Oct 26th, 1:30 PM - 3:00 PM

HUGE MALFORMATION OF HEAD (ENCEPHALOCELA) CASE REPORT

Besnik Elshani
University Clinical Center of Kosova, besnik.elshani@ubt-uni.net

Saliha Sh. Krasniqi
University for Business and Technology, salih.krasniqi@ubt-uni.net

Fëllënza Spahiu
University Clinical Centre of Kosovo

Yllka Krasniqi
University Clinical Centre of Kosovo

HUGE MALFORMATION OF HEAD (*ENCEPHALOCELA*)

CASE REPORT

(University Clinical Centre of Kosovo)

Besnik Elshani[1], Salih Krasniqi[1], Fëllënza Salihu[2], Yllka Krasniqi[2]

Abstract. The skull is one of the most important elements which serve to protect the human head, and including other organs with the jaw. With this being said, sometimes the skull is not strong enough such as when a baby is born, therefore a condition such as Encephalocele may appear. Encephalocele is a neural tube defect that may appear in Parietal Lobe, Occipital Lobe, and Anterior meaning the face of an individual. Moreover, the most common case is the Occipital Lobe, which is located in the back of the head, while Parietal in on the top of the head.

Introduction

As it is known, the skull is a (*strong*) skeleton of a person's head and serves to protect and enclose the most important organ in the human body, the brain. Despite this, skull is also important in other roles as well, such as supporting the jaws, other organs and so on. Additionally, the role of the skull in the human head is highly important when it comes to close damage(s). However, the skull is not always the same, by strength. When a baby is born, the human skull is made out of 44 bone elements, which in this case, tend to be separated, and it is not fully developed until the (*average*) age of **20**.

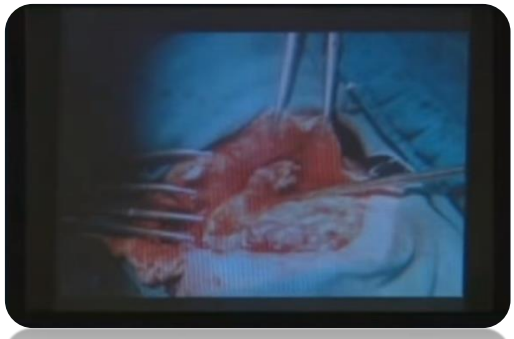
Certainly, when born, there are many cases where the skull is not developed properly, thus, followed with ***defects*** *and / or* ***deformation(s)***, which may also lead to neural tube damage and direct impact on the brain. So, what does the term **Encephalocele** mean? It is a form of condition, and appears usually ***1 in 10,000*** babies each year; these are the statistics from the United States, while the number in Europe may be around the same too [2].

Nevertheless, Encephalocele is a very rare defect that appears during the birth of the baby, more specifically in the neural tubes and affects the brain [2]. This leads to further causes that may damage the baby's health, by lowering the rate of its survival, followed with a very low weight (of the baby), and more upcoming birth defects. Ecephalocele usually belong in the back part of the brain, otherwise known as the ***Occipital Lobe*** (*as shown in the photo below*).

Technically, this condition (as mentioned), happens because the neural tube(s) do not close while the pregnancy of the mother. Furthermore, Encephalocele as seen, happens usually in the ***Occipital Lobe***, however, often happens also at the top of the head, and / or in the area between the ***Frontal Lobe*** (*forehead*) and the nose. Before moving on with its treatments, let's firstly focus on how to diagnose this condition. Despite these facts, such 'rare' cases also happen in Kosovo, particularly in the capital city, Prishtina. Between 2009 and 2010 and 2014, there were approximately four Encephalocele defects that needed to be treated resulting with good condition(s) of the patient at the end. As mentioned earlier that such cases happened in our country too, the regions were as follows: *Kacanik, Dragash and Shtime*.

CASE REPORT (B. Elshani)

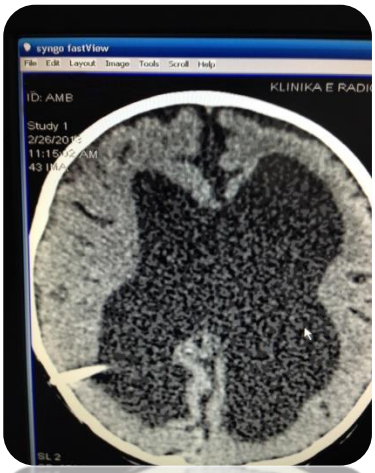
The newborn patient has been admitted for surgery to the neurosurgery clinic by the gynecology-boxing clinic of the children, as congenital malformations born of extremely large size encephalocele. This is the first case of such magnitude in the history of operations in the neurosurgery clinic of this congenital pathology. After pre-operative preparation, the patient, specifically the child, undergoes neurosurgical surgery in the operating room at the Neurosurgery Clinic in Pristina. The operating team resulted from the operating neurosurgeon, assistant neurosurgeon, anesthesiologist and assistant anesthesiologist and anesthesiologist technician as well as instrumentalists in the operating room. During the operation two blood units were delivered.

