UNIDAD EDUCATIVA PARTICULAR JAVIER

BACHILLERATO EN CIENCIAS

MONOGRAPH

CONGENITAL MALFORMATION IN THE NEWBORN

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GRATITUDE

I want to thank all the people who helped me make this monograph, thank you, Dad, for having given me the idea to make this subject and contribute with books and information with what you see daily in your work. To my teacher and tutor who kept giving me suggestions until the end, and to my family: mom, sister, and brother, who waited a long time for the laptop whenever I was doing my monograph.

SUMMARY

The work of this monograph is titled Congenital Malformations in the Newborn, and here we will explain the following types of malformations, with their respective causes and treatments. The first chapter defines it as a congenital malformation, more like they are divided. Then, in the next chapter, you will find descriptions of certain malformations that are normally found in Guayaquil, Ecuador. After the descriptions, an explanation of the corresponding treatments will be made.

INDEX

INTRODUCTION

On the topic: Congenital malformations in the newborn, the author wants to focus on a problem that occurs naturally around the world, which greatly affects the lives of infants. That is why this research work seeks to find out how congenital malformations develop and what the causes are. It is very important to know how these abnormalities appear and perhaps learn how to prevent them in the future.

The chosen topic is a situation that affects the child's life without any understanding reason and at the moment we do not know how to correct them once they are in it. That is why with this monographic work, it is a question of giving and explaining each of the factors, demonstrating how each one of them can be formed, and as it is stated in the previous paragraph, to help perhaps not to stop them, but to prevent them.

It has been analyzed that, over the course of a year, 8 million children are born with congenital malformations, of which 3.3 million of those under 5 years of

age die due to having one of them. These are certain details that have been found in both books and medical journals.

It is not certain that this topic has been chosen before in an Ecuadorian monograph, but I wanted to focus this topic on professional people who see this on a daily basis and can give their different points of view. The doctors will be interviewed in a hospital of the child Francisco de Icaza Bustamante Guayaquil, Ecuador.

In the first chapter, we try to explain that it is a congenital malformation and it is divided into what are the causes of them generally, how they are divided and their types. In the second chapter, he mentions the stages of the embryo where the anomaly is formed and following that he names the syndromes that are formed from the embryo, he talks about what each one of the malformations mentioned is, for example, Langdon Down Syndrome, he describes what it is and what are its causes or environmental factors. Chapter three mentions the treatments mentioned in the

previous chapter. At the end, we have chapter four, which is where the opinions of professionals are analyzed in the form of an interview and graphics.

CHAPTER I

CONGENITAL MALFORMATIONS

1.1. Definition of Congenital Malformations

Congenital malformations can be defined in different ways, as well as disorders or anatomical abnormalities which occur at birth and affect the functioning of the organs depending on the stage of formation of this. Are found both external/physically or within the organism, we mean, they are both macroscopic or

microscopic. Teratology is the branch of embryology that refers to the abnormal development and causes of congenital malformations.

About 20 percent of deaths in the perinatal period are attributed to congenital malformations (MacVicar, 1976). Congenital malformations can be observed in approximately 2.7% of the neonates (McKeown, 1976) and during lactation, 3 additional% are discovered.

Congenital malformations are classified into two big groups, genetic factors, and environmental factors. But, even when this proposed classification it is in use, it doesn't mean that we can exactly determine the factors that caused the malformation.

The author Keith L. Moore in his book ''Clinical Embryology'' (S.F), explains: " In recent years many advances have been made in the search for causes of congenital malformations, but an adequate explanation is lacking for most of them ". What this phrase means is that not all of the abnormalities have an explanation.

1.2. TYPES OF MALFORMATIONS

There are many connatural abnormalities that can affect the human form; the most common ones will be remarked below.

Malformations of the nervous system:

a) Anencephaly: This malformation refers when the baby is born without some parts of encephalon and skull. It is developed when the neural tube does not close completely; it usually occurs if the neonate is born without the upper part of the brain.

