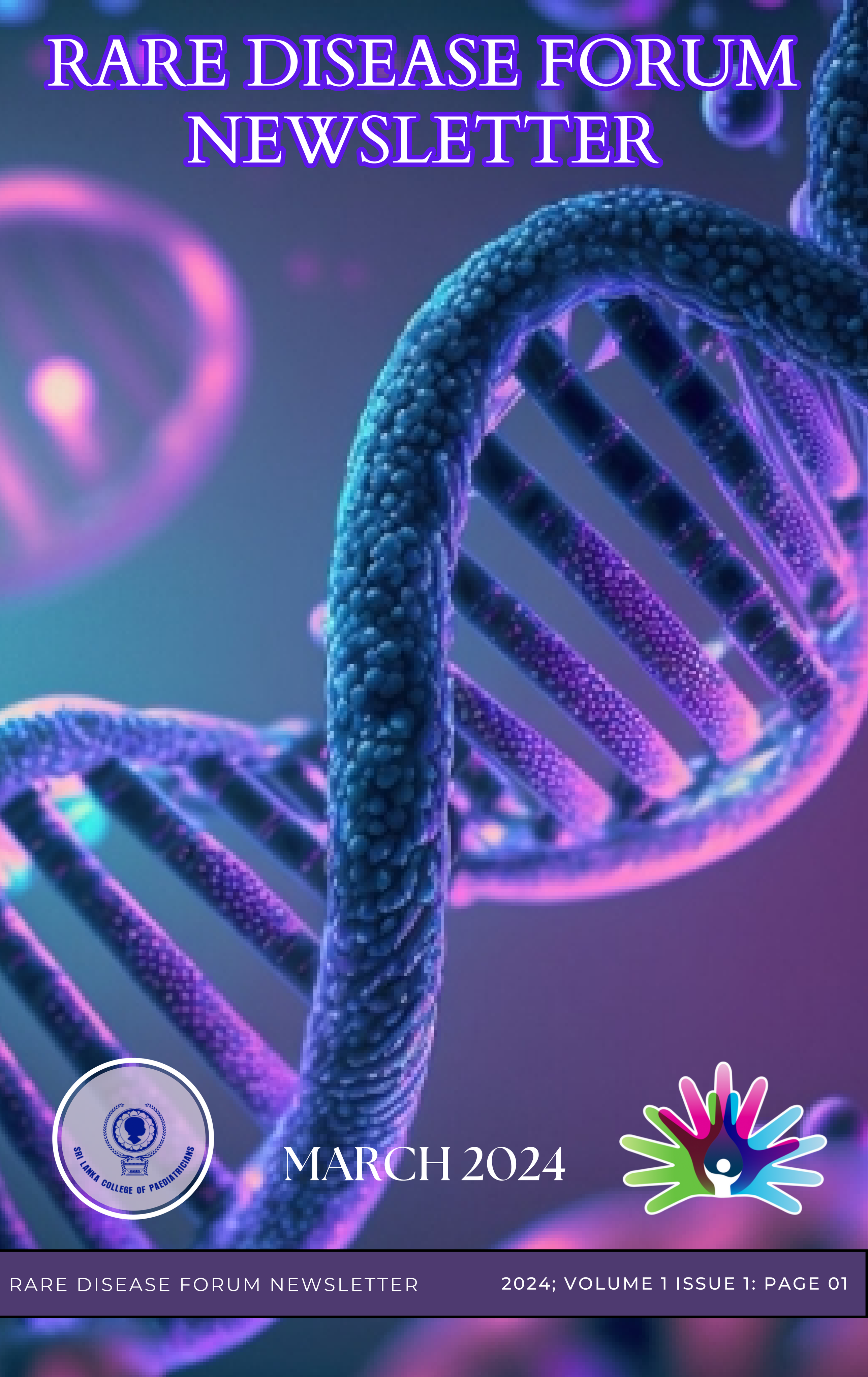
RARE DISEASE FORUM NEWSLETTER



SRI LANKA COLLEGE OF PAEDIATRICIANS

MARCH 2024

> 300 MILLION PEOPLE LIVE WITH A RARE DISEASE WORLDWIDE

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RARE DISEASE FORUM 2024

SRI LANKA COLLEGE OF PAEDIATRICIANS

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8th Rare Disease Day 2024

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8TH RARE DISEASE DAY SYMPOSIUM 2024 SRI LANKA

The Rare Disease Day Symposium 2024, took place on March 1st, 2024, centered around the theme “"Beyond Borders: Bridging Gaps in Rare Disease Care" at the New Auditorium, Lady Ridgeway Hospital for Children. The event garnered participation from a diverse group of 100 attendees, including Consultant Paediatricians, Senior Registrars, Registrars, and members of the Rare Disease Forum.

Throughout the symposium, a series of case presentations were delivered by both paediatricians and postgraduate trainees, enriching the discussions and insights shared during the event.

The speakers of the webinar included:

- Beyond subtle dysmorphism
  Prof Ruwanthi Perera - Professor in Paediatrics, FMS, University of Sri Jayewardenepura

- Genetic testing: an overview for clinicians:
  Dr Hasani Hewavitharana - Consultant Paediatric Clinical Geneticist (Acting), Lady Ridgeway Hospital for Children

- Uric acid & inborn errors of metabolic disorders-clinical experience:
  Dr Eresha Jasinge - Chemical Pathologist, Lady Ridgeway Hospital for Children

- Metabolic emergencies in Paediatrics
  Dr Imalke Kankananarachchi Senior Lecturer in Paediatrics, Faculty of Medicine, University of Ruhuna

- Role of bone marrow transplantation in rare diseases in children
  Dr Prabani Maddumarachchi - Consultant Paediatric/Adolescent Oncology, National Cancer Institute

SPEAKER ABSTRACT

Beyond subtle dysmorphism....

Professor Ruwanthi Perera
Faculty of Medical Sciences, University of Sri Jayewardanepura

Dysmorphism or dysmorpholoy is a Greek terminology depicting a disordered or abnormal shape and the term was first coined by David Smith in 1960s in USA. Dysmorphic features are congenital defects and abnormalities of the body structure that originate before birth and are usually not found in other individuals of the same age or ethnic background. Recognition of dysmorphism leads to early detection and intervention of genetic disorders.

Different terminologies are being used to describe dysmorphism. A malformation or an anomaly is a primary defect occurring before 10 weeks of gestation leading to a basic alteration in the structure. Cleft lip and anencephaly are malformations. A single malformation can lead to a pattern of multiple defects leading to a malformation sequence. A primary lumbar neural tube defect can lead to talipes and hydrocephalus. Several different defects in morphogenesis often with an underlying cause gives rise a malformation syndrome. Syndromes result due to single gene or polygenic disorders, chromosomal disorders, microdeletion syndromes or teratogenesis. Associations are a group of anomalies that occur more frequently than expected by chance and do not have a unified aetiology and VATER & VACTERAL are well known associations. Deformation results due to distortion of an otherwise normal structure by a physical force. Arthrgryposis will result due to a deformation of a limb due to uterine anomaly of the mother. Destruction of a previously normal structure leads to a disruption. Amniotic bands will lead to disruption of a limb extremity and will result in congenital ring constrictions. Dysplasia is due to an abnormal cellular organization with in tissue leading to structural changes and skeletal dysplasias are known examples. All these mechanisms can result in dysmorphic features.

In a child who is suspected to be dysmorphic warrants a systematic approach. It should include information gathering, interpretation of the anomalies and attempting to arrive at a specific diagnosis. History concentrating on consanguinity as well as pedigree are of paramount importance. Pre-pregnancy medical history, folate consumption and identified uterine anomalies of the mother is vital. Antenatal history emphasizing fetal growth, placental structure and function as well as exposure to teratogens yields vital clues. Child's history navigates to birth parameters, condition at birth as well as immediate antenatal complications like feeding difficulties, jaundice and hypoglycemia need to be inquired. Current growth parameters as well as growth patterns should be looked into from previous records. Development delays and specially regressions direct toward the aetiology of dysmorphisim.

Physical examination is the most important step in the process. Growth assessment should include routine height, weight, head circumference as well as

measurement of body segments, limb segments and comparison of sides in cases of body asymmetry.In labeling a dysmorphic feature, standardization of the morphology is important to increase the utility of descriptions of the human phenotype and to facilitate the reliable comparisons of findings among patients. National Human Genome Research Institute has developed the human malformation terminology under head & face, periorbital region, ear, nose & philtrum, lip, mouth & oral region and hands & feet terminologies. (speech - Prof Ruwanthi) Definitions of relevant distances and displacements have been established as well.

It is vital to categorize the anomalies into minor and major. Minor anomalies are defined as unusual morphologic features that are of no serious medical or cosmetic consequence to the patient and are most common in areas of complex and variable features such as face, auricles, hands and feet. Yet, minor indicators may serve as indicators of altered morphogenesis in a general sense or may constitute valuable clues in the diagnosis of a specific pattern of malformation. In addition, before ascribing significance to a given minor anomaly in a patient, it is important to note whether it is found in other family members. Any minor anomaly can be a usual feature in a particular family. Major anomalies are severe in nature, impair body function and lead to serious medical and cosmetic consequences. They may be isolated or affect different body systems. May require surgical inputs for existence.

SPEAKER ABSTRACT

Genetic testing: An overview for clinicians

Dr Hasani Hewavitharana
Consultant Paediatric Clinical Geneticist (Acting)
Lady Ridgeway Hospital for Children

Genetic testing identifies changes in human chromosomes, genes or proteins and helps to reveal if a person is at risk for heritable diseases, if they might pass them on to their children, or even how their body responds to certain medications.
In the past decade, genetic testing has made exponential growth. With a plethora of tests and technologies made available, it has now started to play a larger role and revolutionise healthcare delivery. Its translation into clinical practice by identifying disease causing genes, has paved the way to creating newer treatments that can activate or deactivate these problematic genes and has opened up the doorway to precision medicine.
There are various types of genetic tests, each serving specific purposes. Diagnostic tests identify genetic conditions in symptomatic individuals. Carrier screening determines if a person carries a disease-causing gene which can be passed on to their children. Prenatal tests allow the identification of genetic disorders in developing fetuses. Newborn screening identifies apparently healthy newborns with disease susceptibility and allows early intervention for better outcomes. Predictive and presymptomatic testing assess the risk of developing a disease before symptoms appear and aid in preventive measures. Pharmacogenomic testing analyses how a person's genes may affect their response to medicine. Research genetic testing involves testing for scientific purposes, to understand the contribution of genetic factors in disease causation and drug responses.
Pretest counselling with patients and families should be tailored according to the indication (diagnosis, prediction, screening) of the test and guided by adequate information on risks, benefits, the possibility of genetic results altering the clinical management, alternative options to genetic testing, costs and patient information important for the patient or family members who may carry the genetic mutation.
The benefits of genetic testing are not only limited to the nature of the results (positive or negative), but also gives a sense of relief to people enduring the lack of a proper diagnosis for long periods of time compounded by a feeling of uncertainty in making their healthcare management decisions. Genetic testing gives the answer concerning the necessity to take or neglect a number of investigations and screenings in some cases. It can predict the probability of specific diseases and choose appropriate preventive measures.
Despite there being many indications for its use, most genetic tests come with a high price tag and are time consuming, which has discouraged many clinicians on its use in resource poor settings such as in Sri Lanka.

In many instances though, its benefits can outweigh the risks/costs incurred. For example, it enables paediatricians to avoid invasive testing, like muscle biopsies or kidney biopsies in Duchene muscular dystrophy (DMD) and steroid-resistant nephrotic syndrome (SRNS) respectively. It can also help clinicians to avoid therapies with significant adverse events, to decide the dosing and duration of immunosuppression in SRNS, pre-transplantation therapy and recurrence risk post transplantation. Testing for gene variations can also be used to identify individuals who are at-risk of severe idiosyncratic adverse events, assess drug effectiveness and metabolism (faster versus slower metabolizers), which will improve the effectiveness and safety of epilepsy therapies including using alternative strategies for treatment. In inborn errors of metabolism, a molecular diagnosis will guide successful treatment and genetic counselling. The precise diagnosis in Noonan syndrome and related syndromes is crucial as children with RASopathy have an increased risk of childhood cancer, and thus require close follow-up. Cytogenetic analysis of haematological disorders for identification of chromosomal abnormalities (aneuploidy and chromosomal rearrangements such as deletions, translocations) has become essential for diagnosis, risk-stratification, management and monitoring of minimal residual disease, early detection of relapse, and for implementation of targeted therapy.

As we conclude our exploration of genetic testing, it’s evident that this science helps to unlock the secrets hidden in our unique genetic makeup and if used rationally its benefits can outweigh its cost. With so much new information, recent advancements in disease phenotyping, and the broad spectrum of genetic tests available, the role of healthcare providers with a training in clinical genetics is crucial to guide others to whom, when, and what kind of genetic testing they should ask for. Whether it is the understanding of potential health risks, making informed family planning decisions, or optimizing medication responses, genetic testing empowers individuals to have autonomy over their well-being and offer a promising future of precision medicine.

SPEAKER ABSTRACT

Uric acid and inborn errors of metabolic disorders: Clinical experience

Eresha Jasinge,
Chemical Pathologist,
Lady Ridgeway Hospital for Children

Abstract

Uric acid, a nitrogen containing compound, is the final product of purine metabolism in humans. Liver and intestines are the major sites of its synthesis. Approximately two thirds of uric acid are excreted via kidneys while the remainder is eliminated through the intestines. 90 % of freely filtered uric acid is absorbed.

Altered uric acid level in both serum and urine helps in identifying some of the purine pathway disorders. The diagnosis of purine pathway defects may be overlooked due to variable clinical presentations. Inherited disorders of purine metabolism have a high clinical impact with a wide array of clinical presentations involving different medical specialties mainly neurology and renal. Uric acid, a compound that can be easily analyzed, may be the initial indicator to perform confirmatory tests for purine pathway defects.

Several factors need to be considered when interpreting uric acid levels such as age and sex matched reference ranges, diet, intake of medications, the presence of lactic acidosis and renal failure. Children may have normal serum uric acid level despite its overproduction due to higher uric acid clearance, hence normal serum uric acid doesn't exclude hyperuricemia. Bacterial contamination elevates urine uric acid due to rapid degradation of purine nucleosides and bases.

A chemical pathology lab should play a detective role by introducing uric acid to the routine general biochemistry test panel to screen for rare disorders especially in patients with neurological and renal symptoms. Uric acid can be analyzed easily in blood and urine using general biochemistry analyzers. Uric acid to creatinine ratio and fractional excretion of uric acid are derived parameters using uric acid andcreatinine in blood and randomly collected urine in assisting the diagnosis. In the era of rapidly available sophisticated laboratory tests such as enzyme assays, high performance liquid chromatography and molecular diagnostic methods, uric acid still plays a vital role in a resource limited country like ours. In addition to screening, uric acid is an important marker in the management of some of the disorders.

Pathophysiology of uric acid, and a few case scenarios, all referrals, involving hyperuricaemic hyperuricosura, hypouricaemic hypouricosuria and hypouricaemic hyperuricosuria, will be discussed to highlight the importance of requesting this indispensable analyte.

