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Karnataka Paediatric Journal

Editorial

Mumps resurgence-strategy ahead

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Mumps is a vaccine-preventable disease that usually occurs as parotitis, but it can also lead to several life-threatening complications, including pancreatitis, meningitis, and encephalitis. Mumps disease is caused by mumps virus (MV), which belongs to the genus Rubulavirus of the family Paramyxoviridae. MV is antigenically monotypic and spreads by respiratory droplets. Mumps is a vaccine-preventable disease that is endemic in most parts of the world.

Although mumps is preventable by vaccination, there has been a surge in cases occurring among vaccinated populations. The most common complication of mumps in children is meningitis, sometimes associated with encephalitis. Encephalitis is the most dangerous complication and can result in seizures, paralysis or other neurologic conditions. It is the most frequent cause of the very rare deaths attributed to mumps. The mortality rate for patients presenting with meningoencephalitis has been reported to be up to 1.4%. Other rare complications include, but are not limited to, nephritis, arthritis, thrombocytopenia purpura, mastitis, thyroiditis, and keratouveitis. Post-pubertal males can also develop orchitis in approximately 20% of the cases. Oophoritis in females can also occur but with significantly less frequency. Although hearing loss is a rare (1%) complication of mumps, it is usually unilateral and temporary. However, hearing loss can be permanent, and mumps is the most frequent cause of one-sided sensorineural deafness in children.

Mumps is a public health problem in India; however, inadequate data from different parts of the country underestimate the true extent of the burden. In India, very limited data are available on the epidemiology of mumps. Many outbreak reports, few zero-prevalence studies and vaccine studies on mumps are available from different parts of the country. It has been observed that there is no uniformity in the methodology of surveillance, serological testing algorithm, attempt for virus isolation, and use of available molecular tools and sequencing. Limited information is available about the seasonality of mumps cases in the country. Circulation of two MVs (i.e., genotypes C and G) were reported from India; more genotyping studies are necessary to understand other indigenous MV circulation if any.

Mumps continues to occur in epidemic proportions in India despite the availability of a safe and effective vaccine. Mumps outbreaks or sporadic cases have been periodically reported from the States of Kerala, Maharashtra, Gujarat, Karnataka, Punjab, Tamil Nadu, Uttar Pradesh, and West Bengal. These outbreaks or sporadic cases were confirmed either by clinical presentation or using serological or molecular tools. Only a handful of studies confirmed mumps by serological or molecular tools while in the remaining studies, clinical diagnosis was used for confirming mumps. Reports suggest that due to a lack of surveillance and documentation systems, the burden of mumps is underestimated in India. Similarly, mumps-associated complications and outcomes of patients are not reported systematically.

The recent outbreaks and resurgence of mumps are thought to have occurred for multiple reasons, including declining levels of vaccine-derived immunity and the lack of recommended boosters for the measles, mumps, and rubella (MMR) vaccine. Prevention through vaccination is a key in the management of mumps since there is only supportive therapy for infected patients.

Mumps vaccination has been incorporated into the regular immunization schedule of many countries, usually along with measles and rubella vaccines in a triple formulation. These vaccines have enabled the World Health Organization (WHO) to establish global strategies for the advanced control of measles and rubella leading to an elimination target in some regions. However, in contrast to rubella and measles, secondary vaccine failure occurs frequently in the case of mumps and circulation of MV within highly vaccinated populations has been frequently reported.

Mumps-containing vaccines have not been included in the Universal Immunization Programme (UIP) or National immunization schedule but are available in the private healthcare system. Despite being a vaccine-preventable disease, mumps has never been a part of the UIP because of the disease's no-mortality profile and the perception that it has low public health significance. However, the Indian Academy of Paediatrics (IAP) has always maintained that the public health significance of mumps has been underestimated mainly because of the poor documentation of clinical cases, its complications, and patients' follow-up data as well as the lack of published studies. There is no nationally representative data on the incidence of the disease. There is very little information on the actual long-term morbidity profile of the disease even though the disease is known to have some impact on reproductive organs.

Considering reports on mumps cases or outbreaks and mumps-related complications from different parts of the country, the IAP suggested the inclusion of mumps antigen in the form of MMR vaccine, the first dose at 9 months and the second dose at 16–24 months and a third dose at 4–5 years. The IAP Committee on Immunization has reiterated the inclusion of mumps antigen in UIP as MMR vaccine instead of measles-rubella vaccine.

A major difficulty with curbing the spread of mumps is its long gestation period (the time between infection setting in and disease manifestation). Much like COVID-19, many remain asymptomatic and can be carriers of the virus. The current infections are largely in those who are unvaccinated or yet to complete all three doses. Another theory is the chance that the virus may have mutated to become more infectious and possibly escape the protection offered by the vaccines. However, there is no evidence yet to support the claim. Health officials maintain that creating public awareness about the disease and the importance of isolation is the most important tool in bringing down the transmission of the disease. Mumps is primarily being reported in unimmunized children and adolescents and hence improving general immunization coverage is important. Transmission of the disease begins before the symptoms actually manifest and isolation of the patient for a full 3 weeks is necessary to limit the spread of the disease.

According to the WHO, vaccination strategies targeting mumps control should be closely integrated with existing measles elimination and rubella control. The IAP for one has always advocated the use of MMR vaccine in UIP, which has been available in the private sector for a long time. There are no studies from India on the effectiveness of the mumps vaccine. However globally, the protection from two doses is estimated to be between 70 and 95%, if the coverage is high.

MMR are vaccine-preventable disease. However, morbidity and mortality due to these diseases remain largely unnoticed in India. Measles has received much attention; mumps and rubella still need to garner attention. According to the WHO, near-elimination of mumps could be achieved by maintaining high vaccine coverage using a two-dose strategy. However, the Government of India has not yet decided on the mumps vaccine. Overall, mumps seems to be a significant public health problem in India but does not garner attention due to the absence of a surveillance and documentation system. Thus, the inclusion of mumps antigen in the UIP would have added advantages, with the economic burden imposed by the cost of the vaccine offset by a reduction in disease burden. It is high time that the government includes the mumps vaccine in the national immunization schedule. IAP should once again strongly advocate the inclusion of the mumps vaccine in the national immunisation programme.

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Karnataka Paediatric Journal

Original Article

Pattern of unintentional injuries in children seen in a tertiary hospital in Nigeria

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ABSTRACT

Objectives: Unintentional injuries are bound to occur in children. They are of major public health concern. This study aims to report the type of unintentional injuries and the location of the injuries as seen in a tertiary hospital.

Material and Methods: This is an 'ambispective' hospital-based study lasting for 12 months. This was conducted in the emergency paediatric units of two tertiary hospitals in Southern Nigeria. Every child who reported unintentional injury was included in the study. Structured questionnaires were used to collect the data by trained research assistants. Data were collected and analysed using International Business Machines Statistical Package for the Social Sciences. The results were represented in bar and pie charts, respectively. Ethical approval was given by the Institutional Ethics and Research Committee.

Results: A total of 212 children were included in the study. Males were the highest (60.8%), and children ≤10 years of age were 64.5%, of which those ≤5 years old were 34.6% respectively. Falls were the main cause of injury in all age groups, constituting 53.9%, followed by ingestion of toxic substances and foreign bodies (29.1%). The upper and lower limbs were mostly affected in 79.2% (n = 168) of the children. Males were commonly injured in the lower limbs.

Conclusion: Unintentional injuries are commoner in children ≤5 years old; unintentional falls are the most common cause in all age groups and affect mostly the upper and lower limbs.

Keywords: Unintentional injury, Children, Falls, Toxic substance

INTRODUCTION

Unintentional injuries are of major concern worldwide due its attendant morbidity and mortality. Children of all ages are at higher risk of suffering from these injuries.[1] Unintentional injuries are the leading cause of death among children, but the exact proportions vary. There is variation in the causes of unintentional injuries in children from different races, sexes, and geographical locations. In essence, there is a substantial difference whether the regions are developing or developed.[2]

These injuries could be traumatic and non-traumatic. Traumatic injuries are caused by unintentional falls, road traffic injuries, sporting, recreational, fighting, etc. While the non-traumatic injuries are due to unintentional poisoning through ingestion of toxic substances such as insecticides, excessive alcoholic beverages, drug overdose, drowning, and suffocation, the

occurrence of these injuries can be influenced by weather conditions and time of the day.[3-5]

Unintentional injuries could lead to emotional trauma and financial burden to families and society at large. These can affect the child's learning ability, relationships, and unstructured play. Treatment of the unintentionally injured child requires specialised skills and attention to detail due to their physiologic makeup.

Many studies have suggested ways of mitigating the burden of these injuries, such as caregiver supervision, monitoring, use of helmet seatbelt, penalties for traffic violations such as speed limits, and keeping toxic substances out of reach for children.[2,5,6] Some studies have advocated on cause-specific prevention.

In my country, there is a paucity of data on unintentional injuries to build a national surveillance network, and none of these studies talked about the areas of the body commonly affected.[4,6]

This study aims to outline the pattern of unintentional injuries seen in a tertiary hospital and the location of the injury in the body. This will contribute to national data on unintentional injuries in children to help build a national surveillance network.

MATERIAL AND METHODS

This is an 'ambispective' study Asuquo *et al*.[7] which is midway between retrospective and prospective study. The study is a hospital-based study that took place in the emergency paediatric units of two tertiary hospitals in Southern Nigeria. These hospitals are referral centres in their respective regions. The University of Calabar Teaching Hospital (UCTH) is an 800-bed multispeciality hospital located in South-South Nigeria, while the National Orthopaedic Hospital Enugu (NOHE) is located in South-East Nigeria. They both have paediatric orthopaedic units, while the University of Calabar has both paediatric surgical and medical divisions.

The emergency paediatric unit of UCTH and the emergency unit of NOHE were the primary points of call for all unintentional injuries affecting children. An interviewer-administered questionnaire was used to collect information from a sample of caregivers of children with unintentional injuries attending these institutions. Some patient information was extracted from the medical journals of patients who were seen within the study period and included in the study. A total of 212 was the sample size obtained by using Andrew Fisher's equation[8] for calculating sample size.

The authors and research assistants were used to collect the data from the caregivers and the patient medical journals of those treated and discharged within the study period. The study period spans from February 1, 2023, to January 31, 2024.

The data collected were social demographic characteristics, causes of injury, location of the injury in the body, etc.

The data were analysed using International Business Machines (IBM) Statistical Package for the Social Sciences (IBM Corp., Armonk, NY USA). Descriptive statistics were conducted. The results were presented as percentages and frequencies in pie and bar charts, respectively. Significant inferential statistic was placed at $P \leq 0.05$.

Ethical approval was obtained from the Institutional Ethics and Research Committees.

RESULTS

Two hundred and twelve children were included in the study. The highest age group was 1–5 years $n = 73$ (34.6%), followed by 6–10 years $n = 63$ (29.9%) and 11–15 years $n = 59$ (28%) [Figure 1].

There was a high male preponderance of 60.8% [Figure 2].

The causes of unintentional injuries were falls, which constitute 53.9%, followed by non-traumatic injuries 29.1% (this includes ingestion of toxic substances such as insecticides, drowning, suffocation, foreign object ingestion, drug overdose, and alcohol), while all forms of accidents were 17% [Figure 3].

Regarding the location of unintentional injuries, the upper limbs were commonly affected, constituting 43.4% ($n = 92$), followed by the lower limb with 35.8% ($n = 76$). The head, face, and eyes were 3.3% ($n = 7$), the chest was 7.5% ($n = 16$), the neck was 9% ($n = 19$) and abdomen was 0.9% ($n = 2$) [Figure 4].

Regarding gender and location of the injury, males were commonly injured in the lower limb (M = 54 and F = 22),

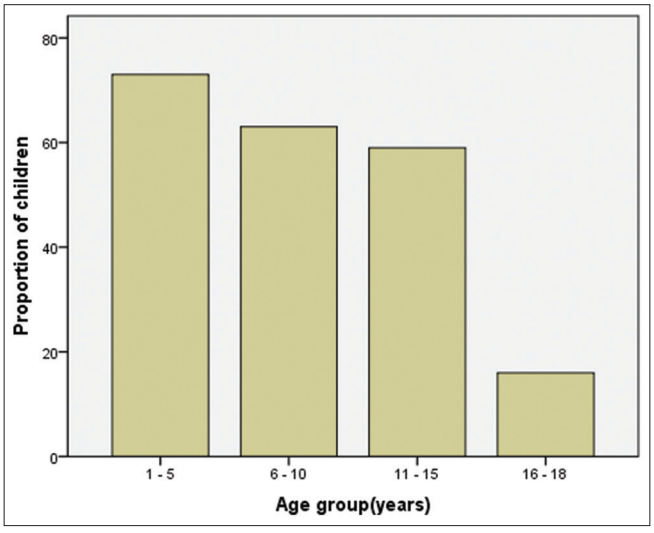

Figure 1: Distribution of age in groups.