Causes or risk factors: An anencephaly can be caused by a change or a combination of genes. Other factors may be the environment in which the mother comes into contact or the foods or certain medications that she was able to digest during pregnancy.

b) Spina bifida: It is a congenital anomaly that occurs when the spine and the spinal cord do not form well. It is classified as a neural tube defect. Spina bifida can be balanced between mild and severe, depending on the type of defect, size, location, and complications.

Causes or risk factors: It can be a result of family history, a risk factor can be obesity, diabetes, medications (anticonvulsants) and folate deficiency.

Malformations of the circulatory system:

a) Eisenmenger Syndrome: Heart defect at birth caused by increased pressure in the blood flow of the lung. As consequence, not enough blood is received in all organs and tissues.

Causes: It is caused by a hole between the cavities of the heart.

b) Tetralogy of Fallot: A condition caused by a combination of four heart defects present at the time of birth. Normally children suffering from this syndrome have bluish skin because blood does not carry enough oxygen.

Causes or factors: There are some factors such as maternal malnutrition, viral disease or genetic disorders.

Malformations of the respiratory system:

a) Pulmonary hypoplasia: Condition where the lungs of a neonate have not been able to maturate correctly during the development in the matrix. This produces that the alveoli, which are very necessary for the exchange of oxygen dioxide and carbon in the lung, are diminished in great quantity, and that damages the capacity of the baby to breathe.

Causes or factors: The most common causes of hypoplasia are the low quantity of amniotic fluid. When the fluid is diminished around the newborn, the movement of the chest begins to be reserved.

b) Atresia Coanas: It prevents the development of the natural formation of the nasal cavities, this atresia is produced by the persistence of the membrane, it also prevents the posterior fragmentation of the duct, making impossible the nasal opening or choana.

Causes or factors: According to research this disorder is due to an imperfection in the thin tissue that diverts the area of the nose and mouth including other assumptions.

1.3.CAUSES OF THE MALFORMATIONS

Not all congenital malformations have an exact explanation, but along the years many of them have been discovered. These factors can produce or be one of the causes that can produce anatomical destruction in a newborn. Next, we will analyze the main ones.

Up to 40% of abnormalities are annexed to certain risk factors that act during the development of the embryo:

• Genetic or chromosomal disorders: In this case, it is indicated by the genetic information of the neonate; however, there will be cases of spontaneous alternation.

• Drugs: Certain medicines can harm the good development of the fetus; a good behavior is that pregnant woman always consults her doctor before taking any pill, or another type of medication.

• Infections: If the mother of the child has some type of infection, it is very possible that the child will get quite injured,

• Food: It is very important that the woman had a balance in their meals during the pregnancy period because if there is any nutritional deficiency, the baby should have several consequences.

• Maternal age: If the pregnant woman is under 20 years or older than 39, the development of the baby can be affected in a negative way producing this type of abnormalities.

CHAPTER II

THE EMBRYO

2.1 THE DEVELOPMENT STAGES OF THE EMBRYO

The formation of the embryo is the most important period of human development, because during it the beginning of all major external and internal structures can be seen. When the embryo is in this stage, any exposure to teratogenic agents can cause quite serious congenital malformations.

A message from author Keith L. Moore in his book "Clinical Embryology" (SF): Although the name "embryo" is often used for parts less than 4 weeks old, theoretically this term should not be used during the first three weeks when the embryonic membranes and germ layers are forming. However, in practice, this word is commonly used from when the embryonic disc is identified until the end of the eighth week. Then, until birth, the product is called "fetus.

2.2DESCRIPTION OF SYNDROMES

We will now describe the syndromes that we have found in the hereditary part

- Crouzon syndrome

What is it?

Crouzon syndrome is an irregularity of genetic origin characterized by the presence of craniosynostosis or suturing of the bones of the premature skull. These disorders can cause complications for the proper development of the brain and all the organs in the head.

The closure causes the skull to grow in the direction of the junction to remain open, which develops malformations in the appearance of the skull and face. It is also likely to affect the relationship between the teeth.