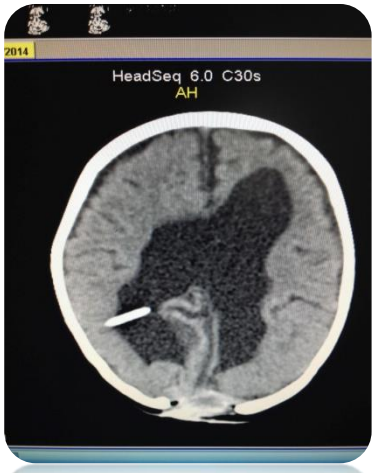


Technically in the neurosurgical aspect with a special delicacy with the help of electron microscope the malatonic congenital brain mass has been resected and carefully preserved, large blood vessels, arteries and veins of normal brain tissue. The surgery lasted about 3 hours and

the patient was taken to ICU Intensive Care Unit, where she was sent to a stable ward after several days of rehabilitation. Fortunately, the patient has survived such a high-risk operation successfully.

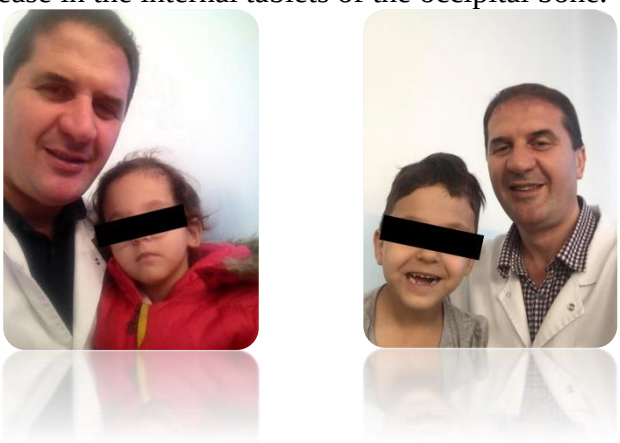
The patient was continuously followed up dynamically, with several head scans examined, where months later, as expected, we developed a hydrocephalus with increased intracranial pressures. The next neurosurgical operative step has been taken, in which case it is also placed in the neurosurgery clinic in the operating room, ventricular peritoneal shunt, to derive cerebrospinal fluid to avoid pressure on the brain tissue.





Lastly, it has been followed for many years, from 2012 until today, where we have noticed rapid and effective improvement. The patient is without any apparent neurological deficit has moderate intelligence, attends school; on a recent MRI examination of the brain, the cerebrospinal fluid derivative system is in full function, no signs of

hydrocephalus, and in the posterior part of the skull in the occipital lobe there is a natural increase in the internal tablets of the occipital bone.



(The child growth over the years)

Prognosis 1.0

Before moving on with the prognosis part, let's firstly focus on the brain tissue, which in this case is one of the most important parts of the human body. So what is brain tissue called?

Brain Tissue is located in the area between the brain and the skull, which results having three layers of tissue(s), and certainly they do serve to protecting the brain consistently and at any given time. Moreover, those three layers of tissues in that particular place are called meninges [4]. It is important to state the fact that ***"dura mater"*** plays a critical role, serving as the strongest layers out of them all. On the other side, the middle layer is consisted of having more elastic tissues and a very thin membrane of blood vessels, and it is named the arachnoid mater [4].

Moreover, before moving on with the treatment of encehalocele, having the correct prognosis of it is a highly suggested step. The prognosis for individuals with encehaloceles is completely depended on one of the types of tissues of brain involved, different brain malformations and last but not least, the location of the sacs. To be more precise, there are cysts within the brain that are and fulfilled with fluid(s) with sacs and may form in the brain at any time. Sacs on the other hand tend to have a very flexible structure.

Treatment 1.0

As stated in the Case Report by Dr. Besnik Elshani, the treatment of encehaloceles which is mostly characterized with the creation of sacs, it can be very critical at a certain point. However, the surgery, most of the time, is performed in a very chronological order, which is:

a. *Placing the protruding tissues back into the skull,*

b. *Completely removing the sac(s),*

c. *Fix / Correct any other associated craniofacial abnormalities of the brain.*

It is at high importance to keep the health of the brain, even on more difficult cases, which can be the removal of larger protrusions, without causing other (*major*) disabilities that may affect the functionality of the brain. Furthermore, when the fluid accumulates in the brain, which typically is founded in children, and sometimes enlarging the head itself, leading with side damages as well; this condition is known as the Hydrocephalus [1].

