# Congenital Brain Malformations

## Clinical and Surgical Aspects

Khaled Fares AlAli
Hashim Talib Hashim
*Editors*

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# Congenital Brain Malformations


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Khaled Fares AlAli • Hashim Talib Hashim
Editors

# Congenital Brain Malformations

## Clinical and Surgical Aspects

Springer

*Editors*
Khaled Fares AlAli
Department of Neurosurgery
Zayed Military Hospital
Abu Dhabi, Abu Dhabi
United Arab Emirates

Hashim Talib Hashim
Department of Research
University of Warith Al-Anbiyaa
College of Medicine
Karbala, Iraq

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*To my Dearest Parents,*

*Your unwavering love, support, and sacrifices have been the guiding light on my scientific journey. Your belief in my potential has fueled my passion for discovery. This book is a tribute to your enduring faith in me.*

*To my Beloved Brothers and Sister,*

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*In this work, I strive to share the discoveries and insights that I have gained throughout my scientific career. Each page is a testament to the collective impact of those who have touched my life in various ways. With heartfelt gratitude, I present this book as a symbol of the profound influence you all have had on my journey.*

*May this book inspire others as you have inspired me, and may the spirit of discovery and learning continue to unite us.*

*With profound appreciation and love,*

***Hashim Talib Hashim***
***1-10-2023***
***H.T.H***

# Preface

In the complex tapestry of human biology, congenital brain malformations represent a profound enigma that has captured the fascination of scientists, clinicians, and individuals affected by these conditions. The pages of this scientific book are dedicated to unraveling the mysteries that shroud these malformations, to shedding light on their underlying causes, and to exploring the implications for diagnosis, treatment, and, ultimately, the lives of those who bear this burden.

As we embark on this journey through the intricate realm of congenital brain malformations, we are confronted by a profound sense of responsibility. This book is the result of years of tireless research, collaboration, and the collective wisdom of countless experts in the field. It seeks to bridge the gap between the scientific community and the wider world, offering a comprehensive overview of these conditions that are both rare and devastating in their consequences.

Throughout these pages, we will delve into the diverse array of congenital brain malformations, from the more common anomalies, such as neural tube defects and microcephaly, to the rare and intricate disorders that challenge our understanding of neural development. We will explore the genetic, environmental, and multifactorial determinants that contribute to their occurrence, as well as the mechanisms that underlie their pathogenesis. We will also discuss the latest diagnostic techniques, the potential for early intervention, and the ongoing quest for effective treatments.

This book is intended for a wide readership, from medical professionals and researchers to patients and their families. We hope that it will serve as a valuable resource for those seeking knowledge, understanding, and support in their journey through the labyrinth of congenital brain malformations. It is our aspiration that the information contained herein will foster collaboration and inspire new research endeavors, ultimately improving the lives of those affected by these conditions.

The study of congenital brain malformations is a testament to the resilience of the human spirit and the power of science to illuminate even the darkest corners of our existence. It is a journey of empathy and discovery, and as we delve into the intricacies of these malformations, we are reminded of the remarkable capacity of the human brain to adapt, evolve, and inspire.

We owe immense gratitude to the scientists, clinicians, patients, and families whose relentless dedication to understanding and confronting these conditions has made this book possible. It is our hope that the knowledge imparted here will pave the way for further breakthroughs and, ultimately, contribute to the well-being of those whose lives are touched by congenital brain malformations.