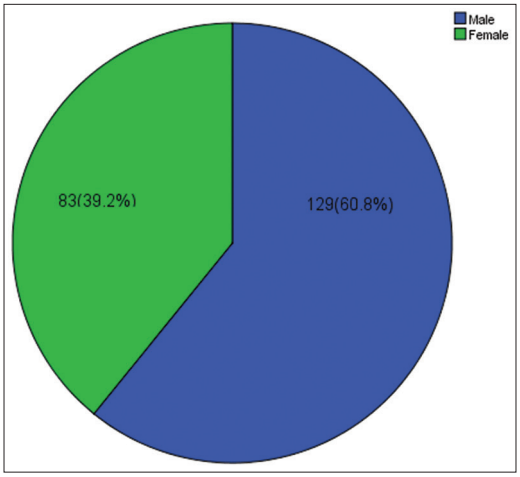

Figure 2: Sex distribution.

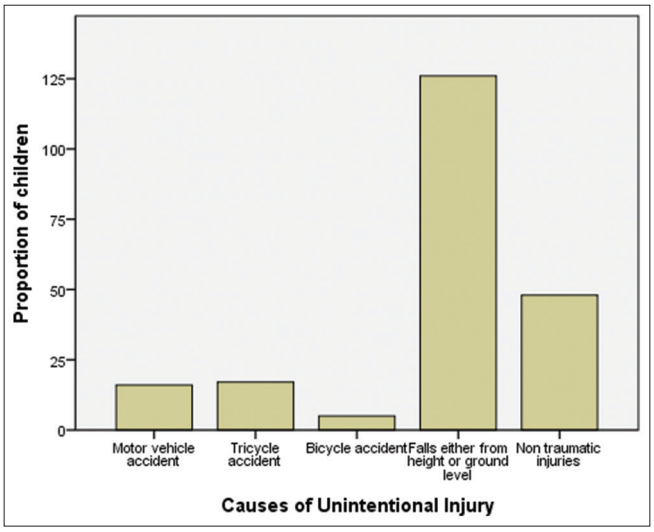

Figure 3: Causes of unintentional injury.

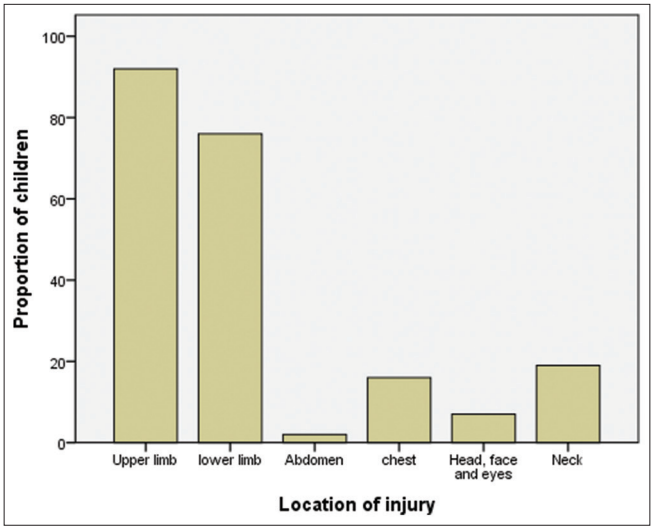

Figure 4: Location of unintentional injury.

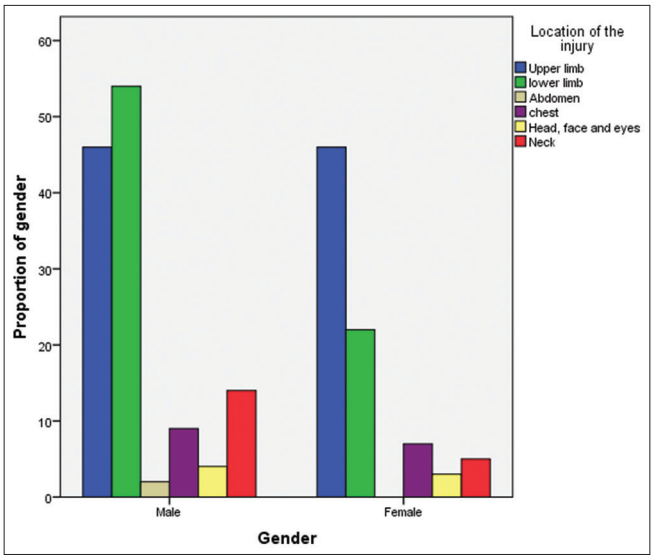

Figure 5: Location of the injury in both genders.

neck (M = 14 and F = 5), chest, abdomen, and head. The lower limb injury was significant, with a *P* value of 0.045 [Figure 5].

Children of the pre-school age are more likely to suffer from unintentional falls and non-traumatic injuries [Table 1].

DISCUSSION

Unintentional injuries are bound to occur in children of all ages. The effects of these injuries can be mitigated by preventive measures.

In our series, the majority of the injuries were traumatic in origin, constituting about two-thirds of all injuries seen. This finding was similar to that reported by a Saudi and Chinese study.[2,9] The non-traumatic injuries range from ingestion of insecticides and petrochemicals, especially kerosene, ingestion of foreign bodies, and suffocation, etc. This finding was similar to that reported by a Northern Nigerian study, Saudi and Chinese study. In the Chinese and Saudi studies, drowning and suffocation were among the injuries, but in our setting, these types of injuries are rarely reported, especially if the outcome was fatal.[2,6,9]

In our study, the hand, arm, forearm, legs, and thigh are the most common locations of the injury in the body, followed by the head and neck. Injuries to the head can be fatal in some cases. Most of the reported literature did not take cognizance of the body parts commonly affected.[1,2,9] This is very important in formulating prevention strategies for these age groups, especially cause–specific prevention.

In our study, pre-school-aged children were more prone to unintentional injury than any other age group. This finding was reported by some studies in Nigeria, the USA, and China.[1,2,6] This is due to their inability to assess and predict

Table 1: Distribution of injuries according to age groups.

Age groups (years) of children	Causes of Unintentional injuries		
	All forms of unintentional road traffic crashes	Unintentional Falls	Non-traumatic unintentional injuries
1–5	6	44	23
6–10	16	38	9
11–15	15	33	11
16–18	1	10	5

dangerous situations. They are also prone to unintentional falls and non-traumatic injuries.

The male gender was more prone to unintentional injury, whether sporting, accident, or domestic injuries. This finding was reported by several studies.[2,10,11] This is due to adventurous behaviour, and they engage in risky activities. Some studies have suggested that female children are less likely to be injured as they get older.[2]

Our study reveals that males are more commonly injured in the lower limb as compared to female children.

There are several preventive measures reported in the literature but we think that they can only reduce the occurrence and mitigate the effects of unintentional injuries.

The limitation of this study is that it is a hospital-based study and the strength is that it shows the location of these injuries.

CONCLUSION

Unintentional injuries commonly affect preschool aged children involving the upper and lower limbs mostly. Injuries result from unintentional falls mostly, and males are commonly injured in the lower limbs.

Preventive measures can only mitigate the effects of these injuries.

Ethical approval

The Health Research and Ethical Committee of University of Calabar Teaching Hospital, Calabar assigned a Protocol number for this study UCTH/HREC/33/408 on 11 January 2023.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Nil.

Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript, and no images were manipulated using AI.

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Karnataka Paediatric Journal

Case Series

A case series of maturity onset diabetes of the young: Searching for a polar bear in a snowstorm

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ABSTRACT

Monogenic diabetes occurs due to a defect in a single gene or a chromosomal locus. The various modes of inheritance occurring include autosomal recessive, autosomal dominant, and non-Mendelian traits or can occur as a spontaneous case secondary to a *de novo* mutation. Although uncommon, monogenic diabetes accounts for 2.5–6% of diabetes in the paediatric age group and usually presents under 25 years of age. Monogenic diabetes can be classified into four major groups: (1) Maturity onset diabetes of the young (MODY), (2) neonatal and early infancy diabetes, (3) monogenic insulin resistance syndromes, and (4) diabetes affiliated with extra-pancreatic features. MODY is the most prevailing form of monogenic diabetes with an autosomal dominant inheritance. There are 14 subtypes of MODY that have been recognised now, and mutations in the genes *hepatic nuclear factor 1-alpha* (*HNF1α*) (MODY-3), glucokinase (MODY-2) *HNF4α* (MODY-1) account for about 95% of all MODY cases. According to a study in the UK, 80% of the MODY cases are misdiagnosed as either Type 1 or Type 2 diabetes mellitus. In our case series, we report 4 cases of MODY and compare their various characteristics. A genetic test was performed for children and adolescents with diabetes mellitus who had a strong history of diabetes in the family and tested negative for islet cell antibodies. Although there have been cases of MODY that have been previously reported worldwide, they usually fall under the types – MODY-3, MODY-2, and MODY-1, whereas we identified one adolescent with Kruppel-like factor-11 gene mutation giving rise to MODY-7 and two adolescents with paired box gene-*4* gene mutation causing MODY-9, both of which contribute to <1% of the cases of MODY reported worldwide.

Keywords: Maturity onset diabetes of the young, Kruppel-like factor-11, Paired box gene-4

INTRODUCTION

Monogenic diabetes occurs due to a defect in a single gene or a chromosomal locus. The various modes of inheritance occurring include autosomal recessive, autosomal dominant, non-Mendelian traits or can occur as a spontaneous case secondary to a *de novo* mutation. The most commonly recognised forms of diabetes mellitus include the polygenic forms of diabetes, such as type 1 or type 2 diabetes. Although uncommon, monogenic diabetes accounts for 2.5–6% of diabetes in the paediatric age group[1] and usually presents under 25 years of age. Monogenic diabetes can be classified into four major groups: (1) Maturity onset diabetes of the young (MODY), (2) neonatal and early infancy diabetes, (3) monogenic insulin resistance syndromes, and (4) diabetes affiliated with extra-pancreatic features.[1] MODY is the most prevailing form of monogenic diabetes with an autosomal dominant inheritance. There are 14 subtypes of MODY that have been recognised now, and mutations in the genes hepatic nuclear

factor 1-alpha (*HNF1α*) (MODY-3), glucokinase (MODY-2) *HNF4α* (MODY-1) account for about 95% of all MODY cases [Table 1].[2] Through this case series, we aim to report rarer forms of MODY and present a comparison of their various characteristics in terms of their presentation and management.

CASE SERIES

Case 1

A 1.6-year-old female child with a history of polyuria, polydipsia for 2 weeks, and lethargy for 1 day presented to the paediatric emergency and was admitted with moderate diabetic ketoacidosis. She was initially diagnosed with type 1 diabetes mellitus (T1DM), started on continuous infusion with insulin, and then shifted to a basal-bolus regimen. As all islet cell antibodies were negative and did not have optimum glycaemic control, a genetic test was done, and she was diagnosed with MODY-1 due to *HNF4α* gene mutation.

Case 2

A 10.9-year-old female adolescent presented to our paediatric endocrinology outpatient department (OPD) with a history of polydipsia and polyphagia for 2 months. She was overweight, and on evaluation, glycated haemoglobin (HbA1C) was 7.9% with negative islet cell antibodies. As she had a strong family history of diabetes, genetic analysis revealed MODY-7 resulting from a Kruppel-like factor-11 (*KLF-11*) gene mutation.

Case 3

A 14.6-year-old male adolescent presented to our paediatric endocrinology OPD with a history of polyuria, polydipsia, and weight loss of 16 kg in 2 months. He was morbidly obese, had an HbA1C of 16.2%, and was initially being treated for type 2 diabetes mellitus (T2DM) with insulin and lifestyle modifications. Islet cell antibodies were negative, and genetic analysis found a paired box gene-4 (*PAX-4)* gene mutation leading to MODY-9.