Hydrocephalus sometimes is associated with encephaloceles, and when this happens, the treatment of this severe condition, will require a shunt, while doing the surgical treatment. Besides this, as side treatments, they can be very much supportive and symptomatic at the same time [1].

Secondly, when it comes to the treatment of encephalocele (and any other malformation or disability of the brain), one of the best Hospitals in the World, is the Boston Children Hospitals [3]. Why Boston? Simply, because it clearly brings together a team of highly experienced neurosurgeons and this includes professionals in plastic surgery, oral surgery and maxillofacial surgery, which is face and jaw, dentistry and even psychologists. Why mentioning this certain case? Simply because, this hospital received a 3D-printed model of the brain structure, including the skull, to help observing different difficulties much easier, as well as creating a specific and clear plan before the surgery [3]. Also, this 3D Model helped the doctors to easier detect and locate diverse disabilities and malformations that if not treated correctly and within time, may lead to more severe and critical conditions, in this

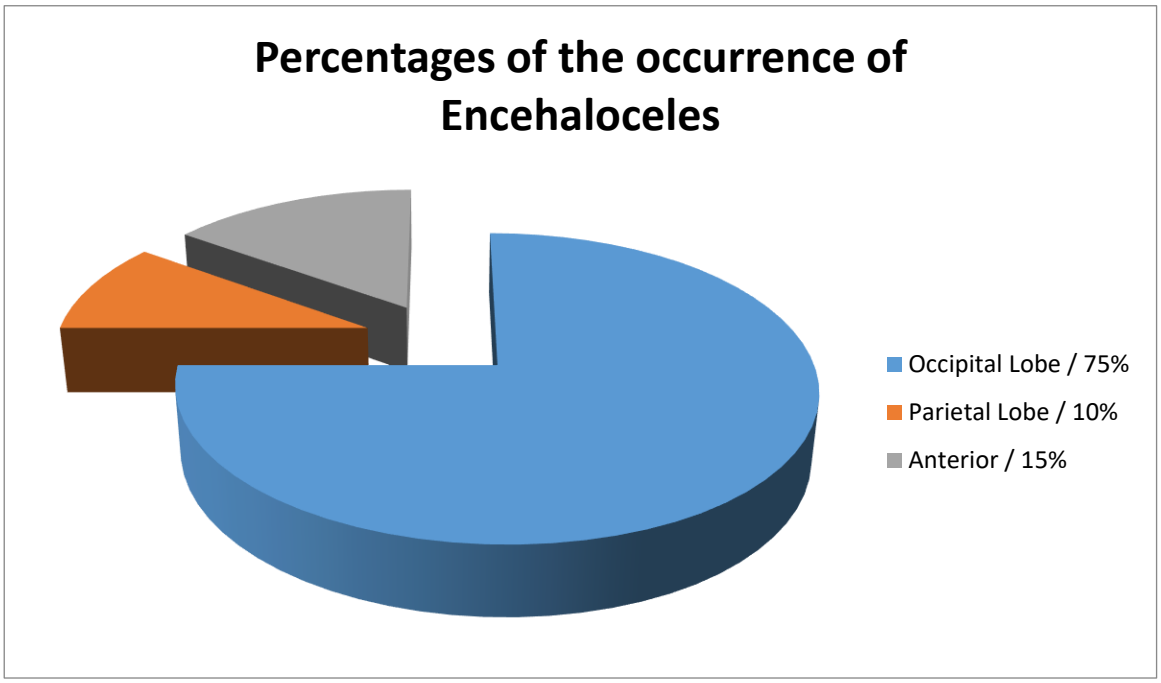
case, of young children. In their website it is presented a special case with a child that had encephalocele and his family was told that he was not going to live; after the operation the child successfully survived, making this a remarkable story to be shared [3]. The very same case happened in Prishtina, Kosovo too, with a child having an encephalocele in the Occipital Lobe, and after the surgery the child survived, and as mentioned in the case report, even after 7 years of the surgery, that child is going to school, doing her studies, being successful and showing not a single damage to the brain at the same time. The shunt is still working perfectly since the day it was placed.

How does 3D Print work precisely? The brain of the child *(and skull)* is perfectly scanned in order to create a unique model from an individual child's imaging, therefore, it allows the up mentioned neurosurgeons to rehearse major operations in more complex cases and minimalizing the disability even from developmental stage of the brain without causing any damage, since the priority focus is to keep a healthy brain and patient [3].