Abu Dhabi, Abu Dhabi, United Arab Emirates — Khaled Fares AlAli
Karbala, Iraq — Hashim Talib Hashim
1-10-2023

# Contents

# Contributors

**Toufik Abdul-Rahman** Sumy State University, Sumy, Ukraine

**Usama Afzaal** King Edward Medical University, Lahore, Pakistan

**Mays Sufyan Ahmad** College of Medicine, University of Baghdad, Baghdad, Iraq

**Aymar Akilimali** Faculty of Medicine, Official University of Bukavu, Bukavu, DR, Congo

**Nooralhuda Sameer Alash** College of Medicine, University of Baghdad, Baghdad, Iraq

**Sajjad Ghanim Al-Badri** College of Medicine, University of Baghdad, Baghdad, Iraq

**Danish Ali** Rawalpindi Medical University, Rawalpindi, Pakistan

**Hossam Tharwat Ali** MBBCH, Qena Faculty of Medicine, South Valley University, Qena, Egypt

**Zahraa Hussein Ali** College of Medicine, University of Baghdad, Baghdad, Iraq

**May Saad Al-Jorani** College of Medicine, Mustansiriyah University, Baghdad, Iraq

**Muntadher H. Almufadhal** College of Medicine, University of Baghdad, Baghdad, Iraq

**Ahmed Dheyaa Al-Obaidi** College of Medicine, University of Baghdad, Baghdad, Iraq

**Mustafa Najah Al-Obaidi** College of Medicine, University of Baghdad, Baghdad, Iraq

**F. A. Ameer** College of Medicine, Al-Qadisiyah University, Babil Governorate, Iraq

College of Medicine, University of Al-Qadisiyiah, Al Diwaniyah, Iraq

**Debrah Fosuah Anastasia** Sumy State University, Sumy, Ukraine

**Rayan Awada** Faculty of Medical Sciences, Lebanese University, Beirut, Lebanon

**Wireko Andrew Awuah** Sumy State University, Sumy, Ukraine

**Tulika Garg** Government Medical College and Hospital, Chandigarh, India

**Ibrahim Saeed Gataa** Oral and Maxillofacial Surgery, University of Warith Al-Anbiyaa, Karbala, Iraq

**Ali Qais Hasan** College of Medicine, University of Baghdad, Baghdad, Iraq

**Ali Talib Hashim** Golestan University for Medical Sciences, Gorgan, Iran

**Hashim Talib Hashim** Department of Research, University of Warith Al-Anbiyaa, College of Medicine, Karbala, Iraq

**Helen Huang** Royal College of Surgeons in Ireland, University of Medicine and Health Sciences, Dublin, Ireland

**Abbas Fadhil Abdul Hussein** College of Medicine, Babylon University, Babil, Iraq

**Mohammed Mohammed Hussein** College of Medicine, University of Baghdad, Baghdad, Iraq

**Karrar Ali Idan** College of Medicine, University of Baghdad, Baghdad, Iraq

**Hafiz Muhammad Iqbal** King Edward Medical University, Lahore, Pakistan

**Arda Isik** Department of General Surgery, Istanbul Medeniyet University, Istanbul, Turkey

**Riaz Jiffry** Royal College of Surgeons in Ireland, University of Medicine and Health Sciences, Dublin, Ireland

**Zaher Odai Khudher** College of Medicine, University of Baghdad, Baghdad, Iraq

**Mrinmoy Kundu** Institute of Medical Sciences and SUM Hospital, Bhubaneswar, India

**Moath Mohammed Madlool** College of Medicine, University of Baghdad, Baghdad, Iraq

**Ali Mahdi Mansoor** College of Medicine, University of Baghdad, Baghdad, Iraq

**Qasim Mehmood** King Edward Medical University, Lahore, Pakistan

**Abbas Mohammad** College of Medicine, Thi Qar University, Al-Nassiryah, Iraq

**Mohammed Qasim Mohammed** College of Medicine, University of Baghdad, Baghdad, Iraq

**Zahraa Qasim Mohammed** Baghdad Medical City, Baghdad, Iraq

**Hadi Mouslem** Faculty of Medical Sciences, Lebanese University, Beirut, Lebanon

**Hadi Mroueh** Carol davila University of Medicine and Pharmacy, Bucharest, Romania

**Ahraaf Munawar** King Edward Medical University, Lahore, Pakistan

**Abubakar Nazir** King Edward Medical University, Lahore, Pakistan

**Awais Nazir** King Edward Medical University, Lahore, Pakistan

**Saira Naz** King Edward Medical University, Lahore, Pakistan

**Jyi Cheng Ng** Faculty of Medicine and Health Sciences, University