Why is this happening?

One of the reasons this malformation occurs is because of a mutation in the fibroblast growth factor receptor genes. It can be considered an autosomal dominant hereditary disease.

This syndrome can be inherited from both parents, such as a new genetic mutation. The result is a mutation of a gene involved in the formation of bones, skin and connective tissues.

In most cases, the sperm transmits these mutations, and it is related to elderly parents.

To prevent excess pressure on the brain, the baby may have surgery in the first few months. Surgery may be necessary for cosmetic reasons or to solve problems such as progressive vision loss. (Bethlehem,2009)

- Apert Syndrome

What is it?

Apert syndrome is a rare genetic condition in which the joints of the skull of a newborn or infant close prematurely. This is also known as craniosynostosis.

The fibrous joints of the skull are usually kept open to allow the brain to grow. Because these structures close very quickly as the wax continues to grow, it pushes the head and face in a distorted way, which can also affect the baby's pressure.

A characteristic of this is the difference with Crouzon syndrome, discussed above, is that this symptom is accompanied by congenital fusion in the fingers and toes.

What are the causes?

Each one of these alterations is of genetic origin, anatomically dominant or also by "novo" mutations, which today is one of the main causes of this syndrome.

The causes of this alteration are not known, but environmental factors such as these are generally considered:

1. First of all, the chemical effects, which are produced by drugs and pharmaceuticals.

2.Nutritional deficiencies, which is a lack of necessary food for the body.

3.General biological factors, such as the age of the parents.

-Treacher Collins syndrome.

What is it is it?

This Treacher Collins syndrome is a rare and incurable congenital anomaly that affects two out of 100,000 births.

This syndrome has one that has always been associated with: hypoplasia of the malar bones, cleft palate, coloboma which means an absence of eyelashes, that is to say, the palate does not grow, or they have an open palate and have a narrow pharynx. Another consequence is that people with this syndrome are born without one or two ears.

What are the causes?

The origin of this syndrome is not yet known, but it is assumed that if one of the parents has or has it, the mutated genes will be passed on to their offspring. The pattern of inheritance is autosomal dominant, there is a 50% chance that the baby has this disease. But most of these cases also develop during embryonic development.

-Pierre Robin Syndrome

What is it? What is it?

It is a condition in which a baby's jaw is smaller than normal, and they also have a tongue that is pulled down in their throat and breathing discomfort.

Because it is a defect of the first-gill arch, multiple ophthalmological problems are attached that can pass unawares to the doctor before the seriousness of the systematic picture, but that can compromise the development of the newborn.

What are the causes?

The appearance of Pierre Robin syndrome may be due to the union of several genetic syndromes. And as an equivalence, the genetic changes prevent an adequate morphological development of certain structures, that is why the cardinal clinical findings are manifested: micrognathia, glossoptosis, and palate of the hay fever.

-Goldenhar Syndrome

Goldenhar syndrome is a rare, eventually occurring condition with a weak genetic component. It is characterized by the appearance of facial anomalies, especially deformities. Clinically, facial asymmetry only occurs in 70% of cases and is most noticeable during childhood.

What are the causes?

The cause of this congenital malformation is not confirmed, but several factors have been reported in the literature, which is divided into:

Environmental:

Cocaine uses during pregnancy.

Bleeding in the 14 weeks of pregnancy.

Fertility treatments.

Maternal diabetes.

Genetic or hereditary:

Inherited cases are more regular when the disease affects the sides of the face.

Unknowns:

Most of the time, the causes are unknown.

Langdon Down syndrome

The Langdon down syndrome is characterized by a genetic alteration in which the human organism has 47 chromosomes instead of 46. It is also known as Trisomy 21 since the remaining genetic material corresponds to par 21.

This disease occurs in one in 700 births and it's one of the main reasons for mental retardation in the world.

There are some physical characteristics of this syndrome, for example:

- They have a flat nose bridge.

