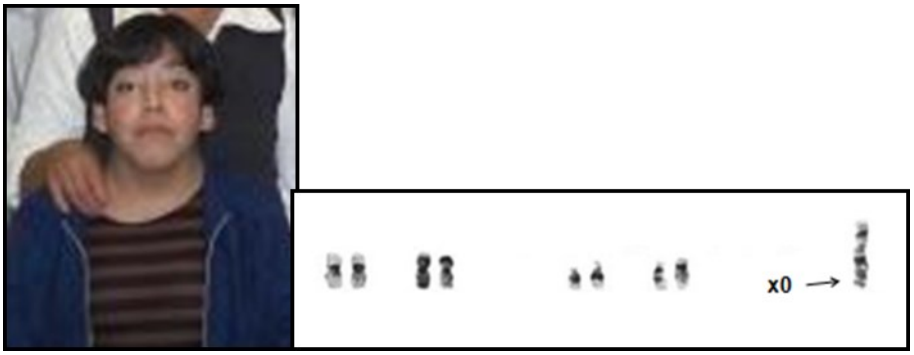
Carriers of balanced translocations individuals, usually do not have phenotypic alterations, so in those cases with clinical alterations the association between two events may be coincidence, but there is also the possibility that in some cases, the clinical picture is result of chromosome rearrangement since subcromosomic translocation may not be balanced either to cracks caused the separation of functional DNA segment and became inactive. These latter cases are extremely valuable, since thanks to them it is possible to do the physical location of the gene that causes the clinical alteration, as it has happened with diseases such as muscular dystrophy Duchenne, Neurofibromatosis, and many others Strachan and Read, 1999; Lai et al., 2000.

The patient with gigantiform cementoma, was found associated for the first time in the literature with a balanced chromosome translocation and it is possible to have relationship with the CGnF, if so, he or the genes involved may be located on any of the chromosomes 1 or 4. Given that the heterochromatin region is involved in the arrangement a possible

mechanism so that there is an abnormal phenotype due to a gene (or their regulatory regions) located in one of the two involved points of rupture, remaining fragmented in both chromosomes and therefore lost its function. It is known that there are clinical cases with balanced translocations and abnormal phenotype, also there are many examples especially in cancer cases of balanced translocations leading to inactivation, Marschalek et al., 1995; Gobbi et al., 1997; Rambaldi A et al., 1996**.**

More evidence is required to affirm that these events are related, however in many cases the first find of a chromosome rearrangement associated with a definite pathology, has allowed stomatologists to see some craniofaciales alterations may be related to genetic syndromes or well -defined chromosome aberrations. Is therefore important multidisciplinary work between different institutions which enrich research and supports one diagnosis of more complete.

**Sexuals Chromosomes : Turner (45X0).** The Turner syndrome also known as: Bonnevie Ullrich syndrome; gonadal dysgenesis; monosomy X is a genetic disorder that occurs only in women. The cells of women typically have two X chromosomes, but only Turner syndrome have a chromosome X or part of an X chromosome. This causes different clinical features or signs, as it is low and the lack of development of ovaries and infertility consistent size. Taking into account the causes, incidence and risk factors, humans have 46 chromosomes containing all the genes and DNA as mentioned in previous studies. Two of these chromosomes, the sex chromosomes, determine the gender of a person. These female sex chromosomes are chromosomes X (XX), the opposite of the male X and Y (XY). These sex chromosomes helps the person develop fertility and sexual characteristics of their gender. In Turner syndrome, the girl does not have the normal pair of two complete X chromosomes. It is more common that the female patient has only one X chromosome in their cells **figure 1 B.** Some affected patients by this syndrome have two X chromosomes, but one of them is incomplete. In other cases, there is the process of the cellular mosaisism where some cells are normal, however others have only one X chromosome. Turner syndrome occurs in nearly 1 per every 2,000 newborns, giving very specific clinical features as you can see in **figure 1 A.**




**A** **B**

**Figure 1 A.** Patient with Turner syndrome, short stature and pterigium coli **B.** The karyotype reveals a chromosome X (45XO).

Simpthomatology in female patients with turner syndrome affects; size, with some degree of malnutrition, wide neck, blepharophimosis, flat chest, broad shaped"shield", delay in the development of the mammary glands and sparse pubic hair. Infertility, amenorrhea, lack of normal moisture in the vagina. Taking into account the exams and tests, it can be diagnosed at the time of the birth or during childhood, puberty or adulthood and also it can be diagnosed by amniocentesis before birth performing a karyotype as part of a prenatal exam. Infants with Turner Syndrome often have swelling of feet and hands (Lymphedema), which probably comes from changes in the drainage of the lymphatic system, since sometimes the pediatrician or neonatologist may think of a renal alteration.