SPEAKER ABSTRACT

Metabolic emergencies in Paediatrics

Dr Imalke Kankananarachchi
Consultant Paediatrician Teaching Hospital Karapitiya
Senior Lecturer in Paediatrics, Faculty of Medicine,
University of Ruhuna

Inherited metabolic disorders have multiple presentations ranging from paediatric emergencies to chronic manifestations. Early identification and management of such metabolic derangements are essential for the overall outcome of the patient. Hypoglycaemia, Metabolic acidosis and Hyperammoniemia cover a large number of metabolic emergencies. However, these manifestations could happen in children without underlying IMD. Irrespective of the underlying aetiology of the metabolic derangement, the management is the same in many of these emergencies. Often, these are associated with intercurrent infection. Therefore, sometimes, clinicians might miss the underlying metabolic diagnosis in the presence of a proven infectious aetiology. In most metabolic emergencies, it's essential to obtain relevant blood and urine samples to arrive at the diagnosis before starting the treatment. However, if there is a significant delay in obtaining samples, the treatment should not be delayed since it can cause irreversible neurologic disability.

The definition of hypoglycemia in children is controversial; generally, blood glucose (BGL) values less than 2.6mmol/L are considered the cut-off value in children. However, a BGL of 3.3mmol/L is low enough to cause signs and/or symptoms of impaired brain function and neurogenic response in some individuals. Idiopathic ketotic hypoglycaemia, Adrenal insufficiency and hyperinsulinemia are common non-metabolic causes of hypoglycemia in children. Glycogen storage disorders, fatty acid oxidation defects and organic acidemias are the most common IMDs that can present with hypoglycemia. A detailed history, examination and basic investigations are important to determine the underlying aetiology. Since IKH remains the most common diagnosis, urine ketone bodies should be checked in every child with hypoglycaemia. In standard settings, blood should be preserved for serum insulin, ammonia, lactate, and Acylcaritine profiles since it's a golden opportunity to obtain the critical sample. However, further analysis of the obtained samples depends on the clinical picture, basic biochemistry results, and the presence of ketone bodies. Treatment aims to return BGL to within the normal range (>3.9 mmol/L). Initial management depends on the patient's consciousness and availability of Intravenous access. If the patient is fully conscious and tolerating feeds, oral glucose 10–20 g, for example, 200 mL milk, 100 mL Lucozade, Glucogel or two teaspoons sugar, followed by a snack of starchy carbohydrates or a milk feed in infants is recommended.

In an unconscious child with intravenous access, 10% dextrose 2-4mL/kg should be given, followed by 10% dextrose infusion to prevent rebound hypoglycaemia. Intramuscular Glucagon is the drug of choice in an unconscious child without IV access. After the initial correction of hypoglycaemia, it's essential to work on the possible aetiology of the presentation and manage it accordingly.

Metabolic acidosis is a frequent problem encountered in paediatric practice. Diarrhoea is the most common cause of metabolic acidosis in children. Increased loss of bicarbonate from the body, excess production of endogenous acids, exogenous addition of acids, and failure of the kidney to excrete acids are the main mechanisms of metabolic acidosis in children.

Metabolic acidosis due to IMD can be due to the accumulation of Ketoacidosis, lactic acidosis or Oragnic acids. Clinical evaluation and baseline investigations will be useful to arrive at the tentative diagnosis. The mainstay of the management of MA is to treat the underlying cause, for example, Insulin in diabetic ketoacidosis and fluid resuscitation in hypovolemia. Intravenous sodium bicarbonate should be used judiciously because of its potential adverse effects in increasing intracellular acidity, hypocalcaemia and hypernatremia. However, IV HCO3 correction is indicated when the PH is <7.1, base excess > 10mmol/L and hyperventilating but not compensating or at risk of exhaustion. The estimated HCO3 deficit is calculated using this formula (Target HCO3 – Current HCO3) × Weight (in kg) × 0.5. Usually, 50% of the calculated dose is given over 30-60 minutes, followed by the rest given over 6-24 hours. If there is clinical suspicion of organic acidemia, Intramuscular B12, Biotine, Carnitine, and Thiamine can be started. Empirical Riboflavin, vitamin C, vitamin E, and coenzyme Q10 can be started if there is a tentative diagnosis of mitochondrial disorders.

Hyperammonemia (HA) is a paediatric emergency which needs immediate treatment. Due to the lack of specificity of the symptoms, it’s difficult to diagnose HA clinically without plasma NH3 levels. Normal plasma NH3 level should be less than 50 mmol/l, but mildly raised values are common – up to 80 mmol/l. Artifactually high NH3 values can be caused by muscle activity, haemolysis or delay in separating the sample. Capillary samples are often haemolysed or contaminated and, therefore, should not be used.

Moreover, a number of non-metabolic disorders, such as sepsis, liver failure, sodium valproate therapy, etc., can present with HA. Severe isolated HA is seen in children with urea cycle defects, and HA with ketoacidosis and hypoglycaemia are seen in OAs and Fatty acid oxidation defects, respectively. Initial management decisions should be based mainly on how severe the NH3 levels and the presence of encephalopathy.

All children with hyperammonaemia should have intravenous therapy and stop oral feeds until the diagnosis is known and the patient is stable. Intravenous (IV) 10% glucose (2ml/kg bolus) should be given as the first option, followed by 0.9% Sodium chloride 10mL/kg bolus. Subsequently, 0.9% Sodium chloride + 10% Dextrose infusion should be started. The target blood glucose value should be 6 - 10 mmol/L. If plasma glucose rises above 14 mmol/L and there is glycosuria, start IV insulin infusion (0.025 units/kg/hr-titrated to blood glucose levels) instead of reducing glucose intake. In an ideal setup, intravenous ammonia scavengers such as sodium benzoate, sodium phenylbutyrate, Carbohydrate, L-arginine, and L-carnitine should be started immediately. The child should not be kept nil by mouth for more than 48 hours and need to start on Intravenous lipids. When blood ammonia drops below 100 µmol/L, proteins can be reintroduced slowly. If there is no improvement despite ammonia scavengers or the ammonia of >500mmol/L, haemodiafiltration or haemodialysis should be started. However, exchange transfusion should not be done due to the increased risk of catabolism.

SPEAKER ABSTRACT

Role of Haematopoietic Stem Cell Transplant in Rare Diseases in Children

Dr Prabani Maddumarachchi
Consultant in Paediatric/Adolescent Oncology and
Special Interest in Stem Cell Transplant
National Cancer Institute (Apeksha Hospital), Sri Lanka

Hematopoietic stem cell transplant (HSCT) stands as the curative treatment for numerous hematological, oncological, immunological, and metabolic diseases in children, many of which lack definitive medical management. Notably, without HSCT, some of these diseases lead to fatal outcomes. Annually, a considerable number of children in Sri Lanka are diagnosed as eligible candidates for HSCT.

HSCT involves replacing the patient's bone marrow with hematopoietic stem cells obtained either from the same patient (autologous) or from a different individual (allogeneic). For instance, in cases of high-risk neuroblastoma, replaced Hodgkin lymphoma, and aggressive brain tumors, stem cell rescue follows myeloablative chemotherapy. Allogeneic HSCT is indicated for various conditions, including relapsed or refractory hematological malignancies (such as acute lymphoblastic leukemia and acute myeloid leukemia), bone marrow failure syndromes, hemoglobinopathies (like thalassemia and sickle cell anemia), inborn errors of immunity, and certain inherited metabolic diseases. In many instances, these conditions carry a 100% mortality rate, making HSCT the sole potential cure. Additionally, HSCT significantly reduces morbidity in hemoglobinopathies, and timely intervention not only lessens the burden on hospitals but also increases survival rates.

The process of allogeneic HSCT commences with finding a suitable donor, which could be an HLA-matched sibling, an unrelated donor, or a haploidentical donor. Advances in the field have facilitated successful haploidentical donor transplants, enabling any biological parent to become a potential donor for their child. This development mitigates delays in finding an HLA-matched donor. Stem cells for transplantation can be sourced from peripheral blood, bone marrow, or cord blood. Conditioning chemotherapy, administered before stem cell infusion, aims to induce myelosuppression to create space in the marrow for new stem cells and lymphodepletion to minimize graft rejection. Serotherapy is often added to mitigate graft-versus-host disease.

The immediate post-transplant period witnesses the bone marrow becoming aplastic before engraftment, often complicated by factors such as mucositis, febrile neutropenia, and viral reactivation. Engraftment is confirmed by the presence of platelets, leukocytes, and neutrophils in peripheral blood. Immune reconstitution inflammatory syndrome (IRIS) may occur during immune system reconstitution, manifesting as worsened clinical manifestations of preexisting infections or unmasking of clinically silent infections.

Unique complications following allogeneic HSCT include graft-versus-host disease (GVHD) which can be acute or chronic. Acute GCVD reflects exaggerated but normal inflammatory mechanisms mediated by donor lymphocytes infused into the recipient, whereas, chronic GVHD characterized by impaired immune tolerance mechanisms affecting innate and adaptive immunity. Transplant-associated thrombotic microangiopathy (TA-TMA), autoimmune hemolytic anemia, immune thrombocytopenia are rare but carries significant long-term morbidity to the patient. Organ-specific toxicity, e.g. hepatic, renal, pulmonary, gastrointestinal are commoner but fortunately, mostly they are transient and reversible with prompt management.

Chimerism is checked upon recipient engraftment to confirm the engrafted cells of donor origin. The aim of HSCT is to achieve complete and durable immune reconstitution with a diverse T cell receptor repertoire, essential for reducing infection, disease relapse, and secondary malignancies.

Early referral to the HSCT team is paramount to prevent infections and provide vigorous treatment before end-organ damage sets in. During the initial assessment, maintaining vigilance and inquiring about the family history of infant deaths from a suspected Severe Combined Immunodeficiency (SCID) patient can prevent disseminated BCG infection by avoiding BCG vaccination at birth. Breastfeeding should be withheld in such patients until the mother's Cytomegalovirus status is known. Moreover, initiating the donor search early on is crucial. Newborn screening for inherited disorders of immunity and metabolism, along with family screening of affected children in such conditions and hemoglobinopathies, significantly impacts early diagnosis, facilitating uncomplicated HSCT.

Pretransplant workup aims to evaluate patient comorbidities and control the disease while simultaneously conducting a donor match. For patients with thalassemia, optimal iron chelation before transplant and hyper-transfusion during the peri-transplant period are associated with improved outcomes. Assessing minimal residual disease in leukemic patients following chemotherapy provides a more accurate evaluation of bone marrow status. Vigorous management of tuberculosis and other infective foci prevents IRIS. Optimizing nutrition from the initial diagnosis is critical for long-term HSCT outcomes. Diagnosis should be genetically confirmed where applicable, as it aids in prognostication and may also modify the conditioning regimen, particularly in disorders with DNA repair defects necessitating modified protocols.

The HSCT team is multidisciplinary, comprising clinicians, hematologists, immunologists, geneticists, microbiologists, virologists, mycologists, nutritionists, and pharmacologists. Prompt involvement of radiology and other subspecialties is vital to ensuring successful HSCT outcomes.

8th Rare Disease Day Symposium 2024

Organized by the Rare Disease Forum Sri Lanka College of Paediatricians

1st March 2024 @ New Auditorium, LRH

08.30-09.00 Poster viewing

09.00-09.30 "An approach to a child with LSD – evolving perspectives and experiences": Dr Ratna Puri
Senior Consultant for Medical Genetics, Sir Ganga Ram Hospital, New Delhi

09.30-09.35 Inauguration ceremony

09.35-09.40 Welcome speech: Prof Harendra de Silva
President, Rare Disease Forum, SLCP

09.40-09.45 President's Speech: Dr Kosala Karunaratne
President, SLCP

09.45-10.15 "Beyond subtle dysmorphism" : Prof Ruwanthi Perera
Professor in Paediatrics, FMS, University of Sri Jayewardenepura

10.15-10.45 Genetic testing: an overview for clinicians: Dr Hasani Hewavitharana
Consultant Paediatric Clinical Geneticist (Acting), Lady Ridgeway Hospital for Children

10.45-11.00 Tea break

11.00-11.30 Uric acid & inborn errors of metabolic disorders-clinical experience: Dr Eresha Jasinge
Chemical Pathologist, Lady Ridgeway Hospital for Children

11.30-12.00 Metabolic emergencies in Paediatrics:Dr Imalke Kankananarachchi
Senior Lecturer in Paediatrics, Faculty of Medicine, University of Ruhuna

12.00-12.30 Role of bone marrow transplantation in rare diseases in children: Dr Prabani Maddumarachchi
Consultant in Paediatric/Adolescent Oncology, National Cancer Institute

12.30-12.40 Launch of the e-bulletin

12.40-12.50 Award ceremony

12.50-01.00 Vote of thanks

01.00-01.30 Annual General Meeting

SLCP 8th Rare Disease Day Zoom Link

Meeting ID: 861 5480 2354

Passcode: 429660

MORE INFORMATION

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ABSTRACTS OF POSTER PRESENTATIONS

P01

A CASE OF MICROTIA – ANOTIA ASSOCIATED WITH OTHER CONGENITAL ANOMALIES

M.H. Kumasaru, K.Withanarachachi, Malinda WAD, Balagalla DD, Peiris WMM, Liyanage J, Dharmawardhane H

Introduction: Anotia is a rare congenital anomaly with absence of one or both pinna and external ear canal which occurs during fetal development. Absence or under development of outer ear structures mainly external auditory canal and pinna is seen. Though there is no exact cause to found genetic and environmental factors are contributing. Antenatal usage of retinoic acid has been given as a cause among external factors. It may present as isolated anomaly or in association with other congenital anomalies or syndromes. Anotia associated with VACTREL anomaly and thyroid agenesis will be further rare

Description: Here we Present a case of a neonate with bilateral absent pinna with possible absence of external auditory canal with congenital hypothyroidism due to thyroid agenesis and possible VACTREL anomaly. Baby was having imperforated anus, posterior urethral valve with bilateral hydronephrosis and posterior urethral valve and cardiac lesions namely PDA and perimembranous VSD.

Investigations (if any):Ultrasound of neck revealed thyroid agenesis and antenatal scan done has revealed B/L hydronephrosis. Postnatal MCUG revealed posterior urethral valve.

Progress: Patient is being followed up at clinics under multidisciplinary care.

Discussion: Child was managed with the multidisciplinary team involvement. Imperforated anus repaired and colostomy creation done. Posterior urethral valve ablation was planned later. BSER was planned to assess hearing with the ENT opinion. CT reporting is awaiting to assess the structure of the ear. Thyroxine was started for congenital hypothyroidism.

P02

HOLOCARBOXYLASE SYNTHETASE DEFICIENCY- RARE CAUSE NOT TO MISSED IN AN ACIDOTIC CHILD

Thayani Arooran , Fernando PMS , Dayasiri K , Kitulwatte N , Schroeder Sabine , Jasinge EA

Introduction: Holocarboxylase synthetase deficiency (HSD) is an autosomal recessive disorder affecting coenzyme function of biotin leading to deficiency of all biotin dependent carboxylases. Presentation is within weeks of birth with seizures, alopecia, hypotonia, skin lesions, severe metabolic acidosis, characteristic urine organic acid (UOA) and acylcarnitine profile (ACYP) with normal biotinidase levels.