- The eyes are shaped like an almond.

- The neck is short.

- The ears without small ears.

- His height is short in childhood and adulthood.

There are also types of Down syndrome.

Trisomy 21: This type is characterized by the fact that each cell in the body has three separate copies of chromosome 21, instead of the two that should be kept.

2.By translocation: It is characterized by having one chromosome 21, along with a different chromosome instead of being separate.

3.With mosaicism: Reverent to a combination, certain cells acquire two copies of chromosome 21. Children with this type of Down syndrome may have similar characteristics. But they have fewer of them because of some cells with the normal number of chromosomes.

What are the causes?

Experts know that this syndrome is caused by an extra chromosome, but it is not clear why it occurs.

But there is one factor that adds up to the possibility of having a baby with Langdon Down syndrome, and that is the age of the mother. When a woman is older than 35 years of age, the chance that her child will be able to go out with Down syndrome is very high.

-Klinefelter's syndrome.

Klinefelter's syndrome is not hereditary but is caused by a genetic condition that only harms the male gender. There are times when children do not realize they have this syndrome until they are adults.

The man born with this syndrome may suffer from low testosterone levels, reduced muscle mass, and poor physical and facial hair. Most of them do not produce sperm.

What are the causes?

The syndrome occurs as a result of random failure where a child is born with an extra sex chromosome. Normally we humans have 46 chromosomes and two sexes that determine the sex of a person.

In females, the chromosomes are two X or XX. And in males, you have XY chromosomes. Certain causes will be shown below.

- The duplication of the X (XXY) chromosome in each cell is one of the most common causes.

- The Klinefelter mosaic, which consists of a repetition of the X chromosome, but in some of the cells.

- This cause is not as common as the others, but it is when you have more than one repeat on the X chromosome.

-Prune belly syndrome

Prune Belly syndrome is the absence of abdominal muscles, either partial or total. Defects in the urinary tract and for children the absence of testicles as they did not develop properly.

The absence of the abdominal muscles can cause problems coughing properly, increase secretions in the lungs, and can cause constipation because of inability to perform abdominal movements to remove the stool,

When having defects in the urinary tract, there are some malformations such as:

- Obstruction in the ureters.
- Pile of urine in the ureters and kidneys.
- Reflux of urine from the bladder to the ureters.

This syndrome affects one in 40,000 infants. It's more likely to happen to men, only about 4% of them to women. It is most commonly found in African Americans.

-What are the causes?

There are two outstanding theories as to what the causes are:

1st: The plugging of the urinary tract is due to a malformation where the urethra closes at the end and by a narrowing of the valves.

2nd: The lack of abdominal muscles may be due to the exit of the cells that form the embryo, between the two months of pregnancy.

CHAPTER III

IMPROVING THE QUALITY OF LIFE

3.1. TREATMENT

-Crouzon syndrome

Treatment for Crouzon syndrome is divided into

a) Surgical treatment: Treatment varies depending on the severity of the congenital malformation. Most of the time it will be treated with surgery, to try to avoid complications that can form in the skull. If this abnormality is not treated, the patient may develop blindness, deafness or learning difficulty.

Surgical treatment is based on opening the scars. If vision loss occurs, what is done is to misunderstand the sutures and improve brain growth. It is preferable to perform this process when the patient is still small, and thus prevent there from being enough space for the brain.

There are 4 types of surgeries, which are:

- Craniotomy: This is based on the removal and replacement of portions of the cranial bone. The surgery proceeds moments after birth to avoid consequences such as Brain pressure.
- Surgery for exophthalmia: The surgery is right in the eye socket or the bones around it. This helps to decrease exophthalmia.
- Prominent lower jaw surgery: It has an effect on the normalization of the appearance of the face.
- And finally, the surgery to repair the sinus fissure.

Orthodontic-surgical treatment

a) Characteristics of the malformation: People with Crouzon syndrome have features that make it necessary to be treated early.

They meet:

Upper jaw:

- Ogival palate.
- Malar bones are depressed.
- Spasm of motor muscles.