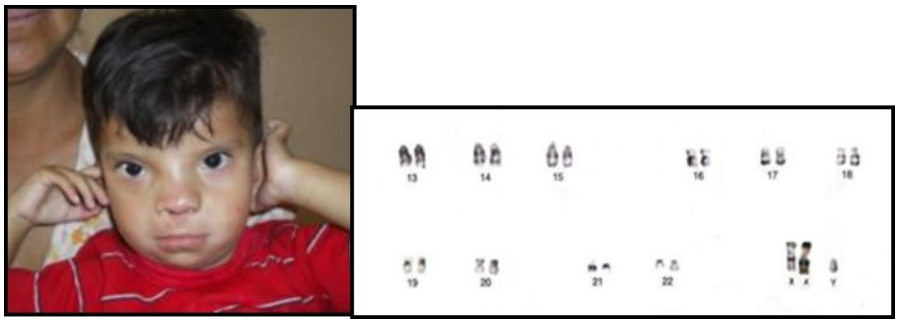
Tests that are usually sent by any physician in the area of health including the areas of pediatrics or nutrition: a karyotype for observing chromosomes, ultrasound to detect small or underdeveloped female reproductive organs. A kidney ultrasound to evaluate kidney abnormalities, in conjunction with Endocrinology may submit serum luteinizing hormone levels that can be elevated to levels of serum follicle-stimulating hormone. Often an echocardiogram (ultrasound of the heart) and an MRI of the breast after the diagnosis, is performed to evaluate any defects in the heart, since the most common problem is an alteration in aortic (stenosis, coarctation, etc), which is important. According to the management or treatment of these patients, hormone somatotropin or growth which must be administered by endocrinology, to help a girl with Turner syndrome not only increase his stature, but also as support in his cell development by

better metabolic processes and oxygenation, can be considered It will thus support the professional in nutrition for better nutrition and development. Estrogen therapy is usually initiated at 12 or 13 years of age to stimulate the development of breasts, pubic hair and other sexual characteristics. Egg donation programs are available to help women affected by this syndrome who want to become pregnant. There are support groups for these patients; www. turnersyndrome.org

The expectations of patients with this syndrome of Turner may have a normal life span and a normal productive life provided a careful medical control occurs. As mentioned above, if necessary a performing any stomatologic procedures, must be performed in multidisciplinary form with nutrition, endocrinology, cardiology, offering better handling and a more favourable for the patient**.**

**Poliploidias; Klinefelter (49 XXXXY).**

Klinefelter's syndrome is a condition that occurs in male patients as a result of the presence of an extra X chromosome and whose most common symptom is high size, change in the distribution of body fat and infertility. Taking into account the causes, incidence and risk factors, males have one X chromosome and one and which is written as XY. The two sex chromosomes help a person develop fertility and sexual gender characteristics, as mentioned previously. In Klinefelter Syndrome, men have one or more of one X-extra (written as XXY **figure 2 B**).



**A** **B**

**Figure 2 A.** Male patient diagnosed as syndrome of Opitz G/B.B.B. hypertelorism, lip and palate cleft with **B.** craniofacial dimorphism The karyotype reveals a doubling of sex chromosome X (Klinefelter's syndrome).

In this case specific note that the patient presents two genetic alterations, a syndrome of Opitz G / B.B.B. and perform a chromosome study, it was observed a doubling of their sexual crosmomas X (47 XXY) **Figure 2 B,** and you can see that the patient presents as part of its clinical features, cleft lip and palate (LPH) bilateral and hypertelorism. It is important to mention that although LPH is known in genetics to be non-hereditary, multifactorial entity 90% about could be valued at LPH clinic in the Dentistry area, like a line defect only and treated as such.

This should be well thought for the professionals in dentistry, nutrition, pediatrics and genetics that a patient with LPH should be investigated through a good clinical history, anamnesis and laboratory studies before considering a single diagnosis in the patient, such as serious in this case, where the patient presents LPH, however is part of a syndrome (Klinefelter). This syndrome occurs in one of every 500 to 1000 newborn males. Women with pregnancies after age 35 are likely to slightly greater than having a child with this syndrome.

Within the most commonly observed signs are as follows: small penis, testicles small and firm, little facial, axillary and pubic hair, sexual dysfunction, enlarged breast tissue (called gynecomastia) and carving high abnormal body ratio (long legs, short trunk). Adults can go to the doctor because of infertility, and school-age children, psychomotor retardation in some of them, who should be treated as such to assess whether they should be handled in the area of dentistry under general anesthesia supported by the area of nutrition. Of the tests that were carried out to patients, the following results can be found: karyotyping that shows 47 XXY, count low semen, low serum testosterone level, level of hormone serum luteinizing and follicle serum estradiol (a type of estrogen) high.