Description: A 2-month-old baby boy, first born to non-consanguineous parents presented with poor weight gain, skin rash and acidotic breathing. He had metabolic acidosis, ketonuria with normal plasma glucose and lactate. Normal biotinidase levels with high 3-OH isovalerylcarnitine in the ACYP suggested the diagnosis of multiple carboxylase deficiency (MCD). Though he responded initially to biotin therapy, due to defaulted clinic follow-up presented at the age of 8 years with metabolic acidosis (pH 6.98, HCO3- 6.9 mmol/L), hyperammonemia (132 µmol/L) and hypoglycemia (2.1 mmol/L).

Investigations (if any): UOA revealed ketonuria, marked lactic acid (pyruvate carboxylase deficiency), with elevated 3-methylcrotonylglycine, 3-hydroxyisovaleric acid (3-methylcrotonyl-CoA carboxylase) and moderate amounts of 3-hydroxypropionic acid, propionylglycine and methylcitrate (propionyl-CoA carboxylase deficiency) suggestive of MCD. Repeat ACYP confirmed MCD with normal biotinidase levels. Two heterozygous variants of uncertain significance in HCLS gene (c.1607T>C and c. 2188G>T) supported HSD.

Progress: Despite biotin therapy and supportive care, he succumbed to illness on 10th day of illness.

Discussion: This case illustrates importance of considering the rare diagnosis of HSD in a patient with severe metabolic acidosis. UOA, biotinidase levels and ACYP are helpful diagnostic aids even in the absence of genetic testing.

P03

GRISCELLI SYNDROME WITH DOWNS SYNDROME COMPLICATED WITH REFRACTORY SEIZURES, TRABECULATED BLADDER WITH PERSISTENT HYPERKALEMIA FOLLOWING INTERSTITIAL NEPHRITIS

D. T Mihishani Dasanayake,

Introduction: Griscelli syndrome is a rare AR disorder with pigmentary dilution, neurologic
Problems, immunodeficiency. Manifestations are depend on specific genetic defect. This neonate was found to have hair, eye and skin pigmentary dilution, Downs syndrome with trabeculated bladder complicated with deranged renal functions and seizures.

Description: A neonate who was born to healthy nonconsanguineous parents, had silvery gray hair, pale skin, hypopigmented fundi and iris, facial dysmorphism with hypotonia. No family members have silvery gray hair, skin hypopigmentation, genetic disorders or unexplained young deaths. Continued Investigations due to persistent hyperkalemia.

Investigations (if any):
MCUG - An abnormal bladder with a trabeculated wall and multiple diverticula. No evidence of VUR or BOO
Cystoscopy - No PUV.
Karyotyping - Down syndrome due to non-dysJunction. (47,XY,+21)
Serum K – 11 mmol/l
Hair shaft - large irregular clumps of pigments.

Progress: Child was managed with CIC, Calcium resonium and seizures were refractory to AEDs, had recurrent admissions and expired at 9 months of age.

Discussion:
Our differential diagnosis were Ocular Cutaneous Albinism, Griscelli, Chediak Higashi and Hermansky Pudlak syndrome. The presence of giant granules in hair shaft favoured of Griscelli. Progressive neurologic involvement may be due to cerebral lymphohistiocytic infiltration but no features of HLH in initial blood films. Immunology evaluation and WES tests are important. GS1 and GS2 die in childhood due to recurrent infections or neurological sequelae and only curative therapy is HSCT. GS3 has a better prognosis. Since This is a very rare presentation with 2 syndromes, MDT approach is essential.

LUPUS LYMPHADENOPATHY-AN INITIAL PRESENTATION OF SYSTEMIC LUPUS ERYTHEMATOSUS

MAF Shahnaz, DS Wijesekara, WKSI.Wanniarachchi, JASD Madhubhashini, KT Mendis

Introduction: Systemic lupus erythematosus (SLE) is an autoimmune disorder that affects multiple systems, with a female preponderance. Diagnosis is based on 2019 criteria by the European League Against Rheumatism and American College of Rheumatology. Although lymphadenopathy is not a diagnostic criterion, in one-third to a half of cases it is reported to be the initial presentation.

Description: An 11-year-old girl presented with a 2-month history of enlarging tender cervical lymph nodes, worsening joint pain, and daily fever. She had left-sided cervical lymphadenopathy for the last 6 months and an erythematous macular popular rash.

Investigations (if any): Erythrocyte sedimentation rate was 107/1st hour, C-reactive protein was 36.9 with a positive antinuclear antibody titter, and anti-double-stranded deoxyribonucleic acid and low C3 and C4 complements. Multiple enlarged lymph nodes with loss of fatty hilum were evident in the ultrasound scan. Lymph node biopsy revealed reactive hyperplasia of lymph node, with histiocytic & plasma cells proliferation which suggestive of connective tissue disease/SLE-associated changes. Later on, renal biopsy disclosed class 1 lupus nephritis

Progress: She responded to hydroxychloroquine, prednisolone, and methotrexate with regression of the lymph nodes.

Discussion: Lupus lymphadenopathy is typically widespread but rarely, cervical lymphadenopathy could be an early sign of SLE. The possibilities of Kikuchi-Fujimoto disease, Castleman disease, Rosai-Dorfman disease, lymphoma, and secondary infections should also be considered as the outcomes and treatments could vary. Differentiation of these pathologies can be done through histology of enlarged lymph nodes while a renal biopsy confirms SLE

A LUMP IN A NEONATE; CONGENITAL HYMEN PROLAPSE

APH Withana , J Liyanage, UB Liyanage

Introduction: The hymen, a fibrous connective tissue. Separates vaginal lumen from urogenital sinus. The abnormalities are commonly an incidental finding. The incidence of vaginal anomalies is unknown. Hymen prolapses is commonly asymptomatic.

Description: Baby girl born to 23-year-old primigravida mother and 25-year-old father at a local hospital. Transferred to teaching hospital Karapitiya for further evaluation of the lump at this baby's vulva. Term baby Delivered by an emergency LSCS due to breech presentation in labour. Birth weight, 2.680kg and admitted to PBU. Baby passed meconium and urine within first 24 hours of life. On examination, pink active baby without dysmorphic features. Cardiovascular, respiratory, neurological, and abdominal examinations were normal except the lump at vulva. Paediatric surgical referral done. Clinically diagnosed as congenital prolapse of the hymen.

Investigations (if any): FBC, RFT, UFR were normal& Urine culture was negative. USS abdomen and KUB were normal; there was no USS evidence of bicornuate uterus. Planned to do a vaginoscopy at 1/12 of age.

Progress: Since the baby was thrieving well without complications the vaginoscopy was postponed. The baby was observed. With the growth of the baby the lump became a smaller prolapse and clinically confirmed the diagnosis.

Discussion: Commonly the pathogenesis of congenital anomalies of the female genital tract has been experimented in animals. They believe that high maternal oestrogen levels have a role in the growth of hymen. A normal baby with prolapse or enlarged hymen doesn't need any treatment. It can be observed. If it associates with any difficulty in urination or defecation, a partial resection could be considered.

TETRABENAZINE RESPONSIVE MYOCLONUS DYSTONIA DUE TO DYT 11 MUTATION

Harsha Munasinghe , Sanjaya Fernando

Introduction: Myoclonus-dystonia is a movement disorder that typically affects the neck, torso, and arms. Individuals with this condition experience quick, involuntary muscle jerks or twitches (myoclonus). About half of individuals with myoclonus-dystonia develop dystonia, which is involuntary tensing of various muscles that causes unusual positioning. In myoclonus-dystonia, dystonia often affects one or both hands, causing writer's cramp, or the neck, causing the head to turn (torticollis).

Description: A 3 year old male child presented with abnormal jerky movements in both upper limbs and difficulty in walking for 4months. He was the 1st child of non consanguineous healthy parents. Antenatal history was uneventful and delivered by Elective cesarean section at 38 weeks of gestation with 2.5kg of birth weight, and had no perinatal or post natal complications.
He was developmentally normal up to 2years and 2months of age then developed recurrent falls due to left side lower limb weakness. Then gradually involves bilateral upper limbs with jerky movements affecting fine movements.
Neurological examination was clinically normal. His brain imaging,EEG,Nerve conduction studies,Basic metabolic studies were normal. These movements poorly respond to Syndopa and Clonazepam

Investigations (if any): Whole exome genome revealed DYT 11 mutation in SGCE(epsilon-sarcoglycan) gene

Progress: jerky movements significantly improved with Tetrabenazine

Discussion: This case history is important for clinical identification of DYT 11 mutation and guiding for effective management strategies in the context of Dystonia.

UNICENTRIC- MESENTERIC CASTLEMAN DISEASE IN A CHILD; AN UNUSUAL PRESENTATION

WMCL Weerasinghe, Kudagammana S T, Hettiarachchi HCM , Dharmadasa K V C K, Jayaweera A H H M

Introduction: Castleman disease (CD) is a rare non-malignant lymphoproliferative disorder. Two subtypes include unicentric Castleman disease (UCD) and multicentric Castleman disease (MCD). Histological subtypes are hyaline vascular-HVCD (80%–90%), plasma cell (10%) and mixed (2%). CD commonly occurs in mediastinum (70%) and rarely in mesentery. UCD is best managed surgically whereas, MCD requires surgery and chemo-radiotherapy. There are a few reported cases of childhood UCD but none in Sri Lanka.

Description: A 12-year-old previously well boy presented with generalized abdominal pain, dyspeptic symptoms, weight loss and anorexia for one month. He was pale, had angular stomatitis and had a palpable mass in the left hypochondrium.

Investigations (if any): His ESR was 62mm in the first hour and CRP was 84mg/L. Blood picture and iron studies suggested iron deficiency anaemia (Haemoglobin 7.5 g/dl, MCV 58fL) with negative HIV and HHV8 antibodies. CECT of abdomen demonstrated a well-defined mass anterior to the left renal hilum (4cm x 3cm x 2.5cm) arising from root of small intestinal mesentery.

Progress: Histology of the surgically removed mass showed a significantly enlarged lymph node with regressive follicles and concentric onion skin appearance to the mantle zone suggestive of HVCD. No atypical cells were seen. Immunohistochemistry studies confirmed the reactive nature of the follicles.

Discussion: This describes unicentric HVCD at the root of the small intestine mesentery, which is rarely involved. HV-CD demonstrates a good prognosis following complete surgical resection. UCD needs follow up due to risk of lymphoma, a rare possibility of recurrence and anaemia.

CONGENITAL FEMORAL DEFICIENCY(CFD) - MULTI-DISCIPLINARY MANAGEMENT

N.S.K.Weththasingha, Thennakoon D, Sholani THDS, Safinaz ZMFZ, Vipulaguna DV

Introduction: congenital femoral deficiency (CFD) is a condition which includes femur deficiency, discrepancy and deformity. The most severe form is proximal focal femoral deficiency(PFFD). It affects the mobility of the hip joint and malorientation of the joint with soft tissue contractures.

Description: A 1-year-old child presented with a L/S absent femur.He was born as a preterm baby(POA-35+2) via an emergency caesarian section due to foetal distress with a very low birth weight and treated as neonatal jaundice,neonatal sepsis. His development was age-appropriate other than the gross motor development. On examination,short left leg with 4 toes,R/S mildly pronated feet with normal reflexes,good muscle strength noted and other systematic examinations were normal.

Investigations (if any): B/L hip and spine X-ray shows R/S hip dislocation,poor development of the neck of the femur,shallow acetabulum, and mild scoliosis in the lower thoracic area(R/S concave),L/S absent femur.

Progress: There was mild pronation of the right feet but he was weight bearing from the right leg.Used ortho-prosthesis for the left leg and child could weight bear holding on to a bar.Planned to use a standing frame to reduce hyperextension of the right knee and to add proper weight to the hip.As the orthpedic opinion monitoring for the scoliosis and B/L hip,pelvis,knee,ankle joint MRI were Planned.Rotational plasty after 5 years of age,R/S hip reconstruction in less than 4 years of age and below knee orthosis was also planned.

Discussion: CFD should be promptly evaluated and timely managed to prevent gait abnormalities, cosmetic, behavioural, psychological issues.

P09

LANGERHANS CELL HISTIOCYTOSIS IN A CHILD PRESENTED WITH VERTEBRAL FRACTURES

R.D.P.Jayawickrama , P.M.Wijayawardhana, Navoda Athapattu

‘Introduction: Childhood histiocytosis constitutes a diverse group of disorders. Langerhans Cell Histiocytosis (LCH) is the best known.Clinical spectrum varies from single organ to multi system involvement. Skeletal involvement is the commonest(85%). Single organ disease is usually benign, in contrast multi system disease requires multi agent chemotherapy with survival rate of 85%.

Description: A 2 years 8 months old girl presented with bilateral painless neck lumps for 10 days. No infective focus or other lumps. Had LOA and weight was flattening. She was lethargic and dyspneic on excretion, had developed abdominal distention but no features of liver or renal failure. Her perinatal period was uncomplicated. Had developed a painless back lump at 6 months , had given a spinal brace but no further investigations were done. She was on treatment for hypothyroidism since 1 year.
On examination ; she was conscious and rational , not dysmorphic , no rashes , no features of bone marrow hyperplasia or glue ears. there were bilateral , non tender firm cervical lymphadenopathy of 4×3.5cm; moderate sized , firm to hard hepatosplenomegaly and a bony lump at T10 level with kyphosis. Other systems were normal.

Investigations (if any): There was severe anaemia and elevated liver functions ; other biochemistry was normal. Skull X- Ray had lytic lesions , X-ray spine showed collapsed vertebral body of T8 and T10 and generalised hypodensity. Cervical lymph node biopsy had massive sinus histiocytosis more in keeping with histiocytosis X(LCH)

Progress: Baby was transferred to Apeksha hospital after blood transfusion and treatment for sepsis.

Discussion: LCH is a differential which should be taken into consideration with its variable presentations. Bone involvement is the commonest of them. Early diagnosis carries a better prognosis.

A RARE CASE OF NAGER'S ACROFACIAL DYSOSTOSIS

LGC Prabhawitha , S Hassan, DD Balagalla

Introduction: Nager acrofacial dysostosis (NAD) is an inherited disorder of morphogenesis that involves defects of development of 1st & 2nd branchial arches and limb buds. This syndrome mainly consists of craniofacial, musculoskeletal and limb anomalies. Though this is mostly recognized as an entity of sporadic nature, there has been cases reported with autosomal recessive and autosomal dominant inheritance.

Description: A baby girl was born at term with a birth weight of 2.4kg to healthy non consanguineous parents. No family history of syndromes or dysmorphism were noted. Antenatal scans revealed no abnormality. At birth baby had respiratory distress and was initially managed with nasal prongs oxygen , 0.5L/min.
Baby was noted to have severe micrognathia, mandibular hypoplasia, cleft palate, downward slanting palpebral fissures, low set ears, stenosed left external auditory meatus, microtia, hypertelorism, bilateral upper limb deformities, bilateral hypoplastic thumbs, bilateral congenital talipes equinovarus.