Lower jaw:

- Mandibular prognathism also occurs in patients.

Teeth:

Defects in the teeth can be either very large or very small. Variation is the most common tonic in patients.

Tongue and lips:

It may be larger than the lower lip, which is usually separated from the shorter upper lip.

-Apert Syndrome

The treatment of Apert syndrome should be multidisciplinary because of the number of defects to be treated. A holistic approach is needed, a large number of professionals are required.

Estimation starts from the moment of birth and childhood. They are needed:

- Pediatrician
- Neurosurgeon
- Psychologist
- Craniofacial Surgeon
- Otolaryngologist
- Ophthalmologist

When it comes to the stages of adolescence and youth, more professionals are required, such as:

- Orthodontist
- Speech Therapy
- Maxillofacial surgeon
- General dentist

It is best to treat the syndrome from birth, with the correct diagnosis. The essential thing is that specialists understand the complexity of the syndrome. For all types of specialists, it is recommended to be in the same place, to minimize the number of appointments and develop operations.

From 5 years old to 9 years old, the boy will have an operation to progress with the middle third of his face, thus improving his profile. And these operations will last until the growth is over, that is until adolescence.

- Treacher Collins syndrome

In Treacher Collins syndrome, surgery is also used, which tries to correct the face and includes:

- Plastic surgery
- Orthodontics
- Dental care

In surgery we found:

- Palatoplasty: Repair the fracture in the palate.
- Gastrostomy: A tube is inserted through the abdomen into the stomach. It is used for feeding or drainage.
- Orthognathic Surgery: Corrects jaw defects, such as underbite.
- Bone graft: A bone is placed through the fracture to heal.
- Bone elongation: Bones are lengthened where two segments are separated so that the placed bone grows in the middle.

-Pierre Robins syndrome

These types of surgeries are to fix abnormalities in the skull.

Jaw lengthening

Palatal cleft closure

Bone distraction

Tongue fixation

Tracheotomy

The main objective is to implement an effective communication tactic through residual capacities and at the same time, the incitement of the transaction of new skills.

-Goldenhar Syndrome

The treatment of this syndrome is one of the most complexes, so there are many parts of the syndrome that must be considered and as some of the malformations, a group of specialists is needed as well as:

- Pediatrician
- Geneticist
- Craniofacial Surgeon

- Plastic Surgeon

- Orthopedic

- Ophthalmologists

- Orthodontist

- Speech therapists

Treatments are performed as necessary to help the infant:

- Bone is grafted to form the jaw

- Palate correction

- Finally, the cleft lip.

Always start as soon as possible to get defects corrected on time.

-Langdon Down Syndrome

Down syndrome cannot be cured, although it is said that by treating them as soon as possible people who have this syndrome from adulthood can have a productive life.

There are 3 things that can benefit children with Down syndrome, which are speech therapy, occupational therapy and exercise to improve their abilities such as hands, feet, etc..

It is necessary to have medical attention that can help in the development of its maximum potential. Specialists such as

- Audiologist
- Cardiologist (pediatric)

- Physiotherapist
- Neurologist (pediatric)
- Optician (pediatric)
- Pediatrician, primary care (childhood)
- Developmental Pediatrician

-Klinefelter Syndrome

As mentioned earlier in congenital malformations there is no specific cure, but for this treatment there is:

If a child is diagnosed with Klinefelter syndrome, it is quite possible that Klinefelter syndrome will be included:

An expert (doctor) who specializes in the treatment of disorders involving hormones and glands in the body.

- A genetic counselor.
- A specialist in reproductive medicine.

- A psychologist
- A pediatrician

Treatment for this syndrome may include:

- Testosterone Replacement Therapy: Testosterone replacement therapy should be started at the beginning of puberty, usually to promote the changes usually caused by puberty. As for example, Growth of muscular mass, an increase of facial and corporal hair, with this therapy fractures can be avoided.