Case 4

A 17.2-year-old female adolescent had a history of polyphagia and easy fatiguability. With an initial HbA1C of 11.8%, she was initially diagnosed to have T2DM by an adult endocrinologist. Despite being on metformin for a year, she continued to have poor glycaemic control and presented to our OPD. Genetic analysis was sent and she tested positive for MODY-9 attributable to *PAX-4* gene mutation. She was switched to insulin, and her glycaemic control improved.

Table 1: Comparison of various parameters among the four cases of MODY.

Parameter	Case 1	Case 2	Case 3	Case 4
Age at diagnosis	1.6 years	10.9 years	14.6 years	17.2 years
Gender	Female	Female	Male	Female
Clinical presentation	Polyuria, polydipsia for 2 weeks, Lethargy for 1 day	Polydipsia, polyphagia for 2 months	Polyuria, polydipsia, weight loss (16 kg) in 2 months	Polyphagia, Easy fatiguability for 4 months
DKA at presentation	Moderate DKA	No	No	No
Associated conditions	Congenital hypothyroidism	Overweight	Morbid obesity	Nil
Family history of diabetes mellitus	Nil	Father and paternal grandparents: T2DM around 35 years age	Both parents: T2DM around 40 years age	Father: DM since 20 years age-genetic test not done Mother: T2DM at 40 years of age
BMI	Not applicable	21.3 kg/m^2	37.17 kg/m^2	19.44 kg/m^2
HbA1C at diagnosis	7.6%	7.9%	16.2%	11.8%
Initial diagnosis	T1DM	T2DM	T2DM	T2DM
Initial treatment	Insulin: Basal bolus regimen	Lifestyle modifications	Insulin: Basal bolus regimen lifestyle modifications	Metformin lifestyle modifications
Islet autoantibodies	Negative	Negative	Negative	Negative
Gene mutation	*HNF4α*	*KLF-11*	*PAX-4*	*PAX-4*
MODY type	MODY-1	MODY-7	MODY-9	MODY-9
Current treatment	Glimepiride+Insulin	Metformin lifestyle modifications	Insulin: Basal bolus regimen lifestyle modifications	Insulin

DKA: Diabetic ketoacidosis, BMI: Body mass index, HbA1C: Glycated haemoglobin, T1DM: Type 1 diabetes mellitus, T2DM: Type 2 diabetes mellitus, DM: Diabetes mellitus, MODY: Maturity onset diabetes of the young, *KLF-11*: Kruppel-like factor-11, HNF4α: Hepatic nuclear factor 4-alpha, *PAX-4*: Paired box gene-*4*

DISCUSSION

The acronym 'MODY' was coined by Fajans and Tattersall in 1974, who defined it as 'fasting hyperglycaemia.'[3] MODY occurs due to impaired insulin secretion due to a defect in the development of pancreatic islet cells. The genes implicated in MODY hamper insulin secretion by impairment of insulin sensing, glucose metabolism in beta-cells of the pancreas, or activation of adenosine tri-phosphate-dependent potassium channels.[4] According to a study in the UK, 80% of MODY cases are misdiagnosed as one of the polygenic forms of diabetes, either T1DM or T2DM, as they have overlapping genotypic features of both. Vaxillaire and Froguel determined the major diagnostic criteria for MODY to be hyperglycaemia before 25 years of age, autosomal dominant inheritance exhibiting transmission in at least three generations, negative beta-cell autoantibodies, and functional impairment in the pancreatic beta-cells.[5,6] In our case series, we report four cases of MODY and compare their various characteristics. A genetic test was performed for children and adolescents with diabetes mellitus who had a strong history of diabetes in the family and tested negative for islet cell antibodies. Genetic counselling was done in view of the autosomal dominant fashion of inheritance.[6] Management differs among the various types of MODY, where some respond to lifestyle modifications, and some require insulin or oral antidiabetic drugs or a combination of both.[7]

CONCLUSION

Although there have been cases of MODY that have been previously reported worldwide, they usually fall under the types – MODY-3 (30–50% cases), MODY-2 (15–25% cases), and MODY-1 (5%), whereas all the other types of MODY contribute to <1% of the cases individually. In our case series, we identified one adolescent with *KLF-11* gene mutation giving rise to MODY-7 and two adolescents with *PAX-4* gene mutation causing MODY-9, both of which contribute to <1% of the cases of MODY reported worldwide. As MODY shares features of the more commonly encountered forms of diabetes, such as T1DM and T2DM, we often miss identifying MODY. Hence, the phrase 'searching for a polar bear in a snowstorm' aptly applies to identifying MODY cases among the numerous cases of type 1 and type 2 diabetes.

Authors' contributions

Both authors have contributed to compiling the data.

Ethical approval

The Institutional Review Board approval is not required.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

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Karnataka Paediatric Journal

Case Report

Presentation of lymphoedema praecox as a septic shock with acute renal failure in an adolescent girl: A case report

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ABSTRACT

Lymphoedema praecox (Meige disease) is a rare chronic disease of disordered lymphatic transport in which 10% of the cases present as non-inherited mutations responsible for defective lymphatic development. The inherent complex inflammatory pathways lead to defective lymphangiogenesis, oedema, adipose tissue deposition and chronic fibrosis. Various factors lead to local impairment of adaptive immunity leading to the increased incidence of bacterial infections. Sepsis and septic shock arising from such infection can be life-threatening. One such case is discussed where a post-pubertal adolescent girl presenting with a chronic painless unilateral limb swelling landed into cellulitis and gangrene with septic shock. She needed care in an intensive care setting and a diagnostic work-up was started to look into the cause. She recovered from this complication and care for this chronic condition was continued later. A high index of suspicion for the occurrence of this rare entity with its associated complications is the prerequisite to a successful outcome.

Keywords: Lymphoedema, Late onset, Hereditary, Meige lymphoedema

INTRODUCTION

Lymphoedema, a chronic disease of a disordered lymphatic transport, consists of primary lymphedema, which is rare (incidence 1.15 in 100,000 in age <20y population) and is differentiated into hereditary Type 1 (Milroy disease), Type 2 (lymphoedema praecox or Meige disease) and lymphoedema tarda. Primary lymphoedema almost always affects the paediatric population; adult-onset disease is uncommon. Boys are more likely to present in infancy, while girls commonly develop the disease in adolescence. The primary disease affects the lower extremities in 93% of cases. Lymphoedema praecox is seen commonly in post-pubertal adolescent girls with involvement of the left lower limb as the common site.

Patients may present with swelling over the limbs, paraesthesias and poor healing of wounds. The assumed hypothesis includes the local proliferation of T regulatory cells causing impairment of adaptive immunity.[1] This, along with the loss of dendritic cell function and a breach in the epidermal-dermal layer, provides entry for the infection.[2] It may present with systemic signs of high fever and rigors and local signs of erythema, pain, warmth and swelling. Beta-haemolytic streptococci can cause local cellulitis, lymphangitis and sometimes gangrene. Septic shock may ensue, if not suspected and treated in time and can be life-threatening. This

case report stresses the need for a timely diagnosis of this rare disorder of the lymphatic system and anticipation of the associated complication of bacterial sepsis which may give rise to a catastrophe like acute renal failure. It may also sensitise the treating physician about the need for long-term care of this chronic condition and about the education of the patient and the family for care at home. They are also taught about seeking help at any possibility of complications.

CASE REPORT

We have discussed a case of a 16-year-old post-pubertal girl from a rural background. Her presenting complaints were fever, swelling, pain and ulceration involving the dorsum of the left foot over 4 days. She also started vomiting and passing loose stools. She presented to the emergency department in septic shock and acute renal failure. Her parents gave an account of her having similar complaints around 2 years back with swelling over her left foot and ankle and needing hospitalisation for 8 days. She recovered from the illness with the treatment but painless left leg swelling persisted for 2 years and discomfort during walking continued. She was worried about the discomfort while walking and doing daily chores, particularly changing clothes. No specific help or any treatment was sought for this purpose and failed to follow up with the hospital where she was previously treated. Her health seemed normal except for the persistent painless leg swelling. Her menstrual history revealed that she attained menarche at 12 years but had irregular cycles and sometimes heavy and prolonged menstrual bleeding. She was born of a non-consanguineous marriage. Her family history did not reveal any significant illness.

Figure 1: Non-haemorrhagic blister over the dorsum of the left foot and limb oedema with skin gangrene over the great toe and adjacent toes.

Her physical examination showed a non-haemorrhagic blister over the dorsum of the left foot and gangrenous skin changes over the great toe and the adjacent toes [Figure 1]. There was non-pitting limb oedema and erythema to the middle third of the leg. Stemmer's sign was positive which was elicited by pinching the skin over the dorsum of the base of the second toe.[3]

Her emergency care in Intensive Care Unit started with the management of septic shock with fluid support, inotropes and antibiotics. She responded to the treatment with recovery from shock and acute renal failure. Ulceration and gangrene were treated simultaneously with the local care of the wound with regular dressing and wound debridement.

Her deranged laboratory parameters were anaemia and raised inflammatory markers and raised serum creatinine. She underwent Doppler of the limb which showed pan reversal of diastolic flow in the left lower limb arteries possibly secondary to systemic infection. Ultrasound of the abdomen showed medical renal disease and echocardiogram was normal. Computed tomography angiography of the limb did not show changes of vasculitis. Her antinuclear antibody was negative. Lymphoscintigraphy could not be done due to a lack of expertise and experience in doing the test.

During recovery, measures were taken like giving compression dressing to the affected limb and leg elevation to decrease limb oedema.[4] She was counselled about the care of the limb with the use of elastic garments[5] and family members asked to keep a watch on new infections, if any.

DISCUSSION

The diagnosis, in this case, is primary lymphoedema looking at the age at presentation in a post-pubertal adolescent girl, the chronicity of painless limb swelling and no risk factors for chronic venous insufficiency. The unilateral firm, sclerotic skin with non-pitting oedema and a positive Stemmer's sign ruled out other diagnoses clinically. Stemmer's sign is more sensitive than specific. If the test is positive, it is likely that the patient has lymphoedema. The staging was done as Stage 3 in view of fibroadipose tissue deposition and the associated skin changes. Causes of secondary type such as filariasis, trauma and malignancy were ruled out by relevant investigations.[6] Secondary lymphoedema is uncommon in children but is responsible for the disease in 99% of adults. This case report highlights the possibility of this rare non-inherited disorder in an adolescent female wherein the primary symptoms like swelling may not prompt the patient to seek medical help. Furthermore, this condition may be missed by the attending physician due to the probability of being unaware about this condition. Children with suspected primary lymphoedema need to be examined for syndromic characteristics that are associated

with the disease. The catastrophe of sepsis and associated poor outcome can be avoided if dealt properly with this entity. For a favourable outcome in such a rare condition with the associated complication of sepsis, a high index of suspicion, gained from knowledge and awareness on the part of the treating physician is highly required. This case report may sensitise the paediatrician to deal with such a catastrophe of this intriguing disorder.

CONCLUSION

Lessons learnt are:

1. Lymphoedema is to be differentiated from other types of oedema clinically and staging is also possible with the help of a positive Stemmer's sign
2. Primary lymphoedema is a clinical diagnosis made only after ruling out other possible causes of lymphoedema
3. A high index for sepsis as a possible complication should be kept to prevent a catastrophe.

Ethical approval

The research/study complied with the Helsinki Declaration of 1964.

Declaration of patient consent

Patient's consent not required as patient's identity is not disclosed or compromised.

Financial support and sponsorship

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Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The author(s) confirms that there was no use of Artificial Intelligence (AI)-Assisted Technology for assisting in the writing or editing of the manuscript and no images were manipulated using the AI.

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Karnataka Paediatric Journal

Case Report

A case of congenital multiple pancreatic cysts in a female child: A case report

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ABSTRACT

Congenital pancreatic cysts are rare in the paediatric age group; they can be isolated or associated with other malformations. Prompt diagnosis and appropriate management are crucial to prevent complications. One and a half year old female presented with an abdominal mass. Surgical exploration revealed multiple cystic lesions occupying most of the abdominal and pelvic cavities originating from the pancreas. A complete surgical excision was done, and the histopathological examination confirmed the presence of islands of well-defined pancreatic tissue with scattered dilated ducts. True congenital pancreatic cysts are rare anomalies to occur, and there is female predominance for such conditions; early diagnosis and treatment are essential to prevent complications.