Discussion: Nager acrofacial dysostosis (NAD) is a development anomaly that affects musculoskeletal, craniofacial & limb development. The cardinal craniofacial features are downward slanting palpebral fissures, malar hypoplasia, micrognathia, cleft lip, cleft palate, low set ears & external ear defects.
Musculoskeletal and limb anomalies are mainly, preaxial anomalies (hypoplastic or absent thumbs), radial anomalies, radio ulnar synostosis, talipes equinovarous, hallux deformities, absent tibia/fibula, lax hips, syndactyly, polydactyly.
Treacher collins syndrome carries phenotypical similarity, but the main distinguishing features from NAD are the absence of preaxial upper limb defects like thumb anomalies, radio-ulnar synostosis in treacher collins syndrome. Limb anomalies are the cardinal feature of NAD.

P11

A CASE STUDY OF CONGENITAL HYPOTHYROIDISM MISSED BY NEWBORN SCREENING

Amarathunga KKC, Liyanage UB, Atapattu N, Lakmal GGM, Alahakoon AE, Suriyaarachchi SE

Introduction: Congenital hypothyroidism, mainly due to thyroid dysgenesis, is often identified through newborn screening, detecting cases before clinical symptoms arise. The global incidence, initially 1 in 4,000 infants, has risen to 1 in 2,000 with enhanced screening algorithms. Ethnic variations show lower rates in African Americans and higher rates in certain Asian and Hispanic populations.

Description: A 33-day-old term infant, displayed prolonged jaundice on day 33 with deep icterus despite normal birth, breastfeeding initiation, and routine tests. Initial examination showed icterus on the trunk and dorsum of the hands with no signs of hyper or hypothyroidism.

Investigation: Despite on neonatal examination TSH level being normal (5mIU/L) Profound indirect hyperbilirubinemia was evident on admission to the ward on day 33 (total bilirubin 261, direct bilirubin 17).Thyroid function tests indicated elevated TSH (>100mIU/L) and low Free T4 (0.9pmol/L). Neck ultrasound revealed absent thyroid glands, while abdominal ultrasound showed no bile duct or liver abnormalities. CRP, blood culture, urine cultures, and TORCH screening were negative. Urinary function was normal.

Progress: Prompt intervention with Thyroxine 25 µg mane addressed severe hypothyroidism. Subsequent management focused on underlying thyroid dysfunction and complications.

Discussion: This case emphasizes the critical importance of neonatal hypothyroidism screening for early detection. Despite normal routine tests, severe hypothyroidism surfaced, emphasizing potential screening protocol errors. Timely Thyroxine initiation prevents long-term neurodevelopmental consequences, advocating for continuous improvement and vigilance in neonatal screening programs. The case showcases the necessity of refining protocols to ensure accurate detection and early management of congenital hypothyroidism, preventing adverse outcomes in newborns.

A CASE OF HAEMOPHAGOCYTIC LYMPHOHISTOCYTOSIS ASSOCIATED WITH KIKUCHI NECROTIZING LYMPHADENITIS

K.A.D.S.R KARIYAPPERUMA, Liyanage U.B, Maddumarachchi P, Vajira G, Sandaruwan WAA, Chandrasiri NKIM, Amarathunga KKC, Azra MNF

Introduction: Kikuchi's disease which is a rare type of necrotizing lymphadenitis and hemophagocytic lymphohistiocytosis which is a hyperinflammatory state are distinct conditions with different clinical presentations and prognoses but can seldom present in conjunction among children and may have a milder course compared to adults but it can still be fatal in some cases

Description: An 8-year-old boy presented with a persistent high fever and left-sided cervical lymph node enlargement lasting more than 16 days. Additionally, he exhibited right-sided cervical lymphadenopathy, bilateral inguinal lymphadenopathy, and mild hepatosplenomegaly.

Investigation: Diagnostic workup revealed cytopenia affecting more than three cell lines, hypertriglyceridemia, elevated ferritin levels, and positive Epstein-Barr virus IgM antibodies. A bone marrow biopsy demonstrated hemophagocytosis, leading to the diagnosis of EBV-driven HLH based on modified 2009 HLH criteria. Furthermore, a lymph node biopsy confirmed Kikuchi necrotizing lymphadenitis.

Progression: Treatment involved administering intravenous dexamethasone at 10mg daily for two weeks, followed by oral dexamethasone at 5mg twice daily for seven days, with a gradual tapering off of the dose.

Discussion: This case illustrates a rare instance of the co-occurrence of Kikuchi's disease and HLH, particularly in childhood. It highlights the critical importance of swift diagnosis and appropriate management in HLH cases to enhance patient outcomes. Further exploration and research into managing such cases and devising optimal treatment strategies are warranted.

P13

NAPHTHALENE BALL POISONING: A RARE CAUSE OF ACQUIRED METHEMOGLOBINEMIA AND HAEMOLYTIC ANAEMIA IN A CHILD WITH G6PD DEFICIENCY

Sandeepani KKI, Senarathne R, Mohideen A.

Introduction: Naphthalene balls contain potent hydrocarbons. Consumption of even a minimal amount can be fatal. We present a case of a 2 years old toddler who presented with acquired methemoglobinemia and intravascular haemolysis and found to have accidental ingestion of mothballs later on.

Description: A 2 years old girl presented with irritability and dark urine. On examination, She was cyanotic, tachycardic, tachypnoeic with saturation 72% and pale. Abdominal examination and cardiovascular examination were normal. Despite high flow oxygen there was no improvement in saturation. Haemoglobin was 7.2 mg/dl and blood transfusion was given.
She was a developmentally normal child with an uncomplicated birth history. There was no reported haemolysis for prior antibiotics administrations.
She was the only child born to non-consanguineous parents. After several inquiries she recalled that the child was playing with mothballs.

Investigations (if any): Arterial blood gas revealed normal partial pressure of oxygen despite low oxygen saturation. spot blood test and tube test showed chocolate brown colour. Methaemoglobin level was 13% . Blood pictures revealed haemolysis and showed bite cells. She was found to have G6PD enzyme deficiency.
2D Echo and CXR were normal.

Progress: Due to G6PD deficiency she was not treated with Methylene blue. She was kept on ascorbic acid. Methaemoglobin level was reduced to 6% on day 8 and she was comfortable with saturation 84% without oxygen. Parents were counselled to avoid precipitating drugs.

Discussion: Exposure of oxidants may cause methemoglobinemia, especially in G6PD deficient individuals. Timely recognition of subtle signs and symptoms of underlying methemoglobinemia and acute intravascular haemolysis along with prompt supportive measures can improve survival outcomes.

A CASE OF CRETINISM

Sandaruwan W.A.A, Liyanage UB, Jayarathna MI, Darmawardane H, Liyanage A

Introduction: Congenital hypothyroidism, resulting from the absence of thyroxin hormone primarily due to thyroid dysgenesis, is a leading cause of mental and physical retardation. Early identification through newborn screening is crucial, as timely intervention allows affected infants to lead normal lives without significant medical handicaps.

Description: A 1½-year-old child, a monochorionic diamniotic twin, initially managed for lower respiratory tract infection, experienced generalized convulsions. Born at a private hospital with a birth weight of 2.4kg at 36 weeks, the child faced respiratory distress, necessitating 2 months of NICU care. Unable to wean off oxygen and feed properly, the child exhibited acyanotic congenital heart disease with mild pericardial effusion. Missed newborn screening revealed typical symptoms, including developmental delays, medical retractable constipation, excessive sleepiness, feeding difficulties, respiratory distress, umbilical hernia, and a distinct physical appearance with myxoedema, bradycardia, and mottled skin.

Investigations (if any): Elevated TSH (>60mIU), absent thyroid gland on USS neck, pericardial effusion on 2D Echo, cerebral atrophy on CT brain, generalized slowing on EEG, delayed bone age with characteristic radiologic findings, and macrocytic anaemia were revealed.

Progress Levothyroxine treatment was initiated, accompanied by early intervention, symptomatic management, and parental counselling.

Discussion: Emphasizing the essential nature of heel prick newborn screening, this case underscores the potential consequences of missed screenings, particularly in monochorionic diamniotic twins sharing circulation. Global developmental delays warrant comprehensive medical evaluations, as thyroid hormone is critical for normal neurodevelopment. Prompt diagnosis and treatment within the first week are imperative to prevent irreversible brain damage.

A CASE OF HYPERMELANOSIS ITO

Sandaruwan WAA, Liyanage UB, Jayarathna MI, Darmawardane H, Liyanage A

Introduction: Hypomelanosis Ito, also known as pigmentary mosaicism or incontinentia pigmenti achromians, is a rare, inherited multisystem disorder, ranking as the third most common neurocutaneous disease after NF1. Characterized by linear nevoid hypopigmentation along the lines of Blaschko, predominantly on limbs and trunk, it presents at birth or early childhood, with a female-to-male ratio of 2.5:1. This pattern is more easily diagnosed in darker skin, and extracutaneous manifestations, such as genitourinary, cardiovascular, and musculoskeletal issues, may arise from various genetic defects.

Description: The case involves a 7-year-old girl, the first child of healthy non-consanguineous parents, exhibiting developmental regression from 6 months to 3 years, below-average IQ, hypotonia, and distinctive linear whorled nevoid hypermelanosis on the skin. Notably, there is no atrophy of skin patches, and palms and soles remain uninvolved. Additional manifestations include multiple dental issues, mild scoliosis with an absent right kidney and evidence of renal scarring on the left. Spontaneous closure of ASD is observed, while ENT and eye examinations remain normal.

Investigations (if any CECT brain yields normal results, DMSA reveals absent right kidney and superior pole scarring on the left, and USS KUB confirms the absence of the right kidney. 2D echo normal.

Progress: currently on solifenacin and oxybutynin therapy for overactive bladder. Early intervention and parental counseling done

Discussion: Hypomelanosis Ito necessitates symptoms-directed multidisciplinary management due to potential complications arising from associated abnormalities. While cutaneous findings generally indicate a good prognosis, overall outcomes depend on the nature of associated abnormalities, and death is a rare occurrence

GOLTZ SYNDROME-FOCAL DERMAL HYPOPLASIA

W.M.D.M.Peiris, Dilani Dehigama, Hansini Kumasaru

Introduction: Focal dermal hypoplasia (FDH) is a rare mesoectodermal disorder inherited by an X-linked dominant gene. Mutations in the PORCN gene on chromosome Xp11.23 has been implicated as the genetic basis for FDH.

Description: A day 1 term female infant, born of non-consanguineous parents presented with multiple hyperpigmented skin lesions on the trunk and extremities from birth.
General examination revealed microcephaly, multiple hyperpigmented atrophic linear streaks, and macules of varying sizes present on the forehead, neck, trunk, thighs, gluteal region, genitalia, and upper and lower limbs along the lines of back. There was syndactyly of left foot and lobster claw deformity of right foot. A few skin-colored growths were present over the proximal nail fold of the right middle finger and right foot. Nails were dystrophic. Systemic examination was normal.
Based on the clinical, dermatological and skeletal abnormalities, a diagnosis of Goltz syndrome was made

Investigations (if any): Ophthalmological examination and X-ray of long bones were normal.

Discussion: Goltz syndrome is a rare inherited disorder characterized by developmental defects of the skin in conjunction with ocular, dental, and skeletal abnormalities. The hallmark of FDH is thinning of the dermis, which results in depressed linear lesions and outpouchings of the skin caused by herniation of subcutaneous fat. Skin biopsy of an atrophic lesion showed a normal or thinned epidermis. Electron microscopy revealed abnormally scattered collagen bundles. Skeletal defects seen in 60-70% include digits, and vertebral abnormalities. Lobster claw deformity is a major distinct feature.
Multisystem involvement, short stature, mental retardation microcephaly and intestinal abnormalities have reported.

RARE PRESENTATION OF NOONAN SYNDROME WITH EPILEPSY

Madhubhashini JASD, Wijesekara DS, Shahnaz MAF, Mendis KT, Withanage CJ, Wijeratne NG, Deshapriya WSA

Introduction: Noonan syndrome (NS) is an autosomal dominant congenital disorder, affects equally both males and females with normal karyotypes. Prevalence 1 in 1000-2,500 live births and characterized by the distinctive facial phenotype, cardiac defects, short stature and neurological symptoms as cognitive defect, Arnold Chiari malformation and epilepsy.

Description: A 10 years 6 month old girl with history of global developmental delay, congenital heart disease, febrile and afebrile convulsions presented with two episodes of afebrile convulsion. She was defaulted clinic follow-up for 3yrs, not on anti-epileptic treatments. Fits developed while awaking without fever, started with Eyes rolled up, GTC movements lasted less than 5minutes, associated with loss of consciousness, postictal drowsiness and postictal vomiting. She is fifth child of non-consanguineous parents, delivered after uncomplicated antenatal period. Past history of evaluated for thrombocytopnea.
Clinical examination found having short her age (below 5th percentile) ,dysmorphic face with hypertelorism, exophoria, microstomia with thin lips, low hanging columella, short neck, prominent ears, low posterior hair line, shield chest, Wide apart nipples, Wide carrying angle, mid-systolic murmur best herd over pulmonary area, low IQ arrived at a clinical diagnosis as NS.

Investigations (if any): EEG showing generalized epilepsy. MRI brain normal, 2DEcho Right aortic arch with Left diverticulum of Komemerell, mild LPA origin stenosis.

Progress: Management arranged with multidisciplinary team on medical, nutritional, occupational and social aspect.

Discussion: NS diagnosis established with NS multi-gene panel , by genomics testing and Genetic counselling is essential. Regular followups are imperative to monitor cardiac health, growth and developmental progress and screening for coagulation abnormalities.

CASE OF PYLORIC ATRESIA WITH DUODENAL ATRESIA

Sandaruwan WAA, Withanarachchi K, Lakmali VGD ,Malinda WAD , Liayanage J ,Gunawardena PVAI

Introduction: Pyloric atresia, a rare congenital intrinsic obstruction occurring in 1 in 100,000 live births, presents with three distinct anatomical variants. Prenatally, it leads to polyhydramnios and is diagnosed by abdominal X-ray, revealing a dilated stomach without air in the distal bowel.

Description The first-born baby to a chronic diabetic mother with a three-year duration of diabetes presented with polyhydramnios and a distended stomach, delivering at 32 weeks with a birth weight of 2200 grams. Postnatally, the infant exhibited bilious vomiting, a distended upper abdomen, and meconium stool output. Abdominal X-ray revealed a distended stomach without the double-bubble appearance. Laparoscopic surgery at Day 7 revealed a grossly dilated stomach, a small part of D1, and no D2 in the proximal pouch. The lower part of the duodenum was attached to the pancreatic head area dividing the common bile duct. Surgical intervention performing an end-to-end anastomosis of the distal end of the pylorus and the proximal end of D3 and D4 using Kimura's technique.

Investigations (if any): Abdominal X-ray displayed a single bubble appearance, while an upper GI contrast study indicated no contrast beyond the pylorus. Post-surgical contrast study showed no obstruction to the flow of contrast
Progress: Successful surgical treatment using Kimura's technique was established after medical intervention.