of Putra Malaysia, Serdang, Malaysia

**Hafiza Qurat ul ain** CMH Multan Institute of Medical Sciences, Multan, Pakistan

**Ahsan Rashid Rana** CMH Multan Institute of Medical Sciences, Multan, Pakistan

**Ali Abid Saadoon** College of Medicine, University of Warith Al-anbiyaa, Karbala, Iraq

**Faizan Saleem** King Edward Medical University, Lahore, Pakistan

**Aalaa Saleh** Faculty of Medical Sciences, Lebanese University, Beirut, Lebanon

**Hadiqa Shahid** King Edward Medical University, Lahore, Pakistan

**Farah Shibli** Faculty of Medical Sciences, Lebanese University, Beirut, Lebanon

**Moatamn Skuk** al-Kindi Medical College, Baghdad, Iraq

**Ta'ef Mohammed** College of Medicine, University of Baghdad, Baghdad, Iraq

**Shehroze Tabassum** King Edward Medical University, Lahore, Pakistan

**Rohan Yarlagadda** Rowan University School of Osteopathic Medicine, Stratford, NJ, USA

**Fatima Yasin** King Edward Medical University, Lahore, Pakistan

**Muhammad Zeeshan** King Edward Medical University, Lahore, Pakistan

# Chapter 1
# General Introduction to the Congenital Brain Malformations

**Hashim Talib Hashim, Mays Sufyan Ahmad, and Mohammed Qasim Mohammed**

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## 1.1 Introduction

The brain is the organ that is contained within the cranium and controls all the body's actions including memory, sensory, movement and emotions. Each part of it is dealing with action and exerts some work and it acts in a synergic way to keep the body moving [1].

H. T. Hashim (✉)
Department of Research, University of Warith Al-Anbiyaa, College of Medicine, Karbala, Iraq

M. S. Ahmad · M. Q. Mohammed
College of Medicine, University of Baghdad, Baghdad, Iraq



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During the embryological development of the brain, there will be very complex processes happening to result in the final shape that we see for the brain, and these processes are affected by many factors in the womb and the most effective factor is the folic acid deficiency during pregnancy [2].

When a defect occurs in one or all of the processes, congenital brain malformations will occur. There is a wide list of malformations that each one of them will be discussed in an independent chapter in this book. Each defect will result from a different process and each defect has its own criteria and nature. Each one of them will have different features from the other.

The malformations that will be discussed in this book will be as follow:

1. Anencephaly
2. Cephalocele
3. Chiari malformation
4. Porencephalies
5. Septo-optic dysplasia
6. Pituitary maldevelopment
7. Posterior fossa malformations
8. Microcephaly
9. Megalencephaly and Hemimegalencephaly
10. Neurocutaneous syndrome
11. Schizencephaly
12. Lissencephaly
13. Heterotopias
14. Polymicrogyria
15. Encephaloclastic disorders

Those 15 malformations will be discussed in our book. Each one of them will be extensively discussed and managed either medically or surgically.

## 1.2 Epidemiology

A study between 1995–1999, conducted in several European countries registered 11 congenital defects,2075 cases with anencephalus, encephalocele, spina bifida, and hydrocephalus were detected. 141 cases had proven to be caused by chromosomal

anomalies. The highest prenatal detection rate for malformation is for anencephalus, with a total number of cases 498 with a termination rate of 85%. There was a significant association between prenatal tests conduction and early diagnosis and termination of pregnancies with congenital diseases [3].

In a meta-analysis studying the prevalence of neural tube defects in Africa,' For 36 studies, "the median value of neural tube defects was 24.5 and the inter-quartile range was between 8.5 and 53 per 10, 000 birth" [4].