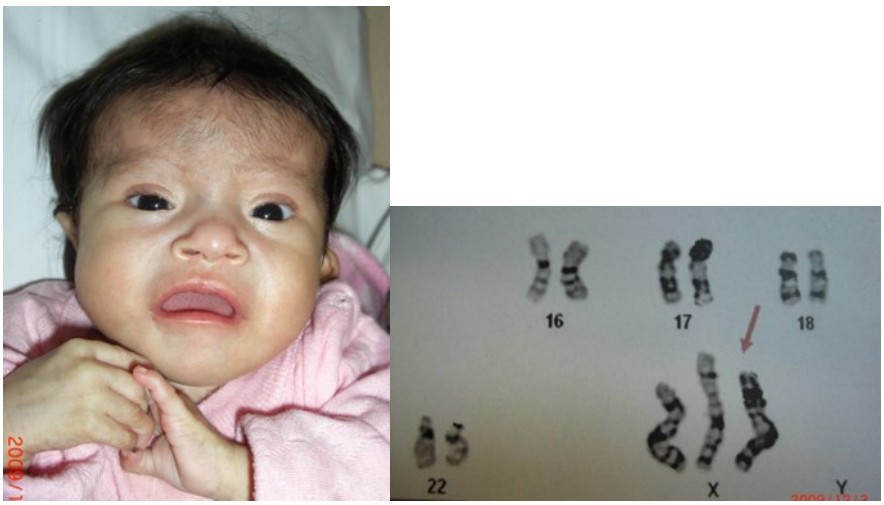
The treatment will consist of therapy with testosterone, which will result: improve the strength, the appearance of muscles, self-esteem and mood, energy and sex drive. Most of the patients are infertile. However, there are some cases of men with an extra X chromosome having offspring. Within support groups, there is the *Klinefelter Syndrome Association* a la casilla postal 119, Roseville, CA, 956780119.

**XXX Syndrome.** This syndrome, Anónimo. Síndrome de Triple X. 2009; Nielsen J., 2009**,** is a genetic disorder that is observed in women that have an extra x-chromosome 48XXX **Figure 3 B.** This chromosome is obtained during the sperm or egg formation that later joined to form the fetus. This extra chromosome may not be disposed of in formation. The probability

that develops this abnormality is approximately 1 of every 1,500 girls.

Clinical features presenting the newly girls with 47, XXX syndrome look like other girls of her age. Some are much higher than the rest of the girls in his family and may have less coordination and therefore with bad eating habits a secondary malnutrition. With 47, XXX syndrome women are most fertile.

This syndrome is one of those associated with mental problems and behavior. A high probability language and speech problems and can cause delays in social skills and learning. Craniofacial dimorphism, palate is ogival and sometimes crowding of dental organs. Small eyes with hypotelorism, turricefalia and hipoplasia **figure 3 A.**




**A** **B**

**Figure 3 A.** Female patient with small eyes, Craniofacial dimorphism, hypotelorism, turricefalia andhypoplasia **B.** The karyotype reveals a Trisomy of sex chromosomes (47, XXX) X.

Triple X syndrome, is generally a sexual Trisomy: XXX sex chromosome. It only affects female patients. Characteristic features: are clinically normal, has been observed craniofacial alteration, sterility and mental retardation. They have a slow motor development, linguistic and emotional. When submitting a linguistic dementia, it is important to visit to the speech therapist, and in terms of psychomotor development is important to practice sports, creative activities of accompanied by proper nourishing

management group. The independence of these girls must be encouraged. They tend to be immature, this can be improved with the collaboration in its development during childhood by their environment. IQ average for triple X girls is slightly lower than the average with a high degree of malnutrition generally.

These patients growth faster than other girls until the age of 8 years. They usually have long legs compared to their body length and height as well as some degree of malnutrition. Usually have a low weight in relation to height. The causes of this syndrome are unknown, although in some cases it is associated with the mother age. The extra chromosome can proceed from the father or the mother, although it is more likely that it comes from the mother.

**Nutrimental management on patients with Chromosome Aberrations.**

Nutrition in patients with different chromosome alterations referred to in this study is very important and complex since it will depend on the clinical and systemic manifestations to each syndrome according to their own medical needs.