Discussion : Antenatal and postnatal detection of pyloric atresia with duodenal atresia, characterized by vomiting and a single bubble appearance on X-ray, emphasizes the importance of medical stabilization before surgical intervention. The chosen surgical technique, Kimura's method, proved effective in resolving the anatomical obstruction

A CASE OF TRANSCOBALAMIN II DEFICIENCY WITH NORMAL B12 LEVELS SECONDARY TO A NOVEL VARIANT IN THE *TCN2* GENE

Charith Prabhawitha, Kaushalya Pussagoda, Imalke Kankananarachchi, Waruna Heshantha, Charith Prabhawitha, Anura Jayawadana, Iresha Jasinghe, Chandima Thewarapperuma, Sabine Schröder, Nayana Liyanarachchi

IIntroduction: Transcobalamin-deficiency is a rare, potentially lethal, autosomal recessive disorder. Currently, twenty-five pathogenic variants in the TCN2 gene have been identified. We present a case of megaloblastic anemia caused by TC II deficiency due to a homozygous nonsense variant in the TCN2 gene (gene OMIM®: 613441): c.471T>G p.(Tyr157*).

Description: A five-month-old baby was treated for failure to thrive and bronchopneumonia. She was the first-born child to second-degree consanguineous healthy parents. Examination revealed features of vitamin B12 deficiency and developmental delay. Investigations revealed pancytopenia. Vitamin B12 was 104.9 pg/mL (200-900). Bone marrow biopsy was inconclusive. The diagnosis was made as vitamin B12 deficiency and intramuscular (IM) vitamin B12 was initiated and continued. The child caught up with the developmental milestones gradually.

At 9 and 16 months, she presented with severe anemia due to 8-week defaulted treatment. Her serum vitamin B12 level was >1000 pg/mL. She recovered with IM vitamin B12 and supportive care. Genetic analysis revealed a homozygous nonsense variant in the TCN2 gene confirming the diagnosis of autosomal recessive transcobalamin II deficiency.

Progress: IM vitamin B12 was given 1- 2 weekly. At present, she is 5 years old and developing normally.

Discussion: Early initiation of aggressive treatment with IM hydroxocobalamin is likely to be associated with disease control and favorable outcomes. The temporary suspension of the therapy leads to the appearance of pancytopenia and its complications.

This highlights that TC2 deficiency can present even with low B12 values in early infancy as well. Moreover, there is a clinical significance of the homozygous c.471T>G variant.

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ANAPLASTIC LARGE CELL LYMPHOMA MIMICKING TUBERCULOSIS; A DIAGNOSTIC DILEMMA

WMCL Weerasinghe, ST Kudagammana, A Herath, AHHM Jayaweera, KVCK Dharmadasa, KPP Kanankearachchi

Introduction: Anaplastic Large Cell Lymphoma (ALCL) is one of the less common types of all Non-Hodgkin lymphomas (NHL). ALCL often presents at an advanced stage with peripheral lymphadenopathy and extra-nodal involvement and accounts for approximately 10 – 20% of NHL in childhood. Common extra-nodal sites include skin, liver, lung, soft tissue and bone. Presentation mimicking tuberculosis is fairly unusual.

Description: A 7-year-old previously well boy presented with a scalp lump for 2 months with a partial response to antibiotics. He had intermittent high fever with drenching sweats for one week. There was loss of weight, anorexia and recurrent respiratory infections. On examination, there was a non-tender lump over the right side (R/S) of the occiput (2.5cm x1.5cm) and matted R/S cervical lymphadenopathy.

Investigations (if any): Initially blood counts were normal with the ESR of 78 . Chest X-ray showed ill-defined inflammatory shadows. Mantoux was negative. Necrotic material and pus were there on drainage of the lump. Cervical lymph node biopsy demonstrated abnormal lymphoid cells.

Progress: Subsequently he developed bilateral pleural effusions, mild pericardial effusion with rising inflammatory markers and blood counts. CECT showed multiple nodular lesions (<5mm) in both lungs and kidneys without mediastinal or para-aortic nodes. Pleural and pericardial aspirates, broncho-alveolar lavage and lymph node biopsy specimens were negative for GeneXpert and acid-fast bacilli. The histology and immunohistochemistry of the lymph node biopsy confirmed an ALCL and the patient was commenced on chemotherapy.

Discussion: This case highlights the need for awareness of ALCL in children with generalized adenopathy and systemic symptoms.

IDENTIFICATION OF PATHOGENIC MORC2 VARIANT IN AN INFANT AND DIFFICULTY IN PREDICTION OF PHENOTYPE

WANSS Wijesooriya , Sanjaya Fernando, Deepthi de Silva

Introduction: Whole exome sequencing (WES) has enabled diagnosis of complex disorders enabling affected patients and families to be informed about prognosis and recurrence risks even when specific therapeutic options are unavailable. This case report discusses a 13-month-old boy with peripheral axonal neuropathy in whom a MORC2 pathogenic variant was identified. Despite this the phenotype remains difficult to predict.)

Description: The 13-month-old male proband is the second child of healthy, unrelated parents. Born by emergency Caesarean section (36 weeks) with weight 2.67kg, OFC 31cm, and length 51cm. Breastfed for 3 months and later expressed breast milk was offered via bottle following refusal to suck. He was weaned at 5 months but prefers blended food. He acquired head control at 3 months and rolling at 4 months but was unable to sit or stand at 13 months. He had a pincer grasp and used 2 words at 13 months. He had absent reflexes.

Investigations (if any): NCS-suggestive of inherited sensory-motor neuropathy. EMG- denervation.
WES -heterozygous pathogenic variant in exon 6 of MORC2 (NC_000022.11: g.30949809G>A (p. Ser25Leu)).

Progress: Discussion: Two phenotypically distinct disorders associated with pathogenic MORC2 variants including Charcot-Marie-Tooth disease (CMT2Z) an asymmetrical peripheral neuropathy with onset in the first decade and progression to motor disability in late-adult life. DIGFAN phenotype includes global delay, impaired growth and IQ, dysmorphism, and neuropathy. This child's developmental problems may be related to CMT2Z but there remains a possibility that the DIGFAN phenotype is present. This phenotypic heterogeneity needs to be included in discussion regarding prognosis.

WISKOTT-ALDRICH SYNDROME; A RARE PRESENTATION IN TWO MALE SIBLINGS: A CASE REPORT

Muhandiram M.R.D.R., Waduge S.K., Pathirana W.M., Salahudeen A., Wicramasinghe W.M.L.N., Eriyagama A.M.S.D., Rajapaksha R.A.V.S.

Introduction: Wiskott-Aldrich syndrome (WAS) is a rare X-linked recessive disease resulting from mutations in the WAS gene that encodes the WAS protein (WASP). It is characterized by primary combined immunodeficiency causing recurrent bacterial, viral and fungal infections, microthrombocytopaenia and atopic dermatitis. Children with Wiskott Aldrich syndrome are at increased risk for development of autoimmune and malignant conditions. Management is focused on providing supportive therapy with appropriate nutrition, skin care for dermatitis and treatment of immunodeficiency with regular IVIG transfusions. Antimicrobial prophylaxis is started early in the course. Hematopoietic cell transplantation remains the only available curative treatment.

Description: A neonate of non-consanguineous parents was detected with purpuric skin rash from day two of life. Subsequently he developed extensive pustules over head and neck region with high spiking fever and generalized atopic dermatitis.
His elder male sibling had been genetically diagnosed with Wiscott- Aldrich Syndrome at four months of age when he presented with recurrent severe infections and microthrombocytopenia . He had serum immunoglobulin levels of increased Ig M, Ig A, IG E and low Ig G. He had been on co trimoxazole prophylaxis and died at age of one year with severe sepsis.

Investigations (if any): Full Blood Count revealed thrombocytopenia with platelet count of 73 x 109/L at birth with lowest platelet of 30x109/L during the course. Blood picture showed microthrombocytopenia . Flow cytometry analysis was significant for low CD 8 cell counts while the CD 4 and CD 19 cell-count remained within normal range. His genetic screening was arranged. HLA phenotyping was scheduled at 3 months of age and Immunoglobulin levels following the 4th IVIG transfusion.

Progress: His acute infection was treated with Iv flucloxacillin for ten days. He was started on monthly immunoglobulin transfusions and long-term prophylactic antimicrobial therapy with oral co-trimoxazole, acyclovir and fluconazole. He is being regularly followed up at dermatology clinic for dermatitis.

Discussion: In low resource countries where confirmatory genetic testing may not be routinely available, it is vital to maintain a high index of suspicion for WAS and start early treatment in male neonates presenting with suggestive clinical manifestations and a positive family history in order to minimize disease related complications.

MERMAID SYNDROME ; SIRENOMELIA TYPE VI - A CASE REPORT

E M D G C H Ekanayake, P V Dissanayake, H M P U Dematawa, K H S Rathnakumara

Introduction: Sirenomelia, also known as "mermaid syndrome" is a rare congenital anomaly with an incidence of 1 : 100,000 pregnancies. Sirenomelia is usually fatal and result in miscarriage, stillbirth or neonatal death.It is manly characterized by varying degrees of lower limbs fusion and is associated with cardiac, genitourinary, gastrointestinal and lumbosacral spine malformations. Sirenomelia is classified into seven groups according to the presence or absence of lower limb bones.Eventhough the exact cause of sirenomelia is not well established, maternal diabetes, drug abuse and teratogenic drug exposure are known associations.

Description: A 26 year old primigravida delivered a baby at term by elective cesarean section due to severe oligohydramnios and intrauterine growth restriction.Prepregnancy folic acid was taken and there was no history of maternal diabetes.Significant congenital anomalies were noted after birth, including fused lower limbs with no feet, absent genitalia and anus and single umbilical artery. The baby also had low set ears, flat nasal bridge and short neck.

Investigations (if any): X-ray of the whole body revealed single femur and tibia, sacral agenesis, and lung hypoplasia.

Progress: The newborn expired at the second hour of life, despite optimal resuscitation.

Discussion: Sirenomelia can be diagnosed by prenatal ultrasound scans as early as 13 - 14 weeks of gestation and antenatal counselling is recommended. However, when there is significant oligohydramnios, the antenatal diagnosis is more difficult.Regular antenatal follow up with maternal blood glucose monitoring and avoidance of teratogenic drug exposure may help in preventing this lethal condition.

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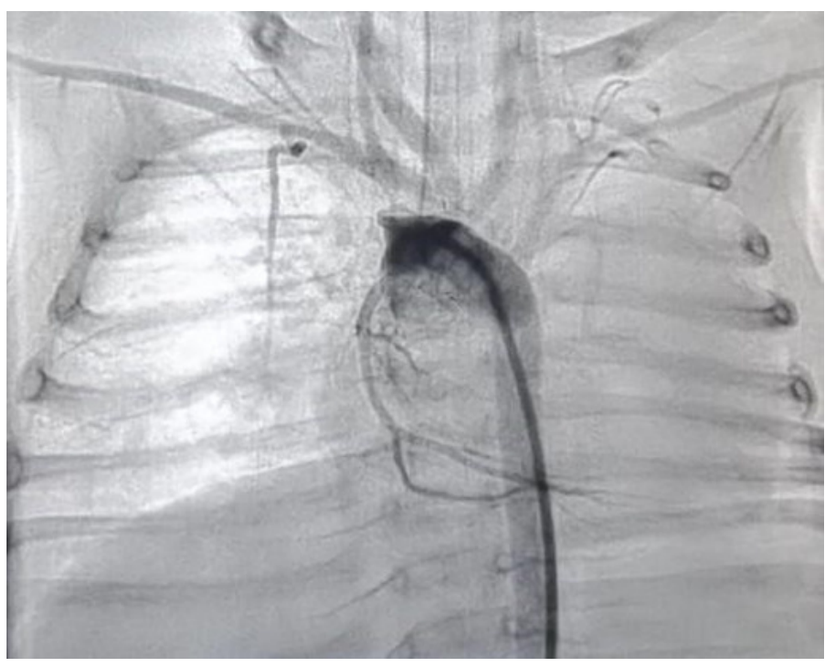
DOUBLE AORTIC ARCH: A RARE BUT TREATABLE CAUSE OF STRIDOR

Puvana A , Hewageegana M, Channa de Silva, Samarasinghe D, De Soysa M, Warusapperuma CR

Introduction: Vascular rings represent 1-2% of all congenital heart diseases. Double aortic arch is a most common type of vascular ring (40%). It encircles the trachea and oesophagus, often resulting in variable airway and oesophagus compression. It is due to the failure of regression of the right fourth arch during embryonic aorta development. In development, the remnant of the right fourth arch will later become the right innominate artery, left fourth arch becomes the aortic arch.

Description: 4-month-old baby who was complicated with persistent respiratory distress which needed high flow nasal cannula oxygen following VSD closure. During ICU stay, he had infrequent stridor. Micro-Laryngo-Bronchoscopy was suggestive of a pulsatile significant mid tracheal anteroposterior narrowing of trachea. Therefore, he underwent bronchogram, aortogram and Computed Tomography Angiogram to delineate the arch and airway anatomy. It was double aortic arch with left dominant arch with significant tracheal compression.

Investigations (if any):



Duplication of aorta

Progress: He was successfully undergone division of the right arch. Post operative period was uneventful.

Discussion: Clinicians should strongly consider the possibility of a congenital vascular ring compression should an infant with a normal upper airway present with stridor. A precise diagnosis can be made by radiological examination. Any stridor is to be evaluated with ENT team properly.

SILVER-RUSSELL SYNDROME, A RARE CONGENITAL IMPRINTING DISORDER

Mendis KT., Wijesekara DS, Deshapriya WSA, Madhubhashini JASD, Withanage CJ, Wijeratne NG

Introduction:
Silver-Russell syndrome (SRS) is a rare congenital imprinting disorder caused by heterogeneous genetic abnormalities: maternal uniparental disomy of chromosome 7, hypomethylation in the imprinting region of chromosome 11 and submicroscopic chromosomal aberrations giving rise to characteristic phenotype of intrauterine growth retardation, feeding difficulties, restricted postnatal growth, facial dysmorphism and body asymmetry.

Description:
A three and half years old boy of consanguineous parents who was born as a term IUGR presented with delayed gross motor milestones. He runs unsteadily, needs help in climbing stairs and is unable to paddle a tricycle, but age appropriate in other developmental domains.
A diagnosis of SRS was made on clinical grounds with characteristic triangular facies, prominent forehead, relative macrocephaly, micrognathia, low set ears and dental malocclusions. He is short for his age (below -3 Standard Deviation (SD)), has growth failure (weight for height below -3SD), asymmetric undergrowth of upper and lower limbs with clinodactyly.

Progress:
Follow up under a multidisciplinary team was arranged with an emphasis on nutritional and physiotherapy aspects.

Discussion:
Although whole exome sequencing and genetic analysis is available, disorder is primarily diagnosed by clinical criteria such as Netchine - Harbison clinical scoring system, as 40% of cases with classic phenotype does not reveal a specific genetic abnormality with currently available molecular diagnostics. Management of SRS requires a multidisciplinary approach with genetic counselling, nutritional support, developmental surveillance, physical, occupational and speech therapy. Therapeutic options such as deformity correction surgery and growth hormone replacement is considered in clinically indicated patients.