- Educational evaluation and support: Normally children who have this syndrome suffer from problems in learning and social communication. Personal or additional assistance may be necessary.

- Speech Therapy and Physical Therapy: This therapy is for children with this syndrome to overcome speech problems.

- Fertility treatment: When a man has Klinefelter syndrome, it is normal that he cannot have children because the testicles do not produce sperm or produce a small amount of sperm. For men who produce a small amount of sperm, there is a useful injection: Intracytoplasmic sperm injection, which is where a quantity is extracted with the syringe and put into the ovule.

-Prune Belly syndrome

In the treatment of Prune Belly syndrome, most cases require pharmacological treatment depending on the percentage of severity of the symptoms. And to mend the malformations, surgical treatment is required.

In the surgical treatment usually includes:

- Reconstruction of the abdominal wall

- Standard pyeloplasty. That is where the narrowing in the ureter, known as "stenosis", is treated.

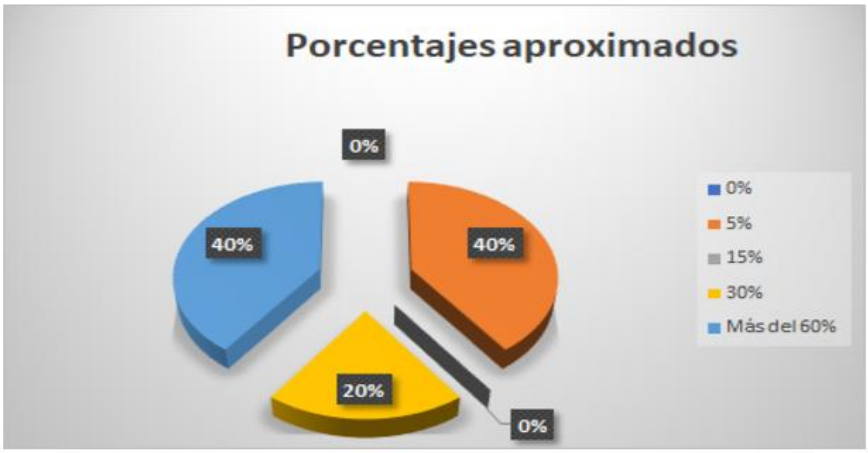
- A surgery to address obstruction in the prostatic urethra.

CHAPTER IV

INTERVIEW

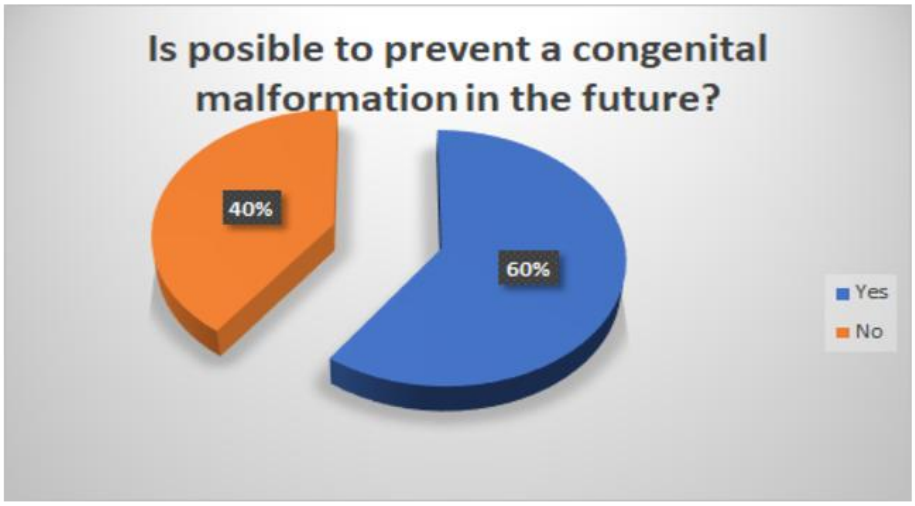
3.1. Interview applied

1.- What is the approximate percentage of children with malformations you see in your environment?