## 1.3 Diagnosis of These Malformations

The diagnosis of brain malformations can be divided into many stages:

1. Prenatal diagnosis by ultrasound or amniocentesis.
2. After delivery diagnosis by laboratory tests, imaging and general inspection and examination (Fig. 1.1).
3. Later one by neurological assessment for follow-up.

Genetic testing and karyotyping are also an important step of diagnosis either prenatally or after birth that can discover the genes defects and leads to the management protocol [5].

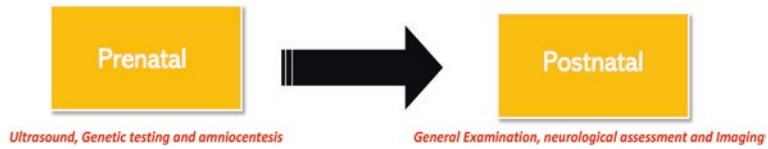


**Fig. 1.1** The diagnosis map for brain malformations

## 1.4 The Management and the Prognosis

The management of the brain's malformations depends on the type and the severity of this malformation. Some of these malformations can be left without any intervention if they are not dangerous or affect the body's functions. Some of them must be operated on as quickly as possible to prevent their progression [6].

The prognosis of the malformations is also dependent on the type of malformation and the severity. And it depends also on the way of choice to treat it and of course, surgical interventions have a worse prognosis than medical management especially since the brain is the operable organ and many complications and consequences are encountered to happen [7].

## 1.5 The Complications of Congenital Brain Malformations

There are many complications encountered with these malformations starting from cosmetic effects to low IQ and finally to loss of movement or paralysis [8].

The most serious complications are muscle paralysis which can involve the respiratory muscles and cause respiratory failure and death.

The complications' extent and severity depend on the site of the malformations and the region of the brain involved and also the management ways. Some surgeries even if they correct the defect but leave permanent damage that cannot be corrected and maybe the damage exists from the beginning that can be reversed after surgery (Fig. 1.2).


**Fig. 1.2** The possible complication for the congenital brain malformations

**Multiple Choice Questions**

1. **Congenital brain malformations(CBM) are:**

   (a) Prenatally developed
   (b) Postnatally developed
   (c) Can be managed medically
   (d) Without any effects on life quality

   ***Answer***: a

2. **The management of CBM depends on:**

   (a) The site of the defects
   (b) The extent of the defects
   (c) The severity of the defects
   (d) All the above

   ***Answer***: d

3. **The complications of the CBM include:**

   (a) Heart failure
   (b) Respiratory failure
   (c) Autism
   (d) High IQ

   ***Answer***: b

4. **The CBM can be diagnosed prenatally by:**

   (a) X-ray imaging
   (b) MRI imaging
   (c) Genetic studies
   (d) Cardiotocography

   ***Answer***: c

5. **If the malformation involves the occipital region and the brainstem, then it has:**

   (a) Good prognosis
   (b) Bad prognosis
   (c) Can be managed surgically with high successive rate
   (d) Cannot be treated

   ***Answer***: b

6. **The congenital brain malformations can be affected by folic acid deficiency:**

   (a) True
   (b) False

   ***Answer***: a

7. **The CBM can be managed surgically without any complications:**

   (a) True
   (b) False
   (c) It depends on the malformation

   ***Answer***: c

8. **The prognosis of the CBM s almost always good:**

   (a) True
   (b) False

   ***Answer***: b

9. **The management of the CBM include:**

   (a) Surgeries
   (b) Medical therapy
   (c) No intervention
   (d) All the possibilities

   ***Answer***: d

10. **Would you abort a fetus with CBM?**

    (a) Yes
    (b) No
    (c) It depends on the parents' choice
    (d) It depends on the country's law and the parents' choice

    ***Answer***: d

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# Chapter 2
# The Epidemiology of Congenital Brain Anomalies