METACHROMATIC LEUKODYSTROPHY, A RARE LYSOSOMAL STORAGE DISEASE

Wijeratne NG , Wijesekara DS, Withanage CJ, Mendis KT, Madhubhashini JASD, Deshapriya WSA

Introduction: Metachromatic Leukodystrophy (MLD) is a rare autosomal recessive lysosomal storage disease causing progressive demyelination of the central and peripheral nervous systems. It has three types, Late infantile, Juvenile and adult form, each having different ages of onset and presenting symptoms but characterized by loss of motor and cognitive skills.

Description: A 3 year old previously well girl, presented with ataxia, increased sleepiness and frequent falls, followed by progressive deterioration of all developmental domains. 2 years later she developed afebrile convulsions. Antenatal, perinatal and family histories were unremarkable.

Investigations (if any): Genetic testing revealed a genetic diagnosis of autosomal recessive MLD. MRI Brain showed bilateral symmetrical periventricular deep white matter T2 and FLAIR hyperintensities with trigoid appearance in both cerebral hemispheres involving the corpus callosum and bilateral posterior limbs of the internal capsule, suggesting MLD.

Progress: Currently at 7 years of age she has quadriparesis, complete loss of speech, hypotonic extremities and diminished tendon reflexes. A multidisciplinary follow-up is arranged where she receives occupational/physical therapy.

Discussion: A diagnosis of MLD was made due to the course of symptoms, MRI and genetic results, in this case Late infantile form. MLD is caused by a deficiency of Arylsulfatase-A which is required for the hydrolysis of sulfated glycosphingolipids, resulted by mutations in the Arylsulfatase-A gene located on chromosome 22q13.33. This leads to accumulation of cerebroside sulfate within the myelin of central and peripheral nervous systems. There is no curative treatment for MLD but allogenic haematopoietic cell transplantation combined with gene therapy is suggested.

A CASE OF DIGEORGE SYNDROME WITH SEVERE COMBINED IMMUNE DEFICIENCY(SCID) LIKE PICTURE

Kumudesh EAS , Kumudesh EAS, TPGJ Shantha, Liyanage UB, Gajaweera H, Rajiva de Silva, Dhanushka Dasanayake

Introduction: DiGeorge Syndrome is a rare disease caused by the deletion of chromosome 22q11.2 leading to disruption of 3rd & 4th pharyngeal pouches during early embryogenesis, resulting hypoplasia or aplasia of thymus, parathyroid glands and with other structural anomalies including congenital heart disease. Severe combined immunodeficiency (SCID) is a rare genetic disorder characterized by disturbed function of T cells & B cells.

Description: 4 months old, baby girl, first born child of nonconsanguineous parents, presented with severe respiratory distress. Antenatal period was uncomplicated. On D2 of life hypocalcaemic convulsions noted with primary hypoparathyroidism, with absent thymus on chest x ray. Echocardiogram was normal and had subtle facial dysmorphism. Calcium levels normalized with Calcium supplementation & no further hypocalcaemic convulsions. Up to 3 months of age baby was clinically well. At age of 3 months developed severe bronchopneumonia. With 2nd presentation of bronchopneumonia managed with multiple antibiotics, antivirals, antifungals, anti TB treatment, IV Immunoglobulin with multidisciplinary approach.

Investigations (if any): Flow cytometry: CD4 =73 (1400 – 5100), CD8 38 (600-2200); Ig G 104 (206 – 1125)

Progress: Poorly resolving pneumonia ultimately needed intensive care. Initially no evidence of infection; later blood culture was positive for Burkholderia cepacia. Despite of appropriate treatment & continuous supportive care baby succumbed to their illness at age of 5 months.

Discussion: DiGeorge syndrome ranges, from normal T-cell to complete deficiency named as complete DiGeorge Syndrome which occurs rarely. Furthermore, complete DiGeorge syndrome with hypogammaglobulinemia behaves like T-negative SCID like picture(extremely rare<1% DiGeroge), with severe susceptibility to opportunistic pathogens with high mortality.

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WILLIAMS SYNDROME, A RARE MICRODELETION DISORDER

Withanage CJ, Wijesekara DS, Shahnaz MAF, Wijeratne NG, Mendis KT, Madhubhashini JASD, Deshapriya WSA

Introduction: Williams-Beuren syndrome (WBS) is a rare microdeletion at chromosome 7q.11.23 with multisystem involvement. Affected individuals exhibit a broad spectrum of symptoms and physical features including classic “elfin-like” facies, cardiac, renal involvement, failure to thrive and hearing impairment.

Description: A 11 month old girl presented with marginal developmental delay, poor eye contact, repetitive hand movements, hypotonia, dysmorphism and loquacious personality. She was born as a term Intra Uterine Growth Restricted baby whose mother had hyperthyroidism, and her neonatal period was complicated with jaundice and hypoglycaemia.

Investigations (if any): 2D Echocardiogram showed Ostium Secundum ASD with mild bilateral branch pulmonary artery stenosis. Ultrasonically kidneys and urinary system were normal. Hypothyroidism and hypercalcaemia were excluded. DNA extracted from the venous blood revealed 7q11.23 microdeletion compatible with WBS.

Progress: Currently at 2 years of age she stands with support, has seven – eight words in her vocabulary and no mature pincer grasp. A multidisciplinary follow up is arranged and her developmental milestones are improving with physical/occupational therapy.

Discussion: A diagnosis of WBS was made by characteristic physical features and DNA testing. WBS could be sporadic or familial resulting from a heterozygous deletion of approximately 1.6 megabases containing 26-28 genes at chromosome 7q11.23. Hemizygosity for the elastin gene (ELN) and LIM-Kinase 1 (LIMK1) is responsible for vascular stenosis like supravalvular aortic stenosis, pulmonary valvular stenosis and visuospatial construction cognition respectively. Intellectual disability, reduced verbal and memory performance is expected in these patients. Screening for possible complications would be beneficial for more favourable outcome.

A CASE OF TOTAL COLONIC AGANGLIONOSIS COMPLICATED WITH NECROTIZING ENTEROCOLITIS

Rukshani WM, Jayawardana PP, Withanaarachchi K, Sandaruwan WAA, Kasthuri S

Introduction: Hirschsprung disease is a congenital motor disorder of the enteric nervous system, characterized by absence of ganglion cells in the submucosal and myenteric plexus. Total colonic aganglionosis is an uncommon form of Hirschsprung disease. Relaxation failure of the aganglionic segment causes affected infants to present with intestinal obstruction. Diagnosis is confirmed by rectal biopsy.

Description: The first-born baby to healthy non consanguineous parents who was antenatally diagnosed with distal small bowel obstruction at 39 weeks of gestation, was delivered by emergency section with 3150g birth weight. Postnatally the neonate had bilious vomiting and abdominal distension with no meconium output. Exploratory laparotomy on day one revealed distended small intestine while histology revealed absent ganglion cells of the whole colon and distal ileum. Surgical intervention included insertion of an ileostomy.

Investigations (if any): Abdominal X-ray on day one displayed grossly distended small bowel while radiographic fluroscopy at one week revealed stricture involving distal descending colon and proximal sigmoid colon. Lower GI contrast study indicated no contrast flow beyond the distal sigmoid colon. Ostium secundum atrial septal defect with a patent foramen ovale were detected during echocardiography. Ultrasound studies of the brain and renal system were normal.

Progress: While awaiting ileostomy pull through surgery, the infant re-admitted at day forty with features of necrotizing enterocolitis. Prompt treatment lead to effective disease resolution.

Discussion: Total colonic aganglionosis poses significant challenges in the management requiring early diagnosis. Enterocolitis is the leading cause of death in these patients. Hydration, bowel decompression and antibiotics are essential in the management of enterocolitis.

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DEVASTATING HYPOGLYCEMIA IN CHILDHOOD TO DIABETES MELLITUS IN ADOLESCENCE : A CASE OF GCK MUTATION

T.J.Hoole, I.Jayasundara, A.K.Nimanthi, R. Balasubramanium, D.S. Gamage, A.K.T.N.de Silva, N. Atapattu

Introduction: Glucokinase (GCK) is a key pancreatic beta-cell sensor that plays a crucial role in the regulation of insulin secretion. Mutations in the gene can cause both hyperglycemia and hypoglycemia.

Description: 9 year old boy was referred from the ENT unit due to hypoglycemia with high serum insulin level of 109.2 pmol/l following increased sleepiness. He was born to non-consanguinous parents. He was on antiepileptics since the age of 4 months. Blood sugar values at that time were not available. At 3 years of age, non-ketotic hypoglycemia with normal free fatty acid profile was detected after an episode of unresponsiveness. The mother was advised on frequent feeding. On presentation, the child was obese, with no organomegaly. He had global developmental delay and hyperactivity. Hypoglyceima was responsive to Diazoxide with hydrochlorothiazide.

Investigations (if any):
Protein challenge test- Serum Ammonia- 53 IU/L
Next generation sequencing – mosaic activating GCK mutation with in-frame duplication on exon 10.
While on treatment-
Na- 145mmol/L
K. 3.5mmol/L
2D-echocardiogram-normal
At 14.5 years-
Bone age- 12-years
FSH 2.3 IU/L
LH 0.09 IU/L
At 15 years-
HbA1C-6.4%

Progress: Marked improvement was noted in his development, although the learning disability persisted. Antiepileptics were tailed off. At 14.5-years-of-age, intramuscular testosterone was commenced for delayed puberty. At 15 years of age, he developed diabetes mellitus, and was started on Glicazide.
Discussion: Patients with congenital hyperinsulinism can develop maturity onset diabetes later. But in this case, the onset has been very rapid and needs further genetic evaluation as activating mutations go without being detected.

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TETRALOGY OF FALLOT WITH ABSENT PULMONARY VALVE SYNDROME: A PATIENT WITH RECURRENT WHEEZING

Puvana A , Hewageegana M, Channa de Silva, Samarasinghe D, Singappuli K, Warusapperuma CR, Menaka A

Introduction: Absent pulmonary valve syndrome (APVS) is a rare congenital cardiac malformation. It has an incidence of 3-6% in cases of tetralogy of Fallot (TOF) and 0.2-0.4% of live-born infants with congenital heart disease.

Description:A 2-year-old child was treated for recurrent LRTI mainly with bronchospasm which was not responded to inhalers and the treatment of heart failure. He is a second born child of non-consanguineous parents with uncomplicated birth history. This time, he presented to hospital for TOF and absent pulmonary valve repair. On examination he was tachypneic with respiratory distress. He had bilateral rhonchi. hyperdynamic chest, systolic murmur in addition to hepatomegaly and always comfortable in seated position. His chest X-ray showed cardiomegaly with enlarged pulmonary conus with plethoric lung. CECT chest showed the evidence of cardiomegaly and PHT with massive dilatation of the main pulmonary trunk, right and left pulmonary artery, compressing both side main bronchus with bilateral segmental collapses.

Progress: After surgery, the child was relatively free from bronchospasm and LRTI.

Discussion: The clinical features in infants with APV include wheezing, right ventricular failure, emphysema, atelectasis, pulmonary infection and rarely sudden death. Thus, prognosis in children with APV is related to the extent of tracheobronchial compression by the dilated central pulmonary arteries and right ventricular failure. The corrective surgery with or without replacement of pulmonary valve done in older children may be useful in TOF with APVs. The cause of recurrent wheeze in this child is probably due to large dilated right pulmonary artery compressing the right main stem bronchus.

READING SKILLS OF SCHOOL-AGED CHILDREN WITH FRAGILE X SYNDROME : A CASE SERIES

Fonseka, D, Amarasinghe, A, Jayathilake, H, Ampemohotti, N ; Lokubalasuriya, T ; Vipulaguna, D

Introduction: Fragile X syndrome manifests significant reading difficulties in individuals. This abstract presents the reading skills of three school-aged male children with FXS who are 12 (A), 14 (B), and 16 (C) years old. A and B are siblings. They attend mainstream classrooms and are being followed up by a multidisciplinary team.

Description: Reading fluency, accuracy, and phonological awareness are below the expected level for all three children. Reading comprehension of the 16-year-old child is in grade one level. A and B had mental age of 5-6 years. Their reading comprehension was at the pre-primary level. All three children didn't have any medical comorbidities affecting their reading skills.

Progress: All three children attended regular learning support sessions. Functional literacy strategies were started with all three children. According to the recent assessment, the reading comprehension of the 16-year-old had improved up to grade 03 level (2 grade levels) and the other two to Grade 1 level, with visual prompts.

Discussion: This abstract emphasizes the impact of functional literacy and learning support to improve the reading skills of children with FXS studying in secondary grade level. Even though there is an improvement in reading with verbal and visual prompts, the difficulties in phonological awareness might have affected the reading improvement. Education-focused appropriate multi-disciplinary interventions are effective in improving the overall functionality of children with FXS.

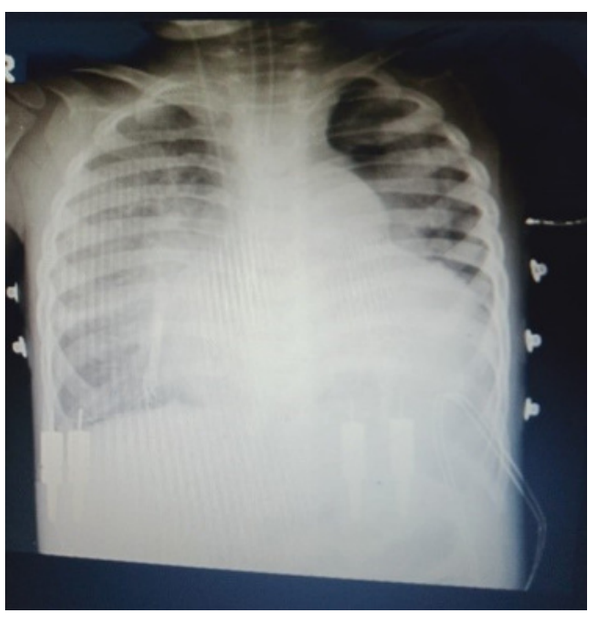
MEDIASTINAL FOREGUT DUPLICATION CYST: A RARE CAUSE OF RECURRENT RESPIRATORY DISTRESS

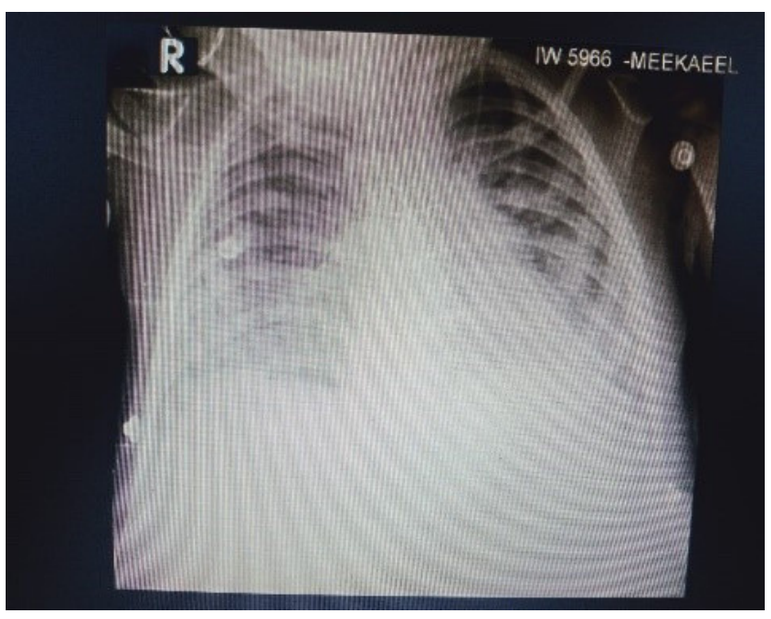
Puvana A , Rasnayake D, Channa de Silva, Vicknarajah S, Prashana R, Warusapperuma CR, Arunath V

Introduction: Foregut duplication cysts are extremely rare benign lesions that contributes 15% of all mediastinal tumours. It is classified into bronchogenic cyst, neuroenteric cyst and other enteric cysts. They represent an abnormal budding from either dorsal or ventral portion of the primitive gut. They can be presented with variety of symptoms which are usually related to compression of the airways or the oesophagus. The Complications of them are infection, pulmonary artery stenosis, superior vena-cava obstruction and peptic ulceration.