In conclusion, the most common percentages seen in hospitals are 5, 30 and more than 60%.

2.- In your opinion, can malformations be prevented in the future?



The interviewees gave their own opinion on this question. And they were asked to explain their answer.

Respondents who answered NO were asked why?

And this was the most prominent answer.

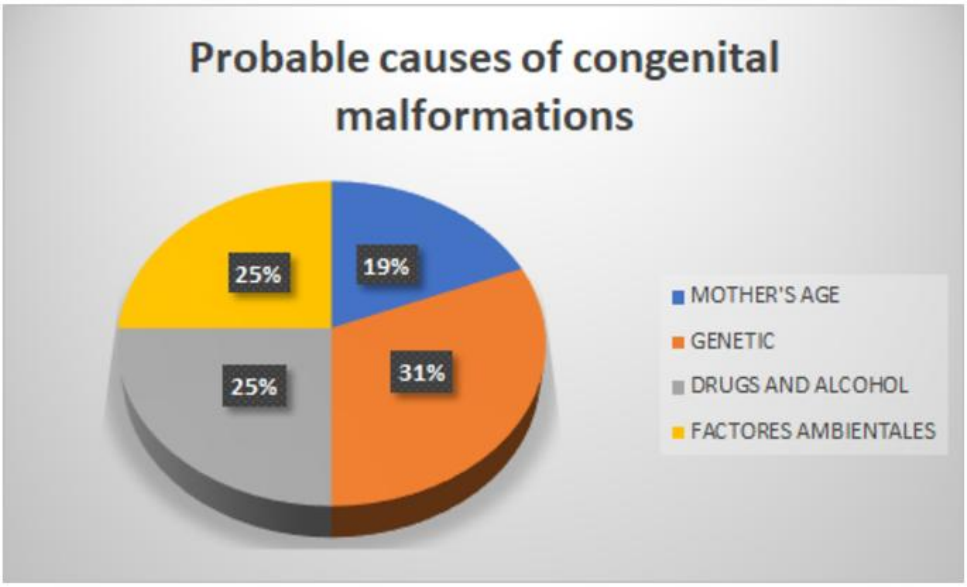
- For lack of early diagnosis. If doctors were to check their patients early, perhaps they could be prevented.

In other words, when a gynecologist or obstetrician does not provide early check-ups, it removes the possibility of preventing a congenital malformation.

Those who answered YES were asked to explain how they could be prevented? And this was the most prominent answer.

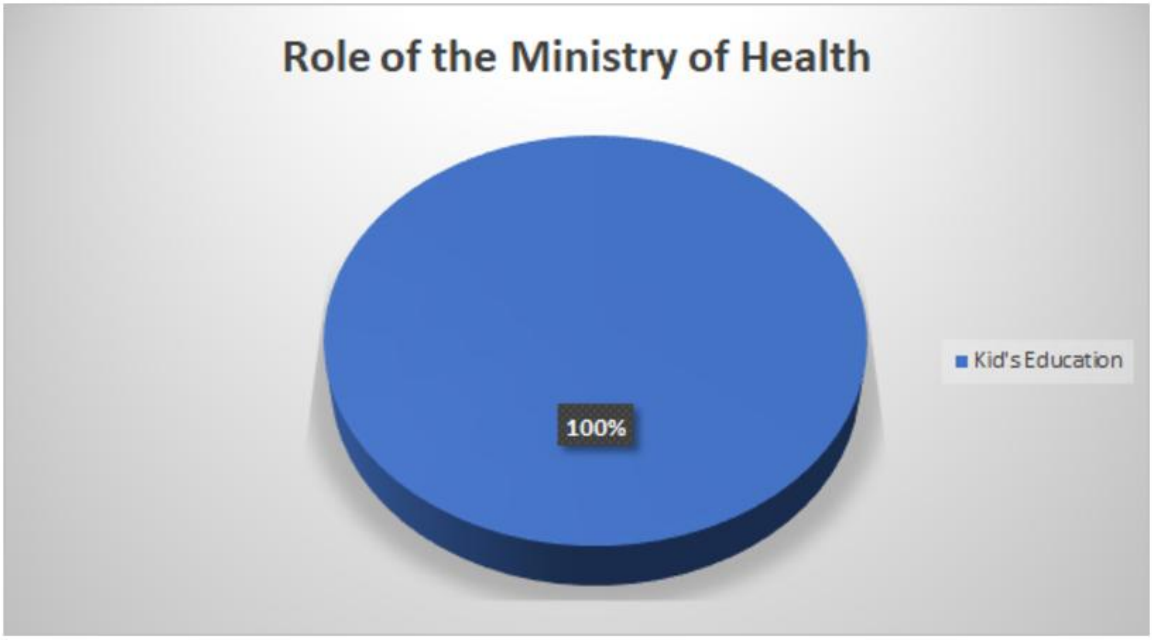
- Congenital malformations could be prevented by giving pregnant women a list of what they should NOT and what they should do during pregnancy.