Keywords: Congenital cysts, Pancreatic cysts, True cysts

INTRODUCTION

Congenital pancreatic cysts are rarely reported in paediatric age group, and they can be an isolated condition or associated with other malformations.[1] Prompt diagnosis and appropriate management are crucial to prevent complications.[2] In our case report, this female baby has multiple pancreatic cysts as isolated anomalies managed by complete surgical excision. We think that the results of this study will be of utmost usefulness for the paediatric surgeon when considering congenital multiple pancreatic cysts in children.

CASE REPORT

One and a half years old baby female was referred to the paediatric surgery department in Child Welfare Teaching Hospital, complaining of abdominal distension for four months, which gradually increased in severity and was associated with decreased appetite and weight loss, but without vomiting or alteration in bowel habit. The patient had no history of fever or changes in sleep patterns. The child had no previous medical or surgical illnesses or admission to hospitals. Family history had nothing significant. During the examination, the baby was conscious, underweight, and pale, with moderate abdominal distension. A palpable abdominal mass was noticed on the right side of the abdomen; it was spherical mobile of about 3 × 4 cm in size.

A complete blood count revealed leukocytosis with normal haemoglobin, platelets, and morphology. All biochemistry investigations serum electrolytes, renal function, serum amylase and lipase, erythrocyte sedimentation rate [ESR], and alpha-fetoprotein were normal levels. Erect abdominal

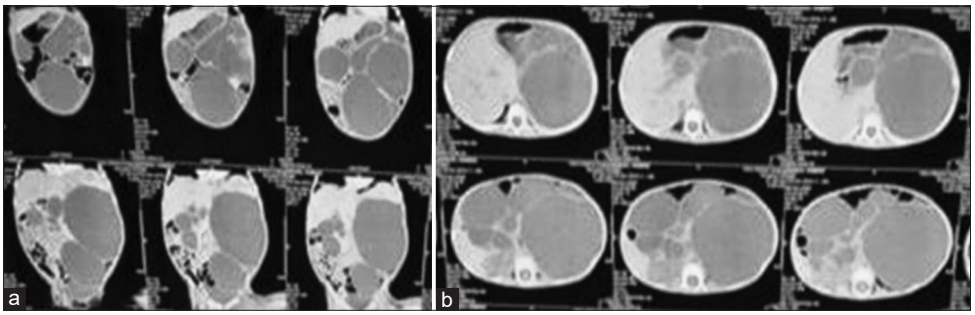

Figure 1: (a and b) Computed tomography scan of the child showing multiple abdominal cysts.

X-ray shows an abnormal soft-tissue shadow. Abdominal ultrasound showed a multiloculated cystic lesion in the entire abdomen and mainly the left upper abdominal quadrant, not related to any organ. Abdominal computed tomography scan with intravenous contrast showed multiloculated cystic lesions extended from the abdominal to the pelvic cavity, with a size of about 20 × 17 × 11 cm, with pressure effect on the abdominal and pelvic organs. There are no solid components and no significant enhancement [Figures 1a and b].

Intra-operative findings show multiple pancreatic cysts of variable sizes. After delicate dissection and release of adhesion, the cysts were excised completely [Figures 2a and b, 3]. The cysts were sent for histopathological examination, which revealed multiple unilocular cysts with a wall thickness of 0.1 cm containing clear serous fluid. Cystic structures are lined by cuboidal and columnar glandular epithelium with thick fibrous walls containing islands of well-defined unremarkable pancreatic tissue and scattered with dilated ducts. No atypia or immature tissue could be seen. Picture consistent with true benign pancreatic cysts is shown in Figure 4. Post-operative follow-up for two years by clinical and abdominal ultrasound examination showed complete excision of cysts without recurrence. Written informed consent has been taken from the patient's parent for publication. The Medical Ethics Committee of the University of Anbar, Anbar, Iraq, approved this study.

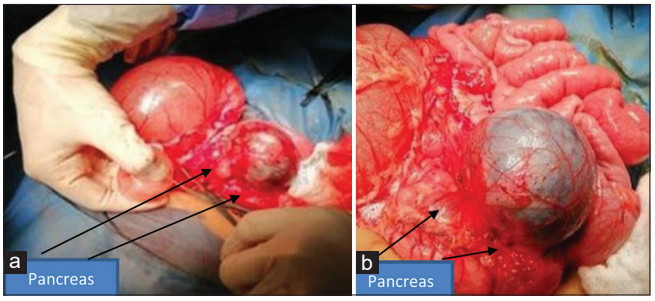

Figure 2: (a and b) Operative picture shows the pancreas with multiple pancreatic cysts.

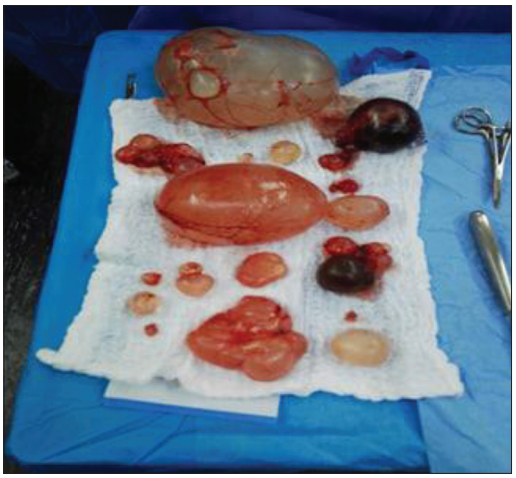

Figure 3: Post-operative pancreatic cysts after excision with variable sizes.

DISCUSSION

Congenital pancreatic cysts in the paediatric age groups are rare. The incidence is <1% of all abdominal cysts.[1,2] Embryologically, true pancreatic cysts occur as a result of developmental anomalies related to the sequestration of primitive pancreatic ducts.[3] These cysts can be single or multiple. Congenital pancreatic cysts may or may not possess an epithelial lining or elevated serum amylase level; the presence of either supports the diagnosis.[3,4] There is female predominance for such conditions as in our case.[1]

Such pathology had been reported to be associated with other anomalies such as von Hippel-Lindau syndrome, polycystic kidney disease, Ivemark syndrome, and other conditions.[3,5,6] diagnosis can be made antenatally or postnatally, but prenatal diagnosis is extremely rare.[1,7,8] Patient may be asymptomatic or have abdominal mass with or without abdominal distension. Pressure symptoms are vomiting and jaundice due to compression on the stomach and biliary tree, respectively.[9,10]

Early diagnosis and treatments are essential to prevent the development of complications such as infection, cyst rupture, peritonitis, and pancreatitis.[4] The latter results either from congenital anomalies of the duct system or from visceral compression by the cyst.[3,11] It may be difficult to

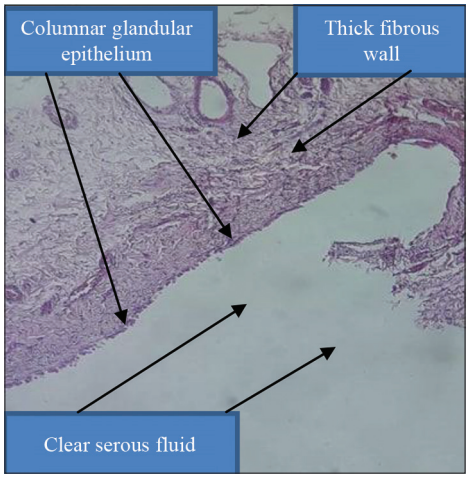

Figure 4: Histopathological finding of pancreatic cyst.

establish the diagnosis by depending on imaging studies, and the differential diagnosis may include choledochal cyst, lymphatic malformations, omental or mesenteric cyst or enteral duplications anomalies. Endoscopic retrograde cholangiopancreatography is useful to rule out communication with the pancreatic duct and biliary tree.[6]

CONCLUSION

We observed that the true congenital pancreatic cysts are rare anomalies with female predominance. Early diagnosis and treatment are essential to prevent complications.

Ethical approval

This research/study was approved by the Ethics Committee of the University of Anbar, and the reference number 211 dated 7th September 2023.

Declaration of patient consent

Patient's consent is not required as the patient's identity is not disclosed or compromised.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript, and no images were manipulated using AI.

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Karnataka Paediatric Journal

Case Report

A nutritional Vitamin B12 deficiency (infantile tremor syndrome) presenting as hemiconvulsion hemiplegia epilepsy syndrome

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ABSTRACT

Hemiplegia Hemiconvulsion Epilepsy syndrome (HHE syndrome) is a condition characterized by the acute onset of unilateral seizures, which progress to cortical atrophy, hemiplegia and later develop epilepsy over weeks to years. Various aetiologies include infectious causes, especially meningitis, encephalitis, trauma, vascular insults, and sometimes idiopathic. Here, we present a 13-month-old female child, first in birth order, born to a non-consanguineous marriage, who presented with mild global developmental delay with acute onset right focal seizures followed by right-sided weakness and drug-resistant epilepsy. The child was found to have clinical and biochemical features of vitamin B12 deficiency with right-sided weakness. MRI of the brain initially showed edema of the left cerebral hemisphere and later atrophy on the left side. The electroencephalogram showed low voltage activity over the left cerebral hemisphere. Post-treatment with cobalamin, improvement in development was noted, with partial improvement in weakness and persistent epilepsy. In the current report, we highlight Hemiplegia Hemiconvulsion Epilepsy (HHE) syndrome on the background of nutritional vitamin B12 deficiency.

Keywords: Hemiconvulsion hemiplegia epilepsy syndrome, Nutritional Vitamin B12, Infantile tremor syndrome, Cerebral atrophy, Hemiplegia

INTRODUCTION

Hemiplegia Hemiconvulsion Epilepsy (HHE) syndrome is a condition in which the patient develops unilateral seizures followed by ipsilateral cerebral oedema progressing to cortical atrophy which manifests as hemiplegia and finally develops epilepsy over weeks to months of varying frequency.[1] The various aetiologies include hypercoagulable states such as protein C/S deficiency, factor V deficiency, metabolic disorders like L2 hydroxy glutaric aciduria, infectious agents like influenza, human immunodeficiency virus, hypoxia, ischemia, toxic agents like theophylline and sometimes unknown cause.[2-4] The main pathophysiology behind the syndrome involves any aetiology which causes prolonged seizures that lead to histotoxic damage of the parenchyma of the brain.[5] There is a complex interplay between genetic, infectious and environmental agents to precipitate HHE syndrome.[4] Here, we present an unusual case of HHE syndrome with nutritional Vitamin B12 deficiency without any apparent aetiology.

CASE REPORT

A 13-month-old female child born to non-consanguineous marriage presented with loose stools, vomiting and fever since the past 5 days followed by right focal status epilepticus. On day 2 post seizures, the right-sided upper and lower limb weakness was noted, in the background of normal birth history and mild developmental delay. At 13 months of age, the child could sit but could not walk, was able to respond to name inconsistently and could not speak any meaningful words. Child was on predominant breast feeds with the mother's diet primary vegetarian and initiated on complementary feeds by 8 months. There was a history of vomiting episodes with decreased acceptance of feeds. On examination, Weight of 7.5 kg (−1 to −2SD), height of 76 cm (0 to −1SD), head circumference of 44 cm (−1 to −2SD), mid-arm circumference of 11.5 cm was noted. There was hypopigmented hair and knuckle hyperpigmentation noted [Figure 1]. Neurological examination revealed Glasgow coma scale: 9/15, right-sided upper motor facial palsy, hypotonia on the right side with power of 2/5 on Medical Research Council (MRC) grade [Figure 1a] and extensor plantar on the right side along with tremors.

Figure 1: Clinical photograph of child before discharge (a) shows hypopigmented hair, pallor, knuckle hyperpigmentation, mild right-sided facial weakness with the right upper weakness at the time of discharge – 12 months of age. During follow-up (b) shows weakness of the right upper limb with wasting and increased tone during follow-up at 4 years of age.