**Fatima Yasin, Qasim Mehmood, Hadiqa Shahid, Ahraaf Munawar, and Ali Abid Saadoon**

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## 2.1 Introduction

Congenital brain anomalies are a group of brain tissue defects that result from abnormal developmental processes during fetal life. The exact cause of these defects is not known, but they are hypothesized to arise as a result of genetic mutations or defects during various stages of fetal and embryonic life. Brain development in the fetus starts at a very early stage, and any disruption in the normal development of neurons or nerve cells can give rise to congenital brain anomalies. They can also occur if the development of the skull and cranium is not proper. Furthermore, various environmental insults to the mother, like toxins, trauma, drugs, and infections, can predispose to the development of congenital brain lesions. The signs and symptoms of these anomalies are highly varied and can range from a delay in early

F. Yasin · Q. Mehmood (✉) · H. Shahid · A. Munawar
King Edward Medical University, Lahore, Pakistan

A. A. Saadoon
College of Medicine, University of Warith Al-anbiyaa, Karbala, Iraq
e-mail: ali.sa@uwoa.edu.iq



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development to life-threatening seizures and epilepsy [1]. Various congenital brain lesions have been noted in children and the epidemiology of some of them is explained below.

## 2.2 Anencephaly

Anencephaly is the most widely recognized CNS problem. The general global estimation of the prevalence of anencephaly is approximately 5.1 per 10,000 births, with a 95% confidence interval between 4.7 and 5.5 per 10,000 births. Globally, the incidence of anencephaly is 8.3 per 10,000 births, with a 95% confidence interval between 5.5 and 9.9 per 10,000 births. Similarly, the worldwide attenuation value for anencephaly is 5.5 per 10,000 births, with a 95% confidence interval between 1.8 and 15 per 10,000 births. The most noteworthy of these values, according to the subgroup analysis, is on the Australian mainland, with 8.6 per 10,000 births and a 95% confidence interval between 7.7 and 9.5 per 10,000 births [2].

## 2.3 Cephalocele

Cephalocele is depicted as an inherent skull vault imperfection with related herniation of hidden intracranial contents, with prevalence changing from 1 out of 3500 to 1 of every 5000 live births [3]. Atretic cephalocele is an interesting and rarely diagnosed condition, with a prevalence of cephalocele assessed to be about 0.8–3.0 per 10,000 births. An atretic cephalocele alludes to a cephalocele that is arrested during the phase of development and addresses roughly 40% to half of all cephaloceles [4].

## 2.4 Chiari Malformations

Chiari malformations are an assortment of hindbrain and craniocervical intersection irregularities, of which the Chiari 1 malformation is the most commonly seen type in clinical practice with a prevalence of 0.56–0.75% on MRI [5]. Chiari malformation type 1 influences roughly one out of 1000 individuals, albeit one out of 100 meet radiological rules, making it a typical neurological issue [6]. Moreover, the rates of Chiari malformation don't vary based on gender in children. Chiari malformation is more common in grown-up women compared to men. Women are diagnosed 3–5 times more often with Chiari malformation than men [7].

## 2.5 Porencephaly

Porencephaly is a neurological condition that can develop previously or after birth and is characterized by cysts situated in any spot inside the brain parenchyma, which by and large is covered by plain walls and encompassed by an atrophic crust. Its inheritance is autosomal dominant [8]. Porencephaly is an uncommon disorder, assessed in 3.5 out of every 10,000 live births [9]. Moreover, porencephaly is rare in the elderly and is seen mostly in neonates because it is usually congenital [10].

## 2.6 Septo-Optic Dysplasia

Septo-optic dysplasia, also known as de Morsier's syndrome, is a rarely occurring congenital disorder. Its occurrence is about 1 in 10,000 live births [11]. A population-based study was done in Europe that tracked down a lower prevalence of 1.9–2.5 per 100,000 live births. One more review from Canada tracked down a prevalence from 53.3 per 100,000 to 113.3 per 100,000 live births showing an advancing rate with time. By and large, there is no preference for gender, with fundamentally the same affection for men and women [12]. The reported incidence for this rare disorder is 1.9–2.5 per 100,000 live births. Most of these cases were reported in childhood or early adolescence [13].