Description: A 1 year and 4-month-old baby presented with stridor, cough and cold for two weeks. On examination, he had right side reduced air entry in addition to stridor and severe respiratory distress. He had right side upper mediastinal mass on chest X ray. Computed Tomography was suggestive of well-defined cystic lesion measuring 4.5*5*6.4cm noted in the middle mediastinum with the significant compression of mid trachea. Ultrasound scan guided aspiration of cyst was done which is suggestive of infected foregut duplication cyst. After aspiration, symptoms transiently settled. Because of recurrence of symptoms and almost static size of cyst on repeat CT scan, Cyst excision was done. During the Post operative period, he had some speech and swallowing issues and his repeat chest X-ray was normal.

Investigations (if any):





Progress: He recovered completely with time.

Discussion: Foregut duplication cysts should be considered in the differential diagnosis of any child or infant have middle or posterior mediastinal mass. Surgical excision is recommended at the time of discovery to prevent the development of complications and to avoid the possibility of malignant transformation.

A CASE OF LANGERHANS CELL HISTIOCYTOSIS

Lakmali VGD , De Silva MAH, Gajaweera H, Sulakshi RU, Sandaruwan WAA, Dissanayake SKVP

Introduction: Langerhans cell histiocytosis is a rare class of childhood histiocytosis, affecting any age with variable clinical manifestations .Hallmark is presence of a clonal proliferation of Langerhans cells. Diagnosis by histopathological examination. Treatment depend on systems involved and severity

Description: 1 year old girl born to healthy nonconsanguineous parents, with unremarkable antenatal, perinatal and postnatal history presented with lower respiratory tract infection with respiratory distress requiring respiratory support throughout. She was having hypopigmented scaly rash over scalp, genital area. She got admitted to local hospital twice at 9months and 10months, treated as bilateral blepharitis, failure to thrive and membranous conjunctivitis, infantile seborrheic dermatitis respectively.

Investigations (if any):X Ray chest bilateral reticular shadows with cystic formation, HRCT cystic lung disease with extensive involvement of both lungs compatible with pulmonary Langerhans cell histiocytosis , scalp skin biopsy favour cutaneous Langerhans cell histiocytosis , bone marrow biopy reactive, Skeletal survey normal

Progress: after medical and nutritional stabilization child was transferred to Apeksha Hospital and chemotherapy started. Died due to severe lung involvement

Discussion: LCH affects many organs, bone (80%), Skin (33%), pituitary (25%),Liver (15%), spleen(15%), lungs (15%), hematopoietic system (15%), lymph nodes (5-10%).Pulmonary infiltrates can be from diffuse fibrosis and disseminated nodular infiltrates to diffuse cystic changes. Although lung is not a risk organ, can be fatal through complications as uncontrolled pneumothorax , chronic emphysematous changes. Early diagnosis and treatment improve survival. Single system disease has chance of spontaneous remission or requires only local therapy, multisystem disease requires multiagent chemotherapy

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GOLDENHAR SYNDROME- OCULO-AURICULO-VERTEBRAL SYNDROME (OAVS)

W.M.D.M.Peiris , Dilani Dehigama, Wasana Mihirani, Waruna Heshantha

Introduction: Goldenhar syndrome is a rare condition which is characterized by a multitude of anomalies involving craniofacial structures, vertebrae, internal organs. The etiology is multifactorial with genetic. Herein, we report a case of Goldenhar syndrome with hemifacial microsomia and microtia along with systemic involvement which was clinically and radio-graphically assessed.

Description: Preterm baby delivered at 34 weeks of gestation born to 1st degree consanguineous parents admitted to NICU for management of respiratory distress.
The newborn is detected to have facial asymmetry with underdevelopment of jaw causing wide mouth with limbal dermoid present in right eye. Microtia along with preauricular tags was present on both sides. On examination there was cardiac murmur with spo2-87% on room air. 2D Echo revealed cyanotic heart disease with Tetralogy of fallots(TOF) physiology and OS ASD.

Investigations: Xray of vertebrae shows T6 abnormality. USS brain detected to have B/L periventricular changes with grade II PVL.USS of abdomen was normal and thyroid scan detected to have thyroid agenesis

Discussion: Goldenhar syndrome oculo-auriculo-vertebral syndrome (OAVS), is characterized by a triad of accessory tragi, mandibular hypoplasia, and ocular dermoid as in our case. The aetiology is multifactorial, autosomal dominant or recessive inheritance.
Abnormal vascular supply of embryo, disrupted mesodermal migration can lead to defective formation of first and second brachial arches and vertebral system as evident in vertebral and thyroid abnormality.
Systemic involvement in Goldenhar syndrome can vary. Among cardiovascular anomalies, tetralogy of Fallot and ventricular septal defects are most commonly associated with OAVS.

MICRODUPLICATION OF CHROMOSOME 2Q24 INVOLVING SCN1A AND SCN9A - A RARE CAUSE OF DEVELOPMENTAL EPILEPTIC ENCEPHALOPATHY

L.N.A.J.P.Nissanka , M.Kajan, Sanjaya Fernando, Channaka Dantanarayana, Deepthi de Silva

Introduction: Among nearly 1000 genes associated with developmental epileptic encephalopathy (DEE), genes clustered in chromosome 2q24 containing SCN1A, SCN2A, SCN3A, SCN7A, and SCN9A are expressed in voltage gated sodium channels. Pathogenic sequence variants or deletions of these genes are common causes of DEE while chromosome 2q24 duplications are rare. This case reports an infant with DEE associated with the smallest described chromosome 2q24 duplication.)

Description: The female proband is the second-born child to non-consanguineous parents. She was delivered by elective Caesarean section at 38 weeks, with a birth weight of 2.95 kg. Seizures started at 6 weeks with a semiology of generalized tonic-clonic convulsions. At 2.5 years, she experiences daily events with various semiologies and is on four antiepileptic medications. She has not achieved head control, developed purposive hand use or language. Brain MRI scan (age 1 year) revealed left mesial temporal sclerosis. Current EEG is consistent with moderate cerebral dysfunction with focal epilepsy.

Investigations (if any): Whole exome sequencing (WES) identified a 0.23Mb microduplication (chr2:166.041.231-166.307.074) encompassing all coding exons of SCN9A but excluding promotor and non-coding initial exons in addition to the promotor and exon 1-13 of SCN1A.

Discussion: The proband’s DEE is likely caused by the duplication with partial over expression of SCN1A. The role of SCN9A duplication involving most of the coding exons minus the promoter needs to be further elucidated. In other reported cases, the seizure frequency improved, but cognitive dysfunctions remained severe. No evidence of congenital pain syndromes associated with SCN9A mutations was present.

DOUBLE AORTIC ARCH PRESENTING WITH RESPIRATORY DISTRESS FOLLOWING MILK ASPIRATION

HA Nasreena Bulkees, MMT Rajeew Ananda, P Arumugam, HGH Udara

Tracheal compression by vascular structures in infants is uncommon and may be masked by nonspecific respiratory symptoms. Double aortic arch (DAA) is a rare type of congenital aortic arch anomaly, affecting approximately 0.005% ~ 0.007% of fetuses, while the prevalence of right aortic arch (RAA) is estimated to be 0.1. When it occurs, the connected segment of the aortic arch and its branches encircle the trachea and esophagus, leading to symptoms related to these two structures.

3 month old female infant with the background history of congenital stridor since day 1 of life which has improved over time presented with acute onset Shortness of breath. At presentation the infant was in respiratory distress; auscultation mono phonic rhonchi with features of possible milk aspiration and the chest x ray showed mild hyperinflation. With worsening respiratory distress the child required invasive ventilation. However, with the conventional ventilation the child had persistent hypercarbia with normal oxygen saturation and difficulty in delivery of tidal volume which improved with manipulation of ET tube towards carina adjusting to the lip level of 12.5mm, which led to suspicion of tracheal obstruction.
Micro laryngoscopy and bronchoscopy revealed lower tracheal antero posterior narrowing, bronchogram showed dynamic obstruction of trachea and the CT angiogram confirmed the presence of vascular ring encircling the trachea and esophagus.

Conclusion :
Stridor in a newborn should necessitate an immediate workup and the double aortic arch should be considered in the differential diagnosis because it is a treatable condition with excellent prognosis and low mortality.

GENETICALLY CONFIRMED CASE OF INTERMITTENT MAPLE SYRUP URINE DISEASE IN SRI LANKA: AS A MIMIC OF MENINGOENCEPHALITIS

Kullugammana MBCN, Zoysa WRS, Karunaratne KW, Yapa DWK

Introduction: Maple syrup urine disease (MSUD) is a rare inborn error of metabolism due to Disorder of decarboxylation of branched chain amino acids (BCAA) with global incidence of 1 in 185000 live births. There are several variants depending on the clinical severity. Though classic type manifest during early infant period, individuals with other variants like intermittent type can be normal except during intercurrent illness.

Description: Five years old boy first child of the second-degree consanguineous parents, presented with fever, poor feeding, and drowsiness for three days. There was a similar episode at 4 years of age managed as meningoencephalitis with normal CSF studies. Perinatal period was uneventful with normal development. No dysmorphism, growth parameters were normal. No neck stiffness or focal neurological signs. He was tachypnoeic without lung signs but normal Cardiovascular, abdominal examinations.

Investigations (if any): High anion gap metabolic acidosis, ketosis and mild hyperammonaemia. Contrast tomography of brain is normal. Further, plasma amino acid analysis, urine organic acid analysis is suggestive of maple syrup urine disease, and it was confirmed genetically.

Progress: He recovered from acute crisis following haemodialysis and during follow up had normal neurodevelopment.

Discussion: MSUD is an autosomal recessive condition due to deficiency of any subunit of branched chain alpha ketoacid dehydrogenase enzyme complex. Out of four subtypes, classic type is early onset with more severe neurological sequalae. But in intermittent type seemingly normal children develop encephalopathy with intercurrent illness. Treatment is similar as classic form, the elimination of BCAA from tissues and body fluids.

HYPOGLYCAEMIC CONVULSIONS DUE TO INSULINOMA IN A TEN-YEAR-OLD GIRL- A CASE REPORT

M. W. A. Nimanthi, Thabitha Hoole, Sumudu Seneviratne

Introduction: Maple syrup urine disease (MSUD) is a rare inborn error of metabolism due to Disorder of decarboxylation of branched chain amino acids (BCAA) with global incidence of 1 in 185000 live births. There are several variants depending on the clinical severity. Though classic type manifest during early infant period, individuals with other variants like intermittent type can be normal except during intercurrent illness.

Description: Five years old boy first child of the second-degree consanguineous parents, presented with fever, poor feeding, and drowsiness for three days. There was a similar episode at 4 years of age managed as meningoencephalitis with normal CSF studies. Perinatal period was uneventful with normal development. No dysmorphism, growth parameters were normal. No neck stiffness or focal neurological signs. He was tachypnoeic without lung signs but normal Cardiovascular, abdominal examinations.

Investigations (if any): High anion gap metabolic acidosis, ketosis and mild hyperammonaemia. Contrast tomography of brain is normal. Further, plasma amino acid analysis, urine organic acid analysis is suggestive of maple syrup urine disease, and it was confirmed genetically.

Progress: He recovered from acute crisis following haemodialysis and during follow up had normal neurodevelopment.

Discussion: MSUD is an autosomal recessive condition due to deficiency of any subunit of branched chain alpha ketoacid dehydrogenase enzyme complex. Out of four subtypes, classic type is early onset with more severe neurological sequalae. But in intermittent type seemingly normal children develop encephalopathy with intercurrent illness. Treatment is similar as classic form, the elimination of BCAA from tissues and body fluids.

PLEUROPULMONARY-BLASTOMA PRESENTING AS A MASSIVE PNEUMOTHORAX- AN UNUSUAL PRESENTATION; A CASE REPORT

WMCL Weerasinghe, HCM Hettiarachchi, WAK Wanasinghe, MGDVK Kiridana

Introduction: Pleuro-pulmonary blastoma (PPB) is a rare, malignant tumour that originates from either lungs or pleura. The age of presentation is usually less than 7-8 years. The patients may present with fever, dyspnoea, cough, constitutional symptoms or symptoms due to brain metastasis. Presentation as a massive pneumothorax is fairly unusual.

Description: A 3-year-old previously well girl presented with progressive shortness of breath and cough over 2 weeks. There had been loss of weight and anorexia for 2 months. There was a family history of breast carcinoma but no familial PPBs. On examination, mild respiratory distress was noted with left sided tracheal deviation and diminished breath sounds on the right side. On air, saturations were maintained in spite of tachypnoea.

Investigations (if any): Chest X-ray showed right sided cystic lesion, pleural effusion with right lung collapse. CT chest showed a large right sided pneumothorax with complete collapse of right lung, mediastinal shift and moderate pleural effusion. Extensive bullous formation in the right middle and lower lobe was seen.

Progress: Child was transferred to the intensive care unit after inserting an intercostal tube. Bronchoscopy excluded foreign body aspiration. Once stabilized, thoracoscopy and resection of the bullous lesion of the R/S middle lobe was done. Histology showed Pleuro-pulmonary blastoma type II. Child was started on chemotherapy.

Discussion: In retrospect, the patient was clinically well so that the degree of pneumothorax and the presentation was very unusual. Family screening for genetics is also warranted in familial PPBs.

A CASE REPORT OF TAKAYASU ARTERITIS

Suriyaarachchi S.E., Liyanage U.B, Dharmawardhana H, Gamage P ,Ubayasiri R, Dias KMGSM , Sandaruwan WAA , Alahakoon A.E

Introduction : Takayasu arteritis(TA) or "pulseless disease" is a rare large vessel vasculitis of unknown aetiology with an incidence of 1.1 per million person years. It is characterized by granulomatous inflammation of the aorta, its major branches and pulmonary arteries resulting in segmental stenosis, occlusion, dilatation and/or aneurysms.