Investigations showed haemoglobin of 8.6 g/dL, mean corpuscular volume (MCV) of 106.7, with normal total leukocyte and platelet counts and megaloblastic anaemia in the peripheral smear with low Vitamin B12 level of 120 pg/mL (normal 180–300 pg/mL) and increased levels of homocysteine of 79.83 µmol/L (normal 3–15 µmol/L). The renal function tests, tandem mass spectrometry for inborn errors of metabolism, cerebrospinal fluid (CSF) analysis, fasting ammonia, lactate and arterial blood gas done were normal. Electroencephalogram (EEG) showed asymmetric background activity with low voltage over the left cerebral hemisphere. Magnetic resonance imaging (MRI) brain shows diffuse oedema of the left hemisphere, corpus callosum, left internal capsule and bilateral globus pallidus regions appearing hyperintense on axial diffusion-weighted imaging [Figure 2a and b] and hyperintense on fluid-attenuated inversion recovery (FLAIR) [Figure 2c] sequences. Magnetic resonance angiography [Figure 3a] shows no obstruction or narrowing of vessels. Magnetic resonance venogram [Figure 3b] shows a reduced number of vessels and flow in the left cerebral hemisphere. Mother investigations revealed haemoglobin of 9.5 g/dL, MCV of 101.7, with normal total leukocyte and platelet counts and megaloblastic anaemia in the peripheral smear with low Vitamin B12 level of 145 pg/mL (normal 180–300 pg/mL) and increased levels of homocysteine of 49.83 µmol/L (normal 3–15 µmol/L). The whole exome sequencing and mitochondrial genome sequencing did not reveal any pathogenic variant.

A diagnosis of HHE syndrome due to nutritional Vitamin B12 deficiency was made. Child was treated with injectable

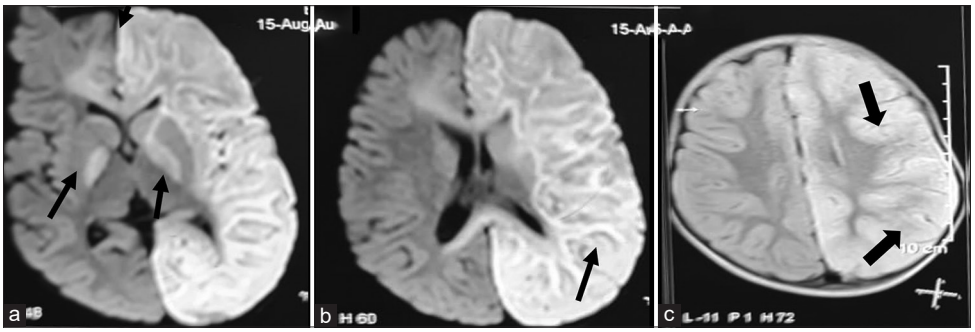

Figure 2: (a and b) Magnetic resonance imaging brain shows diffuse oedema of the left hemisphere, corpus callosum, left internal capsule and bilateral globus pallidus regions appearing hyperintense (black arrows) on axial diffusion-weighted imaging and hyperintense (black arrows) on fluid-attenuated inversion recovery (c) sequences on 7th day at 13 months of age when admitted to the hospital (black arrows).

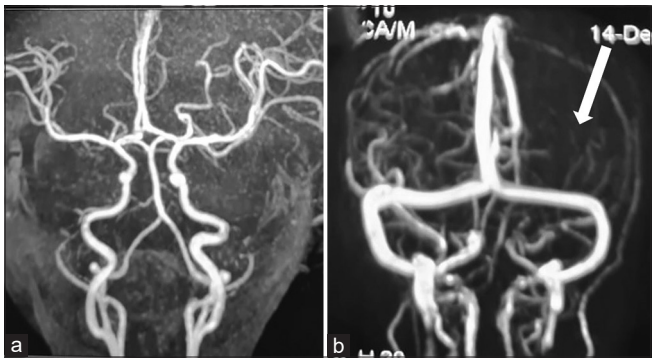

Figure 3: Magnetic resonance angiography (a) shows no obstruction or narrowing of vessels. Magnetic resonance venogram (b) shows reduced number of vessels and flow in the left cerebral hemisphere (thick white arrow) on the 7th day at 13 months of age when admitted to the hospital.

cobalamin for 14 days, folic acid and other supplements with marked improvement in development on follow-up. After 2 weeks of treatment, investigations revealed haemoglobin of 9.6 g/dL, Vitamin B12 level of 859 pg/mL (normal 180–300 pg/mL) and homocysteine of 9.83 µmol/L (normal 3–15 µmol/L). The weakness of the right side improved partially and developed stiffness of the right upper limb [Figure 1b] and right lower limb. Repeat MRI brain at 4 years showed left hemisphere atrophy [Figure 4a] with no restricted diffusion. Diffuse gliotic areas in the left cerebral subcortical white matter appearing hyperintense on axial FLAIR [Figure 4b] and hypointense on T1-weighted sequences [Figure 4c]. EEG of child done at 4 year of age showed low voltage over the left cerebral hemisphere [Figure 5]. Child was on regular follow-up with dietary advice of Vitamin B12 rich diet without any Vitamin B12

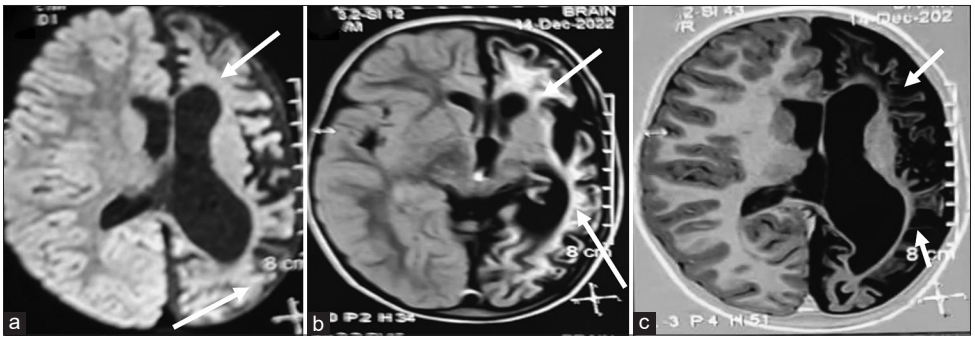

Figure 4: (a-c) Repeat magnetic resonance imaging brain at 4 years. Axial diffusion weighted sequence showing left hemisphere atrophy (thick white arrows) with no restricted diffusion. Diffuse gliotic areas in the left cerebral subcortical white matter appearing hyperintense on axial fluid-attenuated inversion recovery and hypointense (thin white arrows) on T1-weighted sequences.

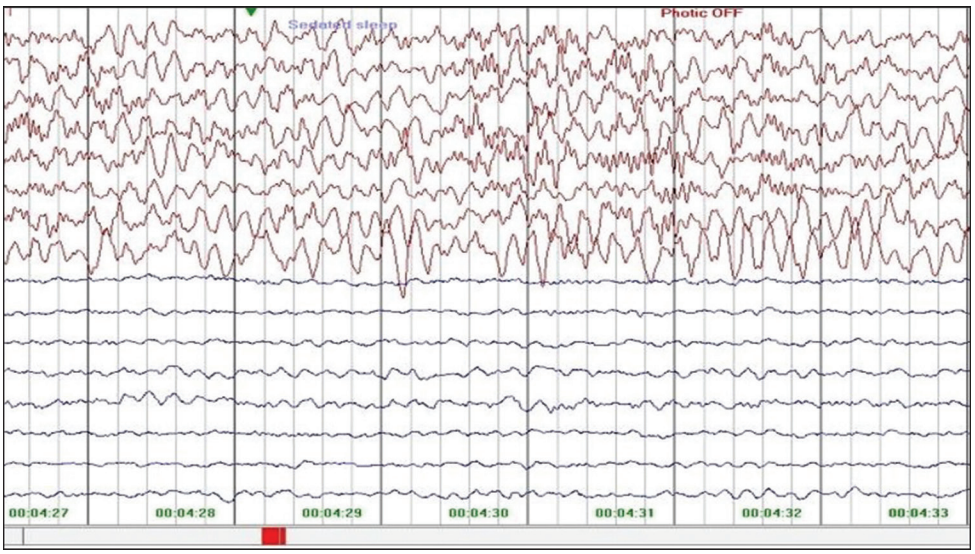

Figure 5: Electroencephalogram of child done at 4 years of age showing low voltage over the left cerebral hemisphere during follow-up at 4 years of age.

supplements and serum Vitamin B12 and homocysteine levels were within normal limits over the 4 years follow-up period.

DISCUSSION

The differential diagnosis considered were HHE syndrome, Rasmussen's encephalitis, infectious aetiologies such as meningitis and encephalitis and cerebrovascular accidents such as cerebral venous thrombosis secondary to acute gastroenteritis induced dehydration. CSF analysis and MRA done were normal, ruling out neuroinfections and stroke. The patient had clinical features and biochemical evidence of Vitamin B12 deficiency; hence, nutritional Vitamin B12 deficiency was considered as a potential aetiology. Rasmussen's encephalitis presents with insidious onset of focal seizures such as epilepsia partialis continua, followed by ipsilateral weakness over a period of months and years with MRI of the brain showing atrophy involving caudate nucleus, and later temporal lobe. HEE children present with focal status followed by weakness maximum at onset later other type of seizures and MRI initially shows contralateral hemispheric oedema involving all lobes followed by hemispheric atrophy of all lobes; hence, our case features are suggestive of HHE, rather than Rasmussen's encephalitis.

Methyl cobalamin is an active form of Vitamin B12 required for maintenance of myelin in the nervous system and deficiency usually leads to myelin disturbances. The neurons of the cerebral cortex with myelin disruption may be prone to glutamate excitotoxicity which could be a reason for seizures.[6] Furthermore, in the absence of cobalamin, homocysteine cannot be converted to methionine which leads to the elevation of the former. Under experimental conditions, high doses of homocysteine have induced convulsive status in animals.[7] In the current case, the patient had elevated homocysteine, low Vitamin B12 which could be the cause for prolonged seizures and the possible aetiology of Vitamin B12 deficiency seems to be nutritional in origin.

The usual course of HHE syndrome is prolonged unilateral seizures of clonic type which leads to brain oedema in acute stages. Initially, the syndrome has only HH component with E component of the disease developing over weeks to months. The neuroimaging shows cerebral atrophy after resolution of oedema.[8] The outcomes are variable with temporal lobe epilepsy in idiopathic and generalised seizures in symptomatic type after a long symptom free period of weeks to months. Many patients have variable degrees of motor, cognitive and language impairment.[9]

The current case showed clinical and laboratory features of nutritional Vitamin B12 deficiency, features of HHE syndrome as focal status seizures followed by ipsilateral weakness and later epilepsy. We ruled out other causes for weakness based on MRI and MRA of the brain, metabolic and genetic causes based on negative metabolic workup and genetic workup. Myers *et al*. reported a 4-year-old female with Cobalamin C disease due to homozygous *MMACHC* mutation who presented with features of HHE syndrome.[10] However, there are no reports which link nutritional Vitamin B12 deficiency to HHE syndrome. This is a rare association.

CONCLUSION

Nutritional Vitamin B12 deficiency should be considered as the risk factor of HHE syndrome as it would have an impact on the treatment and prognosis and needs further studying.

Ethical approval

The Institutional Review Board approval is not required.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

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Karnataka Paediatric Journal

Case Report

Paraesophageal hernia mimicking pneumatocele in an infant: A diagnostic dilemma

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ABSTRACT

A defect in the oesophageal hiatus can result in the herniation of the stomach or other abdominal organs into the thoracic cavity, known as a hiatal hernia. These hernias are uncommon in infants and children, and their symptoms can be vague and non-specific, posing challenges for even experienced clinicians to make a diagnosis. Regardless of the case, surgical intervention is necessary due to the potentially life-threatening complications associated with this condition. We present a rare case of a 2-month-old infant with congenital paraesophageal hernia (PEH) who initially came to our hospital with a diagnosis of pneumonia with pneumatocele. Based on clinical presentation and radiological examinations, the infant was correctly diagnosed with PEH and gastric volvulus. The patient underwent emergency surgical correction and was discharged in a stable condition. It is crucial to have a thorough understanding of the clinical presentation and maintain a high level of suspicion to ensure timely diagnosis and appropriate management in these cases.

Keywords: Children, Pneumonia, Pneumatocele, Paraesophageal hernia, Hiatus hernia

INTRODUCTION

Hiatus hernia is a condition that involves the herniation of the contents of the abdominal cavity, most commonly the stomach, through the diaphragm into the mediastinum.[1] It is more common in adults and uncommon in the paediatric age group, as most cases are caused by acquired age-related laxity of structures supporting the gastroesophageal junction (GEJ). These cases can present with non-specific symptoms such as vomiting, recurrent respiratory system infections or failure to thrive. However, sometimes, they can have an acute clinical presentation with respiratory distress or gastric volvulus.[2,3] As the stomach herniates into the chest, it can be mistaken for cystic lung lesions on radiological images. We present a case of an infant initially misdiagnosed as pneumonia with pneumatocele but later determined to be a para esophageal hernia (PEH).