## 2.7 Pituitary Maldevelopement

Congenital hypopituitarism has an annual incidence of 4.2 cases per 100,000. It is more common in males, with a male-to-female ratio of 1.9:1. Mean age for congenital hypopituitarism is 12.8 years [14]. According to a study, 22 out of 37 patients with idiopathic hypopituitarism had congenital hypoplasia of the anterior pituitary with stalk agenesis and an ectopic posterior pituitary (group 1). 15 out of 37 showed isolated hypoplasia of the anterior pituitary (group 2). 81.81% of children in Group 1 had a history of adverse perinatal incidents, and 68.18% had a breach presentation. 54.54% of group 1 born via vaginal delivery developed multiple pituitary hormone deficiencies and 13.6 born via cesarean section had only isolated growth hormone deficiency. The incidence of breech delivery in group 2 was only 13.33 percent and only isolated growth hormone deficiency [15].

## 2.8 Posterior Fossa Malformation

Anomalies of the posterior fossa are relatively common, but limited data is available on their epidemiology [16]. The most commonly observed posterior fossa malformations include Dandy-Walker malformation, mega cisterna magna, Blake's pouch,

and vermian hypoplasia [17]. According to a European population-based study, the prenatal diagnosis of Dandy-Walker malformation is 6.79 per 100,000 births (with a 95% confidence interval of 5.79–7.96). Live birth prevalence was 2.74 per 100,000 births (95% confidence interval: 2.08–3.61). The Dandy-Walker variant had a prevalence of 2.08 per 100,000 births (95% confidence interval between 1.39 and 3.13) [18]. The prevalence of chromosomal aberrations in Dandy-Walker malformation was 16.3% [17].

## 2.9 Microcephaly

Risk factors for isolated microcephaly include alcohol use, inadequate weight gain in pregnancy, and black ethnicity. Mothers with a previous live birth are protected against microcephaly as compared to nulliparous mothers [19]. There are limited population-based studies available on the epidemiology of microcephaly. The prevalence of total and total severe congenital microcephaly are 14.7 and 4.8 per 10,000 live births, respectively. Young maternal age, higher BMI, and multiparity reduced the risk of unexplained congenital microcephaly [20]. Epidemiological data shows a temporal association between the epidemic of the Zika virus and the incidence of microcephaly [21].

## 2.10 Megalencephaly and Hemimegalencephaly

Megalencephaly is more likely to be an isolated cerebral anomaly in almost 71 percent of cases. The adjusted prevalence of megalencephaly is 0.08 per 10,000 births, with a 95% confidence interval between 0.05 and 0.11. Published incidence figures for this anomaly are not available [22]. Often, hemimegalencephaly is found as an isolated cerebral anomaly or as an occasional finding in a large number of syndromes. A retrospective study showed that 53% of cases of HME were not syndromic, while 47% of cases were part of a known or suspected genetic syndrome [23].

## 2.11 Neurocutaneous Syndromes

Common neurocutaneous syndromes include neurofibromatosis, tuberous sclerosis, Sturge-Weber syndrome, von Hippel-Lindau disease, and Ehlers-Danlos syndrome. They are mostly genetically determined, except for Sturge-Weber syndrome, although sporadic cases may be present [24]. The prevalence of Sturge Weber syndrome is 1 in every 50,000 live births. Males and females are equally affected by it. The prevalence of tuberous sclerosis is 1 in 6000–9000 [25]. The prevalence of neurofibromatosis type 1 is 1 in every 3000 live births [26].