Treatment 1.1

When it comes to the treatment of encehaloceles, it is true that they are classified according to where they appear or where their location is, in this case. Since the head structure is precisely separated in human anatomy, it is much easier to locate one based on which lobe. The most often one is in the ***Occipital Lobe,*** where encephalocele is formed in the back of the head and sometimes it results with touching the neck, secondly there are ***Parietal Lobe*** encephaloceles and thirdly, ***Anterior*** encephaloceles and these ones arise from the base of the nose [4].

Note: To have a better image of the percentages of the Lobes, down below there is a table that clearly shows the occurrence of encehaloceles:



What are the main factors that cause Encehalocele and Hydrocephalus?

According to a study done by Dr. Besnik Elshani, it is true that Encehalocele is caused by the failure of the neural tube, while Hydrocephalus a buildup of the fluid in the ventricles (*or in this case cavities*), within the brain. Both conditions recently are found to be affected by different and diverse environmental factors. Even though, according to affected populations and a study done by The Centers of Disease Control and Prevention (CDC) shows that in the Western Populations the most common encephaloceles are located in the back of the skull, or otherwise known as the Occipital Lobe, while in Southeast Asia region, they are more located in the frontal lobe, while the latest cases in Kosovo [4].

The exact cause is yet unknown, but, there are multiple factors that clearly contribute in the formation of it. Mainly they can be genetic factors, such as having a history of the family with individuals who suffered or had neural tube defects, or a person who already is predisposed to certain disorders carries a gene or multiple genes of the disease. These genes are not necessarily able to function until they are

triggered to some point, under certain situation, such as exposing to different environmental factors as well [4].

Secondly, Hydrocephalus symptoms vary by age. It is said that usually the chances of occurring are to adults who are 60 years old+ and it starts by having an unusually large head, followed by a raid increase of it, and the creation of a soft spot, otherwise known as fontanel, which is located on the top of the head of the individual.

Thirdly, if it happens in toddlers and older children, there are other physical signs and symptoms which help detect the disease, and those include:

1. *Headache*
2. *Blurred or double vision*
3. *Eyes fixed downward (sun setting of eyes)*
4. *Abnormal enlargement of a toddler's head*
5. *Sleepiness or lethargy, poor appetite and more.*

The study done by Dr. Elshani found that another reason that leads to these symptoms are not necessarily the genes, but also from wars, that affected a certain population which then, led to its movement to another place causing irruption and congestion of that particular population. Having this in mind, this is not a preferred situation for pregnant women and women in general; hence, this may lead to disabilities of the future child, or children. Usually, such cases still appear mostly in the Balkan Peninsula as well as covering the Eastern Europe, because of the wars during the years. It is important to state that the exact event that leads to the creation of the hydrocephalus is still unknown too.

Conclusion

To finalize or conclude this topic, the skull is or plays a very important role protecting the most important organ in the human body, the brain. Additionally, as mentioned, the brain is consisted of lobes which are:

a. *Frontal Lobe*
b. *Parietal Lobe*

c. *Temporal Lobe*

d. *Occipital Lobe*

The skull's main duty is to protect the brain, supporting the jaw, and protecting other organs as well from damages that may come from outside, such as an accident. Since the babies are born, the skull certainly does not have the full strength as when it is developed, which means that, when a baby is born, it should be at high focus to not move the head of the baby much, in order to keep it safe from any damage [6]. However, as mentioned at the beginning, there are numerous cases when the skull may be not developed as it should, and this can lead to many defects or deformations [5].

Encephalocele is a skull defect which makes the brain move outside its own structure, damaging the neural tubes, and it creates a non-vital mass which is out of the normal or standard function. Such cases happen mostly in the United States, where 1 in 10,000 babies is born with this rare condition, or 375 babies per year [4]. Moreover, it is important to re-mention the fact that in United States the most common condition of Encephalocele 75% of the time appears in the Occipital Lobe, meaning in the back of the head, which usually sometimes it touches the neck [4].

On the other side, some statistics from different Universities, present the information that in the Southeastern Asia, the most common Encephalocele appears in the Frontal Lobe and in the nose area, meaning the face. When it comes to the Balkan Peninsula, Occipital Lobe is more spread (from all types of cases) but there have been cases which include other lobes too [1].

Besides Encephalocele, Hydrocephalus plays a critical role. It is a neural tube damage which can cause fluids in the brain, relating with our case, more specifically Dr. Elshani Case Report. Another highly important thing is that, this case in Prishtina, Kosovo has been followed for approximately 7 years and the condition of the child is very healthy, meaning, she is attending school, showings signs of intelligence and overall a good consistent health.

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