CASE REPORT

A 2-month-old female child was referred to our hospital with complaints of fever, cough, cold, increased work of breathing and regurgitation of feeds for 12 days. The child had been admitted to a local hospital for seven days, where a chest X-ray and high-resolution computed tomography (HRCT) thorax were performed. The X-ray showed consolidation in the left lung around a pneumatocele, while the HRCT report suggested a small area of consolidation

in the right upper lobe of the lung with a pneumatocele, consolidation in the left lower lung and mild left pleural effusion. The child had received intravenous cefotaxime, amikacin and dexamethasone for seven days at the local centre, but the symptoms did not significantly improve, leading to the referral to our centre. The reason for starting on dexamethasone was unclear from the discharge summary.

The child was a full-term, vaginally delivered baby with a birth weight of 2.7 kg. She had a history of 5-day neonatal intensive care unit admission for neonatal jaundice starting from day 2 of life. The child has received appropriate immunisations up to the present. On admission to our hospital, the child appeared active and awake but pale, with a barrel-shaped chest and tachypnoea (respiratory rate: 54/min). Intercostal retractions were prominent, and decreased air entry was noted on the left side of the lung. In addition, an ejection systolic murmur was heard. Due to respiratory distress, the child was started on high-flow nasal cannula therapy, along with empirical antibiotics and intravenous cloxacillin. Initial investigations revealed a haemoglobin level of 10.5 g/dL, total leucocyte count of 20,800, platelet count of 8,900, C-reactive protein level of 0.5 mg/L and thyroid-stimulating hormone level of 1.28 mIU/L. Chest X-ray, taken with an AP view and lateral view using an orogastric tube, showed a large air bubble behind the heart in the left thorax, the absence of a stomach gas bubble and the orogastric tube ending in the left chest [Figure 1a and b]. A 2D echocardiogram suggested a patent foramen ovale with moderate pulmonary arterial hypertension. An ultrasound of the chest revealed a suspicious air bronchogram in the left thorax, while neuro sonography and abdominal ultrasound detected no abnormalities. A repeat HRCT was performed due to unclear images from the previous scans, and it revealed a PEH with herniation of the GEJ, stomach and pylorus into the left hemithorax, causing collapse and consolidatory changes in the lower lobe of the left lung [Figure 2]. The stomach was rotated along its long axis, with the antrum in an anterosuperior location and the fundus in a posteroinferior location. The mediastinum, trachea, bronchi and cardia were shifted to the right side. A contrast study showed contrast material entering the postero-inferior location, followed by the antero-superior location, suggesting gastric volvulus. A diagnosis of the para esophageal type of hiatal hernia with gastric volvulus was made. The child was transferred to paediatric surgery, and surgical repair was performed. She was discharged in a haemodynamically stable condition after ten days.

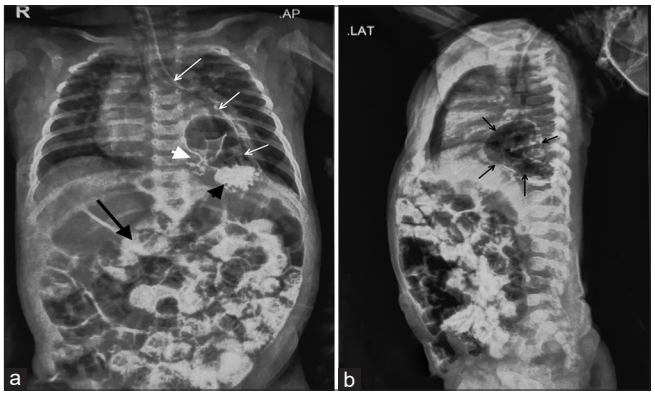

Figure 1: (a) A chest X-ray AP view post-contrast. Stomach bubble is absent in the abdomen. White arrows trace the orogastric tube, which ends in the left thorax. Contrast delineates the anatomy of the stomach, which is present in the thorax. The black arrowhead points to the contrast material in the stomach fungus. White arrowhead points to the pylorus of the stomach. Contrast can also be seen in the intestine (black arrow). (b) A chest X-ray lateral view post-contrast. The black arrow points to the stomach, which is posterior to the heart shadow. Contrast can also be seen in the intestine.

DISCUSSION

Hiatal hernias are of four types: Type I, widely referred to as sliding hernia, occurs when only the gastric cardia protrudes through the oesophageal hiatus and is more common (95% cases of hiatus hernia), and TypesII–IV are known as PEHs and are less common (5% cases of hiatus hernia). Type II occurs when the gastric fundus herniates through the hiatus, but the GEJ remains fixed in position. Type III is a combination of the previous two types in which the GEJ has an abnormal intrathoracic position, and the gastric fundus protrudes into the thoracic cavity. Type IV is the PEH that involves herniation of other abdominal organs such as the colon, small intestine, omentum or spleen.[2,4,5] Due to herniation of the stomach in the thoracic cavity, PEH diagnosis can be confused with cystic lung lesions. In our case, it was confused with pneumatocele. Paraesophageal hernia may present with respiratory distress, especially in Types III and IV[3], as in our case.

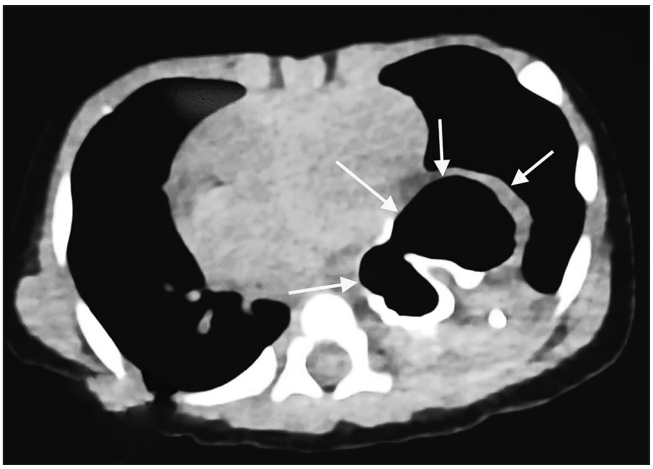

Figure 2: High-resolution computed tomography chest showing the stomach (marked by white arrows) behind the heart shadow.

The first clue to diagnosis can be chest X-ray AP and lateral view. Chest X-ray may show an absence of gastric bubbles in the stomach and the presence of a well-defined radiolucent shadow in the thorax. The orogastric tube tip may be ending in the thorax. These findings were present in the chest X-ray of our case. Contrast studies are helpful in delineating the content of the herniating organs. Computed tomography can be used to delineate the organ and vascular anatomy. PEH must be differentiated from other cystic lung legions.

Other differential diagnoses include large sliding hiatus hernia, lung abscess, congenital lung cysts, hydatid disease, pericardial cysts, foregut duplication cysts, contained perforations and epiphrenic diverticulum.[6]

Underlying surgical causes should be suspected in neonates or infants with certain scenarios such as persistent or recurrent pneumonia, unilateral pneumonia, absence of response to antibiotics and associated symptoms or a family history of congenital lung disease. An infant or neonate having persistent or recurrent pneumonia with normal immunity, despite appropriate treatment, may raise suspicion for an underlying surgical cause.

Unilateral pneumonia can sometimes be associated with underlying surgical causes. This could include conditions such as lung malformations, lung cysts or bronchial abnormalities.

If an infant or neonate does not show improvement or fails to respond to appropriate antibiotic therapy for pneumonia, it may indicate an underlying surgical cause.

Certain symptoms, such as persistent cough, noisy breathing, respiratory distress, feeding difficulties, failure to thrive or other congenital anomalies that may accompany pneumonia may raise suspicion for underlying surgical causes. A family history of congenital lung or respiratory conditions may increase the likelihood of underlying surgical causes, for example, congenital pulmonary airway malformation. When in doubt, prompt imaging such as chest X-ray, HRCT, bronchoscopy, and barium study will help in the diagnosis of underlying surgical causes.

Potentially life-threatening complications of PEHs include gastric volvulus, bleeding, incarceration, obstruction and gangrene or perforation.[7] Our case presented with complications of gastric volvulus. Surgery is mandatory to treat PEH, even if it is asymptomatic or diagnosed incidentally because if PEH is left untreated, it is prone to the above-cited life-threatening complications.

CONCLUSION

PEH in children is a relatively uncommon entity. It can present with symptoms of recurrent pneumonia. The non-specific symptomatology of PEH can lead to delay or miss diagnosis, leading to diagnostic dilemma and delayed management or mismanagement. Physicians caring for these patients should be aware of such underlying surgical causes predisposing or presenting as pneumonia in children. Prompt diagnosis and surgical correction immediately after the diagnosis are essential to ensure good outcomes for these children.

Ethical approval

Institutional Review Board approval is not required.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Nil.

Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript, and no images were manipulated using AI.

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Karnataka Paediatric Journal

Letter to the Editor

Eschar in scrub typhus

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Quick Response Code:

Dear Editor,

Rickettsial diseases are caused by a variety of obligate intracellular, Gram-negative bacteria from the genera *Rickettsia, Orientia, Ehrlichia and Anaplasma*.[1] Rickettsial infections are underdiagnosed in India due to their non-specific clinical presentation.[2] The presence of an eschar, less commonly observed in the Indian population, contributes to the diagnostic process. Therefore, we are sharing this image to create more awareness of this treatable life-threatening condition.

A 2-year-old female child presented with a fever lasting seven days and seizures on day 6 of fever. On examination, vital signs were stable, revealing three eschars: One over the mid back adjacent to the medial border of the left scapula [Figure 1a], over the back below the right scapular region [Figure 1b] and the third over the left mastoid region [Figure 1c]. On systemic examination, a Glasgow Coma Scale of 5/15, meningeal irritations, brisk deep tendon reflexes and hepatosplenomegaly were noted. Investigations revealed a total leukocyte count of 18700/mm^3, haemoglobin of 10.7g/dL, platelet count of 44000/mm^3, total protein of 4.2 g/dL, serum albumin of 1.8g/dL, aspartate aminotransferase of 279.3U/L, alanine aminotransferase of 123.1U/L, serum urea of 122.9 mg/dL and serum creatinine of 0.86 mg/dL. The Weil Felix test was positive for OXK (1:320). Serum and cerebrospinal fluid immunoglobulin M enzyme-linked immunosorbent assay were positive for *Orientia tsutsugamushi*. Work-up for other infections, such as dengue and malaria, was negative. The child was treated symptomatically and with oral

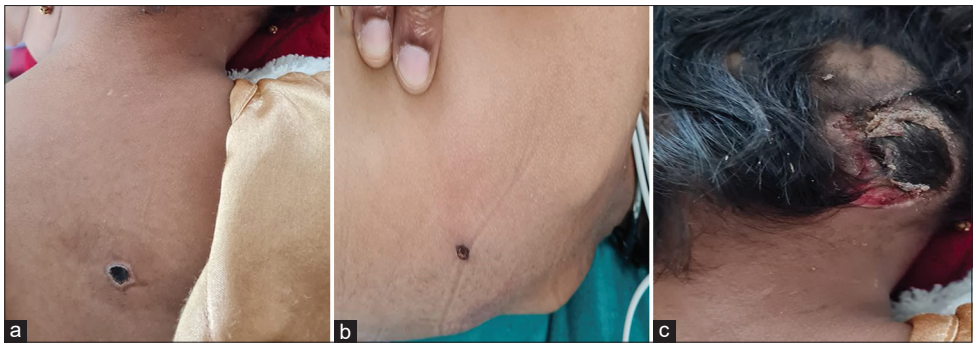

Figure 1: (a) An eschar over the mid back adjacent to the medial border of the left scapula with a black adherent crust surrounded by an erythematous halo, (b) an eschar over the back below the right scapular region with a characteristic black crust and (c) an eschar over the left mastoid region with a black necrotic crust.

doxycycline for seven days, leading to improvement in the child's condition. Rickettsial eschars are necrotic lesions that occur at the site of tick or mite bites and represent locations of primary inoculation of spotted fever group *Rickettsia* and *Orientia* species.[3] A rickettsial eschar begins as a small, painless papule that appears within a few days after the bite of an infected vector. The papule grows, becomes vesicular or pustular and ulcerates, forming a brown-to-black crust surrounded by a red annular halo. The presence of an eschar can aid in the clinical and epidemiologic differentiation of less severe spotted fever rickettsioses from the more severe Rocky Mountain spotted fever. Rickettsial eschars serve as an important clinical specimen.[3]

Ethical approval

The Institutional Review Board approval is not required.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

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Karnataka Paediatric Journal

Letter to the Editor

Leigh like phenotype secondary to 3-hydroxyisobutyryl-CoA hydrolase deficiency: A first Indian case

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Dear Editor,

Biallelic mutations in the 3-hydroxyisobutyryl-CoA hydrolase *(HIBCH)* gene causes HIBCH deficiency (HIBCHD) (HIBCHD: OMIM#250620) and present with neurodegeneration, psychomotor retardation, lactic acidosis and abnormalities of basal ganglia.[1] We are reporting a case of HIBCHD presenting as Leigh phenotype.