## 2.12 Schizencephaly

It is a rare disorder [27], which is why its exact prevalence has always been difficult to estimate [28]. A study in the UK has shown the prevalence of schizophrenia to be 1.48 in every 100,000 births [29]. A retrospective study, however, was done in southeast Hungary to estimate the prevalence of schizophrenia over the age of 14 years. Among 185,486, only 10 patients were suffering from Schizencephaly, among which 6 were boys and 4 were girls. This study concluded a prevalence of 0.54 per 10,000 births [30]. A study done in Japan concluded the incidence of schizophrenia to be 5.2 per 100,000 live births [31]. A study of more than four million people in California showed that the population prevalence of Schizencephaly was 1.54/100000 births from 1985 to 2001 [32]. Another study by Oxford University said that this rare malformation is associated with an estimated incidence of 1.5 per 100,000 live births and 1 in 650 to 1 in the 1650 children suffering from epilepsy [33].

## 2.13 Lissencephaly

Lissencephaly is a range of disorders that encompasses a wide variety of brain malformations. It includes agyria (that is, the absence of brain gyri), pachygyria (that is, the presence of broad brain gyri), and subcortical band heterotopia [34], but it is a very rare CNS malformation [35]. It consists of two types, type 1 and type 2 [36]. Type 1 Lissencephaly has an estimated prevalence of 11.2 per 10,000 live births according to a study in the Netherlands [37]. However, the prevalence of this type has not been described in any study to date [36].

## 2.14 Heterotopia

Heterotopias are of various types. The reported prevalence of gastric heterotopia of the cervical esophagus is 0.18–14%. The gastric heterotopias are also called inlet patches (IP). However, there is a motion that this prevalence is underestimated [38]. Because a study in the tertiary care hospital showed that the prevalence there is much lower, at 0.18% [39]. According to [40], the prevalence of gastric heterotopia was 1. 9% and 2.2% of these patients had associated H pylori gastritis. Another study reported it to b 4.3% [41] and comparable results were reported by [42], which is almost 3%.

## 2.15 Polymicrogyria

It is a disorder of late migration and then post-migratory organization of the cortex in a way that results in a malformation. This malformation is accompanied by many small but fused gyri [43]. It is a rare disease; however, it is more common as compared to other malformations. The prevalence of polymicrogyria is 1 in 2000 [44]. Another study showed that 10.37% of patients were found to have polymicrogyria [45].

## 2.16 Encephalopathy Disorders

Hashimoto encephalopathy (HE) is a common clinical manifestation that presents as an encephalopathy; however, a central nervous system infection or tumor is not associated with it. The HE is associated with low prevalence [46]. However, Wernicke encephalopathy has a prevalence between 0.4% and 2.8% [47, 48]. The highest prevalence was found in Austria (1.1–2.8%), as shown by [47, 49, 50]. However, the use of alcohol is associated with an increased prevalence of 12 percent, as reported by [47]. However, chronic traumatic encephalopathy is associated with 0.79% of them, and 60% of them were associated with a history of traumatic brain injury [51].

**Multiple Choice Questions**

1. **Q 1. A rare brain malformation accompanied by slits in the cerebral hemispheres is called:**

   (a) Schizencephaly
   (b) Lissencephaly
   (c) Hemimegalencephaly
   (d) Polymicrogyria
   (e) None of these

   **Answer: a**

   **Explanation: Slits are observed n cerebral hemispheres of patients suffering from Schizencephaly.**

2. **Q 2. How many types of Lissencephaly are there?**

   (a) One
   (b) Two
   (c) Three
   (d) Four
   (e) None of the Above

   **Answer: b**

   **Explanation: There are two types of Lissencephaly, Type 1 and Type 2.**

3. **Q 3. Polymicrogyria is a CNS malformation characterized by:**

   (a) Early migration
   (b) Late migration
   (c) Non migration
   (d) Excessive migration
   (e) None of the above

   **Answer: b**

   **Explanation: Late migration leads to a new subcortical organization that results in small fused gyri known as Polymicrogyria.**

4. **Q 4. Gastric heterotopias in the cervical esophagus are also known as:**

   (a) Inlet patch
   (b) Outlet patch
   (c) Cerebral Heterotopia
   (d) Both A and B
   (e) None of these