That is why it was decided in this study to mentioned the food management of Down syndrome patients, taking into account that the incidence is higher in comparison with all chromosome alterations. Nutritional therapy is included in the health monitoring of children with Down syndrome. It is necessary to choose an appropriate anthropometric standard. During the monitoring of health and follow-up consultation is necessary to guide eating habits and healthy physical activity and boost their income to early stimulation systems. The nutrition plan must be placed in the patient's current health problems; for example, many patients need to achieve an adequate weight to deal adequately with cardiac surgery. It is important to properly define the energy and protein intake, and the only way to assess it is through the growth pattern of the patient. There are many alternatives of nutrition, such as breast milk, formula milk and meals, when they are appropriate introduced. Power paths can be oral or use a stomach tube and even perform a gastrostomy if necessary.

In an Italian study mothers of children with Down syndrome attended at University hospitals were interviewed, to determine the status of breastfeeding, in a universe of 560 children, of whom 246 were still in the neonatal unit. It was found that 70% of children who had to be hospitalized and 46% of those who are not hospitalized, they did not receive breast

milk; and the duration of breastfeeding was 54 days (deviation are give ± 111 days), especially in children who are hospitalized in neonatology. Thus, 57% of children with Down Syndrome did not receive breast milk. Frequent child diseases; depression and frustration felt by mothers for having a child with Down syndrome; the fear of having insufficient milk production; and children weak suction, were the reasons identified by the mothers of these children.

**Antropometry and physical composition**

There is little information on the body composition of patients with Down's syndrome, but it is known that they are characterized by:

1) low size, with average height between 1.45 and 1.50 m in adulthood, i.e. very below the normal average height.

2) early pubertal development, which begins to 9.5 years in girls and at age 11 in children; and

3) very common in adolescence and adulthood overweight and obesity.

Nutritional evaluation of these patients was carried out according to the anthropometric standards used in the healthy population, and diagnosed as malnutrition and low size. However, in the same way for many morbid pictures, own growth standards were developed in various countries such as Spain, United States, Sweden, United Kingdom, Ireland, the Netherlands and Germany, Cremers et al., 1996.

Each table is designed with different methodologies and the choice of the most appropriate, is the one who can guide the expression of the maximum development potential of these children.

The tables that are most used are the Foundation Catalan Down's syndrome, which has two versions (1998 and 2004) and the tables of Cronk et al., 1988, which considers the United States population, and published in 1978 and 1988. Swedish growth curves were obtained from a longitudinal and transversal study with 4.832 measurements in 354 patients, of whom 151 were women and excluding patients who were using growth hormones.

These data were made only by graphics that were expressed in standard deviations weight for age, height-for-age, cranial perimeter for age and BMI (BMI) for the age. The Swedish table bmi chart.

In the study conducted in the United Kingdom and Ireland 5,913 measurements were made in 1089 patients, excluding all patients with heart disease, premature or those who die during follow-up.

With Down syndrome in a pediatric hospital. It is likely to be one of the most demanding tables. Also available in percentiles and the parameters considered are: weight, length and perimeter of the skull for age, Myrelid et al., 2002; Pastor et al., 1998.

The growth charts of Spanish with Down syndrome children, developed by the Catalana Fundación of Síndrome de Down, date back to 1998 and are included in the book "Standards anthropometric for evaluation of the nutritional status", Gladys Barrera, published in the years 2004 and 2006 by INTA, mentioned by Mario Bildoso in 2006. These tables begin at 2 months of life, so it is very difficult to extrapolate information to determine the nutritional status of a child before that age Piro et al., 1990; Mario Bildoso, 2006**.** In 2004, the Foundation developed tables with percentiles, allowing you to accurately determine the status of the child. For the year 2004 growth curves were 1.718 measurements, 763 of them in women and excluded patients with diseases that affect growth, such as heart disease, hypothyroidism, etc.

**Nutrimental problems:** Early childhood have been observed with feeding problems and poor gain weight in children with Down syndrome, mainly in those who suffer from severe congenital heart disease. These children may eat poorly and does not develop. However, as soon as heart disease is corrected, they begin to gain the right weight.

Many teens and older people with Down syndrome have obesity. This is probably due to a lack of physical activity and familial bad habits. However, some young people with Down syndrome have increased weight while maintaining a normal caloric diet. It is important that, from childhood, children maintain a proper diet to avoid excess weight. The best for all children, including those who have Down's syndrome, is to maintain proper food habits, balanced diet, avoid high-calorie foods and perform regular physical activity.

## CONCLUSIONS

To design a nutritional plan, in a patient with Down syndrome, is:

To evaluate the current health problems, including nutritional status of the patient, compared to the more appropriate anthropometric standard; establish the most adequate energy and protein contribution; and determine the types of food and the way of power that will be used.

These children should receive the same supplements than healthy children. It should be additionally encouraged the incorporation of infants to early learning programs, as well as the creation of healthy lifestyle habits in relation to food and physical activity, in order to prevent chronic diseases of adulthood and find and treat in properly and early associated pathologies.

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