A 16-month-old male born to consanguineous parentage presented with developmental delay followed by regression, stiffness of limbs, and encephalopathy. Developmentally, attained partial neck controls from 8 months, roll over at 12 months, monosyllables by 13 months, social smile by 6 months, and recognition of mother by 7 months. At around 15 months of age, following febrile illness, the child lost all attained milestones and developed stiffness of limbs. On examination, the occipitofrontal circumference was 44 cm (−1.36 Z score), weight was 8.35 kg (−1.34 Z score), hypertonia with intermittent dystonia, power of 4/5 and exaggerated deep tendon reflexes with extensor plantar response [Figure 1a]. Tandem mass spectrometry (TMS) screening showed elevated C4 hydroxyl carnitine (1.42: Normal range: 0–0.50: NMT - 5.56). Magnetic resonance imaging revealed bilateral symmetrical T2 hyperintensities involving globus pallidus, diffuse cerebral atrophy, and thin corpus callosum with diffusion restriction in bilateral globus pallidus, with hyperintensity on diffusion-weighted images and hypointensity of corresponding apparent diffusion coefficient maps. Small cystic changes representing necrosis were noted in bilateral globus pallidus [Figure 1b-f]. Exome sequencing was sent in suspicion of Leigh like syndrome and identified a novel frameshift deletion c.131delG, p.(Gly44fs*6) in exon-3 of the *HIBCH* gene. The variant is classified as likely pathogenic as per ACMG classification.

The HIBCHD presents with developmental delay, dystonia clinically, and basal ganglia involvement radiologically.[1] Biochemically, the elevation of hydroxy C4 carnitine and urinary excretion of methacrylyl CoA metabolites and S2 carboxy propyl cysteine is found.[2] In the current case, we found elevation of C4 hydroxyl carnitine on TMS. The differentials considered were mitochondrial disorder, mainly Leighs and Leigh like syndrome, and short-chain enoyl CoA hydratase deficiency (*ECHS1* mutation). There was a significant improvement in development and dystonia with dietary management with a low protein diet, symptomatic therapy, carnitine supplements and rehabilitation. To conclude, we report the first Indian child with HIBCH deficiency presenting as Leigh like syndrome.

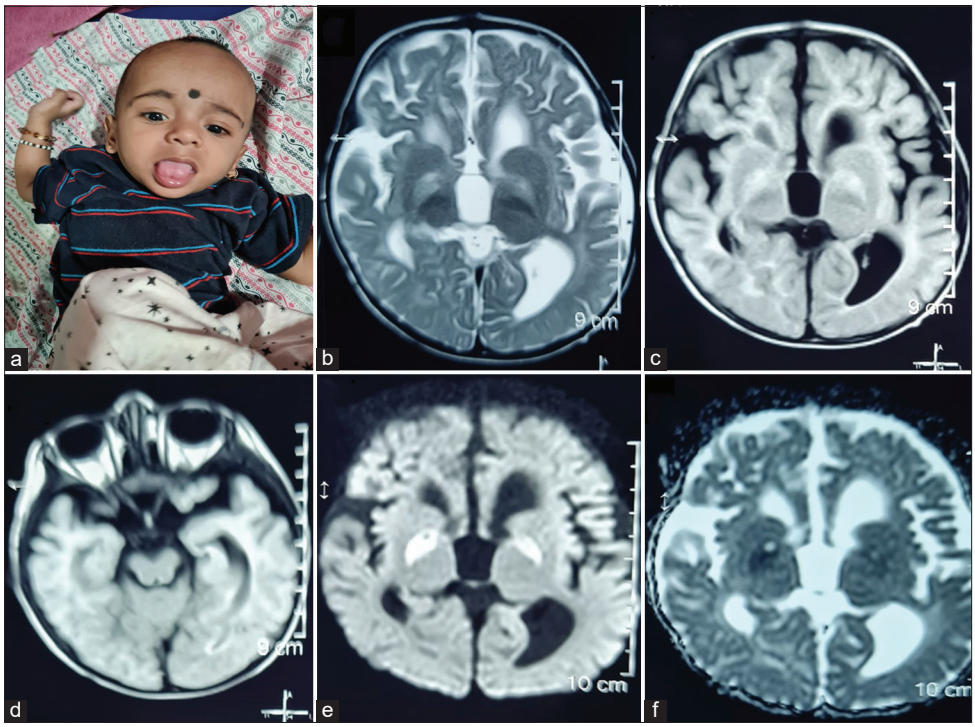

Figure 1: (a) Clinical photograph of the child showing tongue and upper limb dystonia, (b) axial T2-weighted (Two headed arrow), (c) fluid-attenuated inversion recovery sequences of magnetic resonance imaging of brain images showing bilateral symmetrical globus pallidus hyperintensity, bilateral basal ganglia atrophy, predominantly involving globus pallidus (left more than right) (Two headed arrow), (d) cerebral peduncles appear normal (Two headed arrow), (e) axial diffusion weighted images (DWIs) (Two headed arrow) show restriction of diffusion in bilateral globus pallidus, with hyperintensity on DWI images and (f) hypointensity of corresponding apparent diffusion coefficient maps (Two headed arrow).

Ethical approval

The Institutional Review Board has waived the ethical approval for this study

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

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2. Peters H, Ferdinandusse S, Ruiter JP, Wanders RJ, Boneh A, Pitt J. Metabolite studies in HIBCH and ECHS1 defects: Implications for screening. Mol Genet Metab 2015; 115:168-73.

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Journal Summary

Advances and challenges in pediatric care: Insights from diverse clinical perspectives

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1. Reassessing febrile seizures: Long-term neurological and psychiatric implications

Source**:** Wang DS, Chung CH, Hsu WF, Chen SJ, Chu DM, Chien WC, Tzeng NS, & Fan HC. (2024). Higher risk of psychiatric disorders in children with febrile seizures: A nationwide cohort study in Taiwan. *Pediatric Neurology*, 154, 26–35. https://doi.org/10.1016/j.pediatrneurol.2024.02.005

Febrile seizures are a prevalent occurrence amongst children aged 6 months–6 years, as evidenced by previous research, such as a Danish study demonstrating a positive correlation between febrile seizures and the overall incidence of psychiatric disorders. In recent years, evolving perspectives on febrile seizures have challenged the notion of their benign prognosis. In this population-based observational study conducted in Taiwan, researchers sought to delve deeper into this association and identify associated risk factors.

Utilising data from the comprehensive Taiwan National Health Insurance Research Database spanning a 15-year period from 2000 to 2015, which covers over 99% of the population, the researchers analysed records of approximately 1,936,512 individuals, accounting for roughly 10% of Taiwan's populace. Following stringent exclusion criteria, the study included 2,464 children diagnosed with febrile seizures as cases, alongside 7,392 controls.

Febrile seizures were examined as the primary exposure, with psychiatric disorders serving as the main outcomes of interest. The retrospective cohort study, encompassing nearly two million children, revealed a noteworthy finding: Children with febrile seizures faced a substantially heightened risk, 4.7 times higher, of developing psychiatric disorders compared to their counterparts without such seizures.

These psychiatric disorders encompassed a spectrum of conditions, including anxiety, depression, bipolar disorder, sleep disorders, substance-related disorders, psychotic disorders, and organic mental disorders, underscoring the breadth of the impact. Of particular significance were comorbidities such as attention-deficit/hyperactivity disorder (ADHD) and intellectual disability, which were identified as significant contributors to psychiatric morbidity amongst children with febrile seizures.

Furthermore, the study highlighted a nuanced risk profile, indicating that children with febrile seizures, particularly those with underlying ADHD, exhibited heightened susceptibility to specific psychiatric morbidities, notably anxiety.

The implications of these findings are profound, emphasising the critical role of vigilance amongst various stakeholders, including parents, clinicians, nurses, and educators, in early detection and intervention. Specifically, the study underscores the importance of considering psychiatric

symptoms in children with febrile seizures and the potential benefits of early referral for appropriate management and support.

2. The retinoid hypothesis: A two-decade review of congenital diaphragmatic hernia

Source: Rivas JF, & Clugston RD. (2024). The aetiology of congenital diaphragmatic hernia: The retinoid hypothesis 20 years later. *Pediatr Res*, 95, 912–921. https://doi.org/10.1038/s41390-023-02905-7

Congenital diaphragmatic hernia (CDH) poses a significant threat to neonatal health, affecting approximately 2–3 in 10,000 births with considerable mortality and long-term morbidity amongst survivors. Despite its clinical significance, the underlying mechanisms of CDH remain elusive. In 2003, Greer *et al*. introduced the retinoid hypothesis, suggesting that aberrant retinoid signalling contributes to abnormal diaphragm development in CDH. Twenty years later, the authors meticulously review the literature, spanning both animal models and human studies, to provide a thorough evaluation of the hypothesis's validity and implications.

CDH presents a complex clinical picture, ranging from isolated defects to more intricate manifestations, such as Bochdalek hernias and Morgagni hernias. Genetic and environmental factors play pivotal roles in CDH aetiology, with up to 40% of cases linked to identifiable genetic causes and various environmental risk factors implicated in disease development.

The retinoid hypothesis posits retinoic acid (RA) signalling disturbances during early gestation as crucial contributors to CDH pathogenesis. RA, a metabolite of dietary Vitamin A, exerts potent developmental regulatory effects, forming the cornerstone of this hypothesis. Since its inception, the retinoid hypothesis has guided significant research efforts, with findings from animal models and human studies providing substantial support.

Over the past two decades, numerous studies have reinforced the association between RA signalling and CDH formation, consolidating the evidence base for the retinoid hypothesis. By synthesising decades of research findings, this review underscores the enduring relevance and significance of the retinoid hypothesis in elucidating the molecular mechanisms underlying CDH development. Furthermore, it highlights the crucial role of RA signalling in diaphragm morphogenesis, offering valuable insights for future investigative endeavours and therapeutic interventions aimed at improving outcomes for affected individuals.

3. Does prednisolone impact recovery in paediatric Bell's palsy? A critical follow-up inquiry

Source: Babl FE, Herd D, Borland ML, Kochar A, Lawton B, Hort J, West A, George S, Oakley E, Wilson CL, Hopper SM, Cheek JA, Hearps S, Mackay MT, Dalziel SR, & Lee KJ. (2024). Facial function in bell palsy in a cohort of children randomised to prednisolone or placebo 12 months after diagnosis. *Pediatric Neurology*, 153, 44–47. https://doi.org/10.1016/j.pediatrneurol.2024.01.011

Inadequate data exists regarding the medium-term recovery of paediatric patients afflicted with Bell palsy or acute idiopathic lower motor neuron facial paralysis. This investigation embarked on a 12-month follow-up study encompassing children aged 6 months–under 18 years, subsequent to their involvement in a randomised trial exploring the effectiveness of prednisolone. Evaluation of facial function employed both the clinician-administered House-Brackmann scale and its modified parent-administered counterpart.

A cohort of 187 children underwent randomisation, with 93 assigned prednisolone and 94 allocated a placebo. At the 6-month juncture, recovery of facial function, as adjudged by the clinician-administered scale, was apparent in 98% (78 out of 80) of the prednisolone cohort and 93% (76 out of 82) of the placebo group. Concurrently, using the modified parent-administered scale, recovery rates stood at 94% (75 out of 80) versus 89% (72 out of 81) at 6 months and 96% (75 out of 78) versus 92% (73 out of 79) at 12 months.

Despite the majority of participants experiencing complete facial function recovery within 6 months, a subset of children exhibited incomplete recovery at the 12-month interval, irrespective of prednisolone administration. These findings underscore the variability in outcomes and underscore the significance of prolonged monitoring in this patient demographic.