   **Answer: a**

   **Explanation: Inlet patch is another term used for cervical gastric heterotopias.**

5. **Q 5. Alcohol abuse is associated with the following encephalopathy:**

   (a) Hashimoto encephalopathy
   (b) Hepatic encephalopathy
   (c) Wernicke encephalopathy
   (d) Hypoxic Ischemic encephalopathy
   (e) Chronic traumatic encephalopathy

   **Answer: c**

   **Explanation: Research has proved the association of alcohol with the increased prevalence of Wernicke encephalopathy.**

6. **Q 6. Congenital hypopituitarism is more common in**

   (a) Males
   (b) Females
   (c) Equal distribution
   (d) Transgender
   (e) None of these

   **Answer: a**

   **Explanation: Congenital hypopituitarism is more common in males having male to female ratio of 1.9: 1**

7. **Q 7. Posterior fossa malformations include all except:**

   (a) Dandy Walker malformation
   (b) Mega cisterna magna
   (c) Porencephaly
   (d) Blake ‘s pouch cyst
   (e) Vermian hypoplasia

   **Answer: c**

   **Explanation: Mostly observed posterior fossa malformations include Dandy Walker malformation, mega cisterna magna, Blake ‘s pouch and vermian hypoplasia.**

8. **Q 8. Risk of congenital microcephaly is decreased by:**

   (a) Black ethnicity
   (b) Nulliparous mothers
   (c) Maternal alcohol use
   (d) Multiparity
   (e) Lower BMI

   **Answer: d**

   **Explanation: Young maternal age,higher BMI and multiparity reduced the risk of unexplained congenital microcephaly.**

9. **Q 9. Congenital brain malformation characterized by unilateral enlargement of cerebral hemisphere is called:**

   (a) Megalencephaly
   (b) Hemimegalencephaly
   (c) Archinencephaly
   (d) Holoprosencephaly
   (e) None of the above

   **Answer: b**

   **Explanation: Congenital brain malformation characterized by unilateral enlargement of cerebral hemisphere is called hemimegalencephaly.**

10. **Q 10. Which of the following neurocutaneous syndrome is not genetically determined?**

    (a) Neurofibromatosis
    (b) Tuberous Sclerosis
    (c) Sturge Weber syndrome
    (d) Von hippel Lindau disease
    (e) Ehlers Danlos syndrome

    **Answer: c**

    **Explanation: neurocutaneous syndromes are mostly genetically determined except for sturge Weber syndrome.**

11. **Q 11. What is an inherent skull vault imperfection with herniation of intracerebral contents?**

    (a) Anencephaly
    (b) Porencephaly
    (c) Cephalocele
    (d) Septo-optic dysplasia
    (e) None

    **Answer: c**

    **Explanation: Cephalocele is the skull vault imperfection that is assosiated to herniation of intracerebral contents.**

12. **Q 12. What is the other name for Morsier's syndrome?**

    (a) Anencephaly
    (b) Chiari malformations
    (c) Porencephaly
    (d) Septo-optic dysplasia
    (e) None

    **Answer: d**

    **Explanation: Septo-optic dysplasia is the other name for Morsier's syndrome and is a rarely occuring congenital disorder.**

13. **Q 13. Chiari malformation is more common in:**

    (a) Women
    (b) Men
    (c) Equal incidence
    (d) None
    (e) Transgender

    **Answer: a**

    **Explanation: Chiari malformation is more in grown up women as compared to men.**

14. **Q 14. Which of these presents as cysts occuring anywhere inside the brain parenchyma?**

    (a) Anencephaly
    (b) Hemimegalencephaly
    (c) Holoprosencephaly
    (d) Porencephaly
    (e) None of the above

    **Answer: d**

    **Explanation: Porencephaly manifests itself as cysts that can be present at any part inside the mind parenchyma.**

15. **Q 15. Septo-optic dysplasia affects more:**
    (a) Women
    (b) Men
    (c) No gender preferences
    (d) Equal distribution
    (e) Transgender

    **Answer: c**

    **Explanation: Septo-optic dysplasia shows no gender preference for men or women.**

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