3.- In your opinion, what causes a congenital malformation?



In conclusion, according to the opinion of those interviewed, the most likely cause of a newborn being born with a congenital anomaly is genetics. This is followed by the consumption of drugs and alcohol during pregnancy and environmental factors. The maternal age was quite discarded for the experts, but it is not necessary to leave aside that the maternal ages are one of the number one causes of these congenital malformations.

4.- What is the role of the ministry of health or government in this issue



According to interviews, the Ministry of Health is responsible for the education of newborns with congenital disorders.

5.- Is it easy to deal with situations like this?

Finally, several of the interviewees gave the congenital malformations that are mostly found at the local level:

- Down's syndrome with 29%
- Apert syndrome with 18
- Crouzon syndrome with 23%
- Prune Belly syndrome with 18%
- Pierre Robins syndrome with 12%

3.2. Analysis of the applied interview

The malformations are something terrible that happens in the life of several human beings (newborns), but nobody knows of this subject better than the experts, that is to say, doctors. For this reason, several of them were interviewed to get their different opinions.

The interviews turned out to be quite effective, the interviewees gave to know all type of information that was unknown, from the approximate percentage of malformations that they see daily, to the types of them that they observe at a local level. That is to say that they gave their answers and respective opinions plus their explanation. An example could be: To the question "In your opinion, can congenital malformations be prevented in the future?". Answers whether or not they were given and explained.

In conclusion, all the interviews were a success and each one of the experts gave the help that was in their reach, answering each question.

CONCLUSIONS

At the end of this monographic work, it is concluded that: Every second, minute, an hour that passes is another child who is born with a condition, so it is good to take precautions during pregnancy, whether not to consume alcoholic beverages or drugs, take into account maternal ages, etc..

As explained in Chapter III, where treatments are discussed, each of these anomalies has something in common, which is when they say, "treat them as soon as possible. It is good that they imply that when it comes to this kind of thing, it is necessary to put all the effort into giving a better quality of life to the child who has the malformation.

The congenital malformations are infinite, all have their types, causes, and treatments. It is impossible to cure them but not to prevent them.

RECOMMENDATIONS

At the end of this work it is recommended that: The most recommended things for the future are:

1. Moms from the moment they know they are pregnant start taking the proper precautions prescribed by the gynecologist or obstetrician. (Do not drink alcohol, do not eat sushi, do not ingest drugs and do not exceed physically).
2. The gynecologist or obstetrician should explain in depth the subject of malformations (what they are, how they are formed, etc.) and how to prevent them.
3. If the newborn is born with some kind of congenital anomaly, treat it immediately.

4. On the part of the mother it would also be good for her to learn on her own a little bit about how a malformation is formed and its different treatments.
5. El ginecólogo podría hacer una revisión o diagnóstico temprano, y así poder ver si se puede tratar o no al neonato.

Congenital malformations are very common nowadays, and therefore these recommendations are very useful to help prevent them in the future.

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