4. Does neonatal phototherapy heighten cancer risk in children?

Source: Kuitunen I, Nikkilä A, Kiviranta P, Jääskeläinen J, & Auvinen A. (2024). Risk of childhood neoplasms related to neonatal phototherapy – A systematic review and meta-analysis. *Pediatric Research*, 10.1038/s41390-024-03191-7. Advance online publication. https://doi.org/10.1038/s41390-024-03191-7

According to this systematic review and meta-analysis, children with a history of neonatal phototherapy face a 1.2-–1.6-fold elevated risk of hematopoietic cancers and solid tumours. However, several considerations emerge when interpreting these findings, including limitations in the reporting quality of the primary studies, potential causal pathways and confounding factors.

Some studies suggest that the heightened cancer risk may stem, at least in part, from hyperbilirubinemia rather than phototherapy, a phenomenon known as confounding by indication. This speculation aligns with evidence indicating that cancer incidence amongst children with hyperbilirubinemia, but without phototherapy, falls between

the rates observed in children without hyperbilirubinemia and those subjected to phototherapy.

Initially intending to analyse cancer risk by phototherapy duration and intensity, the researchers found scant reporting on phototherapy duration across most studies. Prematurity, a factor linked to both phototherapy and cancer risk, further complicates the analysis. Notably, while prematurity did not correlate with cancer incidence amongst treated and non-treated premature infants, full-term infants subjected to phototherapy exhibited a slightly elevated risk of hematopoietic cancers.

Comparing their findings to prior meta-analyses, the researchers noted similarities alongside key disparities and concerns. Unlike earlier studies that pooled case-control and cohort data, this review refrained from doing so, thus mitigating heterogeneity in reporting. In addition, while previous analyses lacked sensitivity assessments, this review conducted a more comprehensive evaluation.

Despite its strengths, including adherence to a pre-registered protocol and meticulous analysis, this review is constrained by the limitations of its constituent studies. Many studies exhibited a high risk of bias due to inadequate adjustment for potential confounders. Moreover, the inability to assess mortality or exposure-outcome gradients, coupled with variations in phototherapy practices and incomplete covariate adjustment, underscores the need for further research.

In light of these findings, caution should guide clinical practice. Although neonatal phototherapy appears associated with increased cancer risk, current evidence does not warrant changes in its use. Nonetheless, adherence to guidelines and judicious use of phototherapy are essential to mitigate potential harm. Moving forward, rigorous studies are imperative to deepen our understanding of the relationship between phototherapy and neoplasia and unravel potential causal pathways.

5. Does inhaled salbutamol improve outcomes in preterm infants with chronic lung disease?

Source: Ng G, Bruschettini M, Ibrahim J, & Da Silva O. (2024). Inhaled bronchodilators for the prevention and treatment of chronic lung disease in preterm infants. *The Cochrane Database of Systematic Reviews*, 4(4), CD003214. https://doi.org/10.1002/14651858.CD003214.pub4

Chronic lung disease (CLD) is common amongst preterm infants and is linked to respiratory morbidity. Bronchodilators are often used to alleviate muscle hypertrophy in small airways, potentially improving compliance and reducing airway resistance. Despite their widespread use, it is uncertain whether they lead to better clinical outcomes. This updated Cochrane review aims to evaluate the impact of inhaled bronchodilators on mortality and other complications in preterm infants at risk for or diagnosed with CLD.

Two randomised controlled trials were included in this review, with one providing usable outcome data. The trial, conducted in six neonatal intensive care units, involved 173 infants with a gestational age of <31 weeks. The intervention group received salbutamol for CLD prevention.

The evidence suggests that salbutamol may not significantly affect mortality or CLD at 28 days compared to placebo. However, certainty regarding these outcomes is low due to limited data. The effect of salbutamol on pneumothorax remains uncertain, with no significant differences reported between groups.

No trials evaluated bronchodilator therapy for CLD treatment, highlighting a gap in current research. Future trials should consider including relevant clinical outcomes beyond pulmonary mechanics to better assess the efficacy and safety of bronchodilator agents in preterm infants.

6. Are post-natal corticosteroids safe and effective for preventing bronchopulmonary dysplasia in preterm infants?

Source: Van de Loo M, Van Kaam A, Offringa M, Doyle LW, Cooper C, & Onland W. (2024). Corticosteroids for the prevention and treatment of bronchopulmonary dysplasia: An overview of systematic reviews. The Cochrane Database of Systematic Reviews, 4(4), CD013271. https://doi.org/10.1002/14651858.CD013271.pub2

This Cochrane review delves into the persistent challenge of bronchopulmonary dysplasia (BPD) in pre-mature infants, exploring the potential of post-natal corticosteroid therapy as a solution. Through a meticulous examination of nine systematic reviews, which encompassed a substantial body of evidence comprising 87 randomised controlled trials and involving 9419 preterm infants, the researchers aimed to ascertain both the efficacy and safety of postnatal corticosteroids in this vulnerable population.

The findings offer insights into the nuanced effects of various corticosteroid regimens. Early initiation of systemic dexamethasone or hydrocortisone emerges as a promising intervention, demonstrating a potential reduction in the combined outcome of death or BPD at 36 weeks' post-menstrual age. However, this potential benefit is juxtaposed with notable adverse effects such as cerebral palsy or gastrointestinal perforation, underscoring the need for cautious consideration.

Moreover, late initiation of systemic dexamethasone presents another avenue for intervention, showing efficacy in reducing the risk of death or BPD. Despite its potential benefits, this approach also carries inherent risks, albeit to a lesser extent compared to early initiation.

Inhaled corticosteroids, particularly when initiated early, demonstrate favourable outcomes without apparent adverse

effects, highlighting their potential as a safer alternative. However, further investigation is warranted to elucidate their efficacy in late initiation scenarios.

An intriguing finding emerges regarding endotracheal instillation of corticosteroids with surfactant, showing promise in reducing the risk of death or BPD without evident adverse effects. This novel approach holds potential for clinical application pending further validation through ongoing large-scale trials.

However, amidst these promising findings, the study underscores the need for cautious interpretation and further research, particularly regarding the long-term effects and optimal timing of corticosteroid administration. This nuanced understanding is essential for guiding clinical practice effectively and ensuring the optimal care of preterm infants at risk of BPD.

7. Are there unexplored links between congenital anomalies?

Source: Morris JK, Bergman JE, Barisic I, Wellesley D, Tucker D, Limb E, Addor MC, Cavero-Carbonell C, Dias CM, Draper ES, Echevarría-González-de-Garibay LJ, Gatt M, Klungsøyr K, Lelong N, Luyt K, Materna-Kiryluk A, Nelen V, Neville A, Perthus I, Pierini A, Randrianaivo-Ranjatoelina H, Rankin J, Rissmann A, Rouget R, Sayers G, Wertelecki W, Kinsner-Ovaskainen A, & Garne E. (2024). Surveillance of multiple congenital anomalies; searching for new associations. Eur J Hum Genet, 32, 407–412. https://doi.org/10.1038/s41431-023-01502-w

This study scrutinised 1386 unique combinations of dual anomalies co-occurring in individual cases, revealing 31 significant associations, with 20 previously established and 11 novel associations. Amongst these, six pairs of anomalies emerged as new associations, distinct from any known associations or sequences, warranting ongoing surveillance within the EUROCAT framework for potential clustering and trends.

Known associations, delineated based on published literature, included ten pairs associated with limb body wall complex, OEIS complex or VACTERL association. For instance, the association between neural tube defects and omphalocele elucidated three anomaly pairs. In addition, the link between diaphragmatic hernia and pulmonary hypertension explained another pair. Less recognised associations, albeit documented, constituted the remaining three pairs.

Amongst the novel associations, three pairs overlapped, such as severe Congenital heart disease (CHD), Tetralogy of Fallot and common Atrio-ventricular (AV) canal with duodenal atresia, suggesting a potential genetic basis. Notably, the association between encephalocele and an/microphthalmos was scarcely documented in the literature, similar to an/microphthalmos and cleft lip. The association of hydrocephaly and hypoplastic right heart syndrome, predominantly in males with associated renal/genital anomalies, was identified in a few publications, hinting at possible genetic implications or inclusion within VACTERL. Continued surveillance is imperative to monitor these new associations.

Comparable methodologies, such as the co-occurring defect analysis approach, have been employed in prior studies to unveil new associations. However, this study exclusively focused on cases with at least two anomalies, without adjusting for clustering tendencies observed in isolated anomalies. Moreover, the analysis excluded cases with known chromosome or genetic anomalies to spotlight potential novel anomaly clusters rather than reiterating known associations.

The study's strength lies in its extensive EUROCAT dataset, encompassing 32 congenital anomaly registries and over 6.5 million births. Standardised coding methods and stringent data quality monitoring enhance the reliability of findings. Nonetheless, limited access to detailed genetic information for all cases poses a notable limitation.

In essence, while most identified associations corroborate existing literature, the discovery of six new associations underscores the need for continued surveillance, highlighting the evolving landscape of congenital anomalies warranting ongoing investigation within the EUROCAT surveillance system.

8. Navigating uncertainty: Rare disease diagnosis through newborn screening

Source: Raspa M, Kutsa O, Andrews SM, Gwaltney AY, Mallonee E, Creamer A, Han PK, & Biesecker BB. (2024). Uncertainties experienced by parents of children diagnosed with severe combined immunodeficiency through newborn screening. Eur J Hum Genet, 32, 392–398. https://doi.org/10.1038/s41431-023-01345-5

This study sheds light on the uncertainties faced by parents when their child receives a diagnosis through newborn screening. Despite early identification of their child's condition, parents of infants with severe combined immunodeficiency (SCID) encountered various uncertainties about the future. The study findings resonate with prior research on uncertainty in pre-natal or newborn screening diagnoses. For example, parents of infants diagnosed prenatally with congenital heart disease expressed immediate uncertainties post-diagnosis and longer-term uncertainties about prognosis. Similarly, parents of infants screening positive for cystic fibrosis with inconclusive diagnoses also reported uncertainty. In addition, parents of infants identified with Pompe disease through newborn screening faced uncertainties about symptom onset and treatment initiation.

Contrary to expectations from diagnostic odyssey literature, a pre-symptomatic diagnosis through newborn screening did not alleviate uncertainty for parents of children with rare disorders. Instead, uncertainties persisted regarding treatment decisions, outcomes and the long-term course of their child's condition.

A key finding of this study was the dynamic yet persistent nature of uncertainties experienced by parents of children with SCID across their journey. Certain uncertainties were more pronounced at specific stages, whereas others persisted throughout multiple phases. Scientific uncertainties, particularly related to diagnosis and treatment, were prevalent early in the SCID journey, while prognostic uncertainties emerged later, in the post-treatment phase. Personal and practical uncertainties, including logistical, financial and relational aspects, were prominent in the pre-treatment and new normal stages. Existential uncertainties were frequently reported early on but resurfaced in the new normal stage and could persist throughout the child's life. Similar patterns of evolving uncertainty have been observed in other chronic conditions, underscoring the importance of ongoing support for parents throughout their journey.

Parents in this study expressed a range of negative emotional responses to uncertainty, including anxiety, worry, fear, guilt and grief. Managing uncertainty for parents of children with rare genetic conditions such as SCID is a complex process that may culminate in adaptation and acceptance. Hope, optimism, psychosocial support and the provision of information by healthcare providers are vital factors in coping with uncertainty. While this study provides valuable insights, it has limitations. The researchers utilised a convenience sample of predominantly English-speaking mothers recruited through patient advocacy groups, limiting the generalisability of its findings. In addition, the diverse geographic locations and treatment options of participants may have influenced their experiences with uncertainty. Finally, their semi-structured interview approach may have overlooked certain subtypes of uncertainties experienced by parents.

In conclusion, this study underscores the chronic and multifaceted nature of uncertainties in the SCID journey and emphasises the importance of preparing parents for this journey by addressing and assisting them in coping with uncertainty. Healthcare providers, including genetic counsellors, immunologists and transplant specialists, play a pivotal role in supporting parents and providing both informational and emotional support. Leveraging external resources and intrinsic coping mechanisms will aid parents in navigating the road ahead and fostering positive adaptation.

Ethical approval

The Institutional Review Board has waived the ethical approval for this study.

Declaration of patient consent

Patient's consent was not required as there are no patients in this study.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

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