Description : A 12-year-old previously healthy boy presented with convulsions following a brief febrile illness. He was noted to have severe persistent hypertension with low volume right radial pulses and marked discrepancy(>10mmHg) in blood pressures between two upper limbs. There was no history suggestive of limb claudication, abdominal symptoms or cutaneous manifestations of systemic vasculitis.

Investigation : The full blood count and urinalysis was normal with negative inflammatory markers and immunology. The echocardiogram revealed left ventricular hypertrophy. Kidneys were ultrasonically normal with normal doppler flows. CT angiogram detected narrowed segments of right subclavian artery and coeliac trunk. MR angiogram revealed segmental narrowing and wall thickening of abdominal aorta and stenosis at the onset of the coeliac trunk.

Progression : Although blood pressure control was initially difficult despite using 3 antihypertensives, it was achieved following immuno-suppression with steroids and cyclophosphamide.

Discussion : The diagnosis of TA needs high index of suspicion and is based on clinical criteria and angiographic abnormalities. The inflammatory markers may be misleading in acute disease and in long term monitoring of disease activity. Glucocorticoids remain the mainstay of therapy to induce remission with addition of cyclophosphamide in life threatening situations. Methotrexate, azathioprine, MMF are often required to sustain remission.

NONSPECIFIC CHRONIC ABDOMINAL PAIN IN A CHILD, COULD IT BE A TUMOR?- A CASE OF GANGLIONEUROBLASTOMA

R.Balasubramaniam, T.J.Hoole: I.Jayasundara, A.K.Nimanthi, D.S. Gamage, A.K.T.N.de Silva, I.Kumarasiri, N. Atapattu

Introduction: Neuroblastic tumors represent the predominant extra cranial solid tumors encountered in pediatric patients. Often, they manifest with nonspecific abdominal pain, a common complaint among children, typically attributed to functional causes. Among the spectrum of neuroblastic tumors, ganglioneuroblastoma is a relatively uncommon subtype.

Description: Here, we present the case of a 6-year-old girl, who presented with a 6-month history of nonspecific abdominal pain, recently exacerbated in frequency.

Investigations (if any): Evaluation revealed a right-sided adrenal mass during ultrasound examination, with no significant additional clinical features and a normal physical examination, consistent with a prepubertal stage. Contrast-enhanced computed tomography (CECT) of abdomen confirmed the presence of a contrast-enhancing lesion within the right adrenal gland. Laboratory analysis indicated normal levels of dehydroepiandrosterone-sulphate (DHEAS) and testosterone.
Progress: Subsequently, the patient underwent surgical resection, experiencing an uneventful recovery. Histological examination confirmed the diagnosis of ganglioneuroblastoma, with no evidence of lymph node involvement. Presently, the patient is being followed up with oncology input.

Discussion: Neuroblastic tumors could be a rare cause of chronic nonspecific abdominal pain in children. Timely evaluation leads to favorable prognostic outcomes.

ACUTE HAEMOLYTIC EPISODE IN A PREVIOUSLY UNDIAGNOSED PATIENT WITH G6PD DEFICIENCY ; A RARE PRESENTATION FOLLOWING INGESTION OF HEEN MADURUTHALA (OCIMUM TENUIFLORUM L.)

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Introduction: Glucose-6-phosphate dehydrogenase (G6PD) deficiency is an X-linked disorder affecting enzymes in Hexose monophosphate pathway. The clinical spectrum of G6PD deficiency includes episodic haemolytic anaemia and chronic non-spherocytic anaemia, often presenting as neonatal jaundice and acute haemolytic episodes. Diagnosis relies on clinical findings as well as demonstrating reduced G6PD activity in red blood cells. Blood transfusions for severe acute haemolytic episodes and prevention of predisposing factors play an important role in the management

Description: A 3-year-old child of healthy parents was transferred from a local hospital complaining yellowish discoloration of sclera, the passage of dark urine, fever and mild difficulty in breathing for one day. Further history revealed ingestion of the plant Ocimum tenuiflorum L. (Heen Maduruthala) a day prior to the onset of symptoms.

On examination, the child was febrile, severely pale, icteric, with a thready pulse, tachycardia and gallop rhythm. There was no hepatosplenomegaly respiratory examination was normal

Investigations (if any): FBC revealed normochromic normocytic, severe anaemia (5.1g/dl) with reduced haematocrit (16.3%), and leukocytosis. Direct agglutination test was negative, and further investigations revealed elevated LDH (1836 U/l), reticulocyte count (11.5%), CRP (62mg/l) serum ferritin (13,395 ng/ml) blood urea (8.15mmol/l) and AST (104 U/l).

The peripheral blood smear revealed normochromic, normocytic red cells with bite cells, blister cells, occasional spherocytes, polychromasia, and NRBCs. He was found to have low G6PD enzyme levels after six weeks of acute episode

Progress: In the acute setting, the patient was managed with close monitoring of vitals and blood transfusion. The patient began showing improvement both clinically and in his blood counts. On discharge, Parents were advised on avoiding triggering factors in the future

Discussion: Differentiating causes of acute haemolytic episodes is important in first presentation of haemolytic anaemias. Isolating a causative trigger remains a challenge in G6PD deficiency prevention of risk factors is important in the management

SEVERE INTERLECTUAL IMPAIRMENT IN A GIRL WITH TURNER VARIANT SYNDROME

W A I Nilmini Warnakula, Lokubalasuria T , Madushani L , De Silva D C , Fernando M S S, Fonseka O D S, Prabath A, Vipulaguna D

Introduction: Turner syndrome (TS) is a chromosomal disorder characterized by partial or complete loss of one of the X chromosomes.

Description: This case report details a 15-year-old female with Turner variant who exhibits severe intellectual impairment. It emphasizes the need for a multidisciplinary approach to address multiple impairments in TS children.
She had webbed neck, wide carinal angle, mal-occluded teeth on examination. Her height was with in 10th centile for her age, but within mid-parental height. Psychometric assessment indicated mental age of 9 years and 6 months and extremely low non-verbal IQ (<1%). She had impaired working memory, listening comprehension, reading fluency, reading accuracy and writing fluency. She was an alphabetical reader and had poor sentence organization. Her writing displayed syntactical and morphological errors. She had basic self-awareness and the adaptive skills were partial independent. She also had epilepsy and primary amenorrhoea.

Investigations (if any): Genetic testing showed the presence of Turner variant with 46, X,?dup{X} {q26-qter}, MRI brain and EEG revealed no abnormalities.

Progress: A multidisciplinary team, including paediatric specialities, genetics, psychology, and education, occupational therapy, developed a comprehensive management plan. This included tailored educational strategies, preparation for possible vocational training, ensure safeguarding, improving adaptive skills and behavioral therapy. She is on anti-epileptics and estrogen replacement therapy.

Discussion: The case underscores the significance of a functionality focused multi-disciplinary approach to Turner Variant Syndrome (TS). In comparison to typical TS, this child has poor reading skills and severe IDD. A multidisciplinary approach is crucial for comprehensive assessment, interventions, and support.

A CASE OF DIPHALLIA WITH ANORECTAL MALFORMATION

D. Sachini Kaushalya Weerakkody, Charith Prabhavitha, Aswin Isira Wijerathne, Tikiri Gunawardana

IIntroduction: Diphallia is an extremely rare urological anomaly. It can present as complete or incomplete diphallia, varying from a small accessory penis or duplication of the glans to complete penile duplication. In most cases it is associated with complex urological, gastrointestinal, or anorectal malformations.

Description: A baby boy was born to non-consanguineous parents at term. Mother is 20-year-old who was in her second pregnancy without any complications. Baby has passed urine and meconium within the first 24 hours of life. Both phalluses of his penis had normal-shaped glans with 2 urethral openings located at the normal position. However, he was passing urine from the right phallus only. He had a single scrotum and well-formed rouge with palpable testes. He had an anterior ectopic anus with normal tone without faecal incontinence, which was located at the base of the scrotum.

Investigations: Ultrasonography of the urological system showed normal kidney and bladder morphology. Both penises had normal internal morphology, but a normal urethra was seen within the large right penis.
The micturition cystourethrogram, 2D Echo and spinal x-rays were normal.

Discussion: Diphallia is a rare congenital anomaly comprising two structurally and anatomically separate phalluses. A widely accepted classification separates true diphallia from bifid diphallia. In true diphallia each phallus has two corpora cavernosa and a corpora spongiosum. Bifid phallus has only one corpora cavernosum in each penis. Diphallia is commonly associated with other congenital anomalies. Management of these patients is a challenge to the multidisciplinary team.

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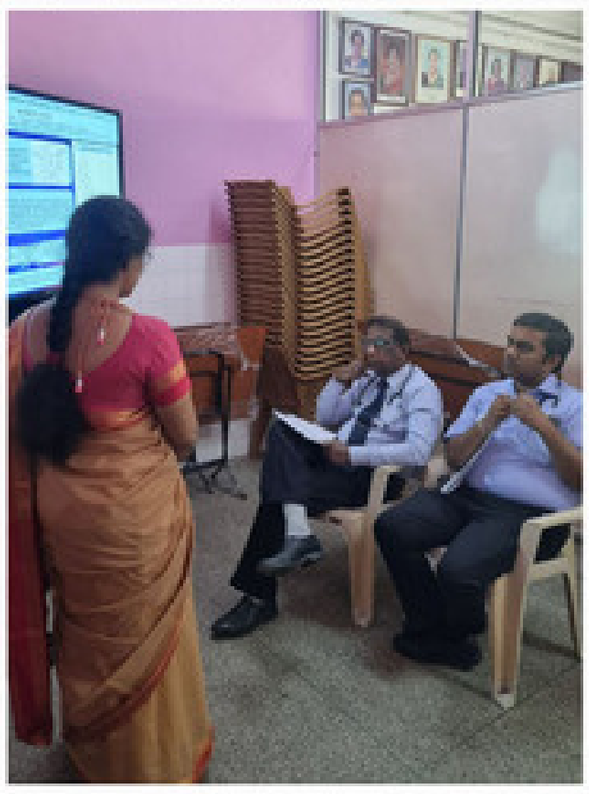
PHOTO GALLERY



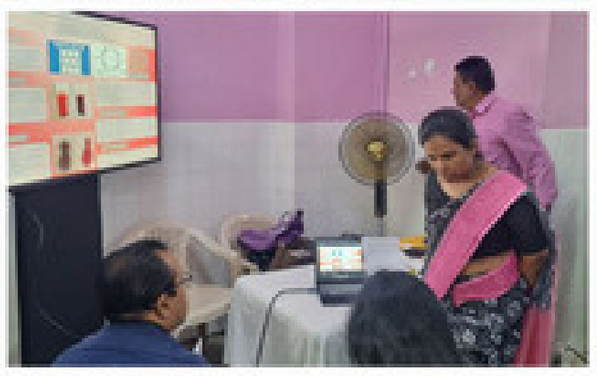




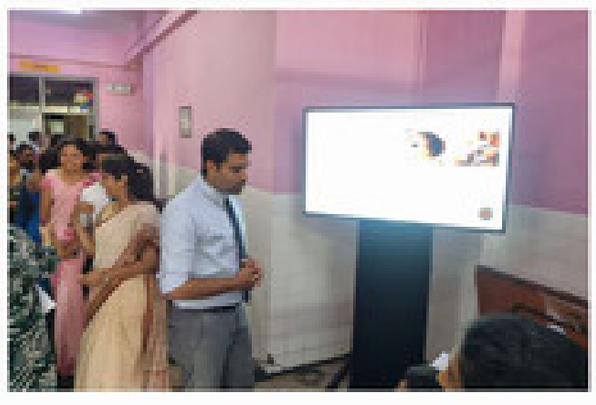






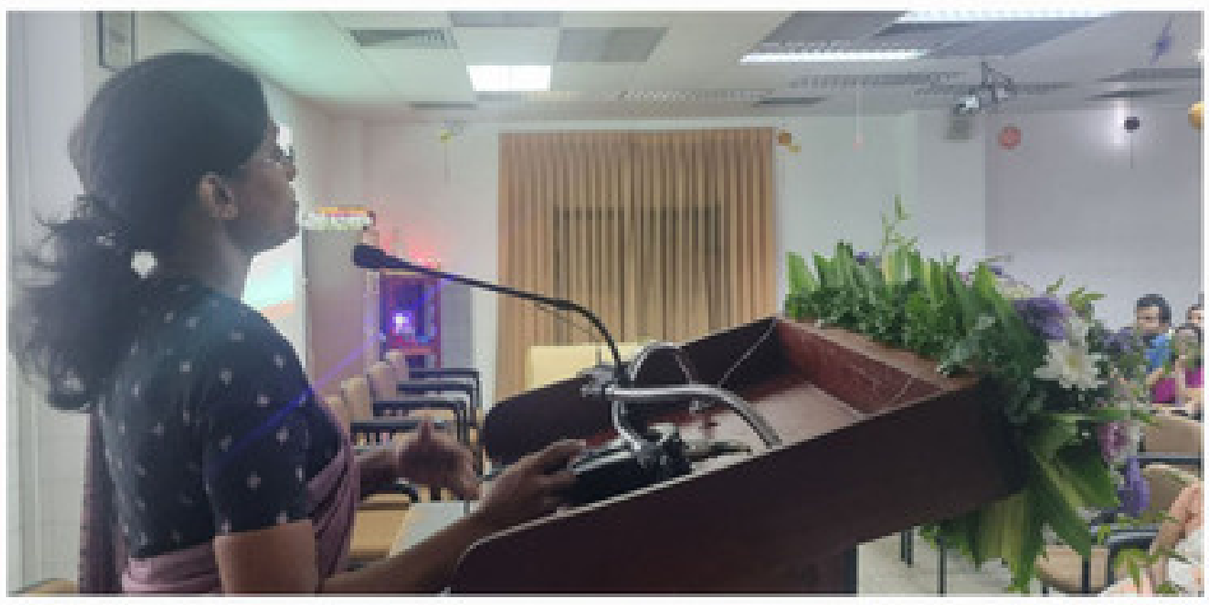




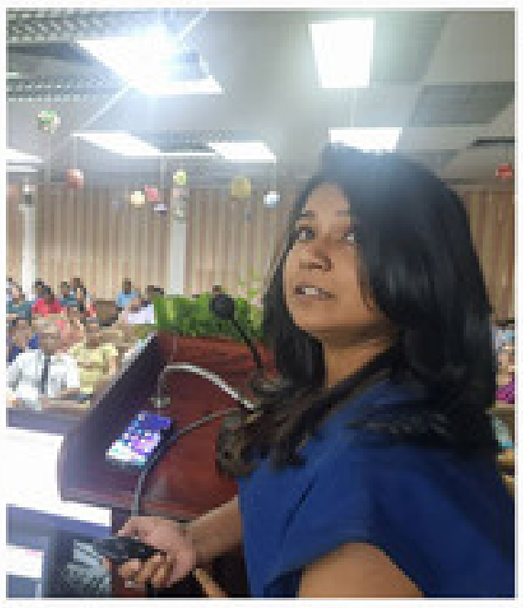




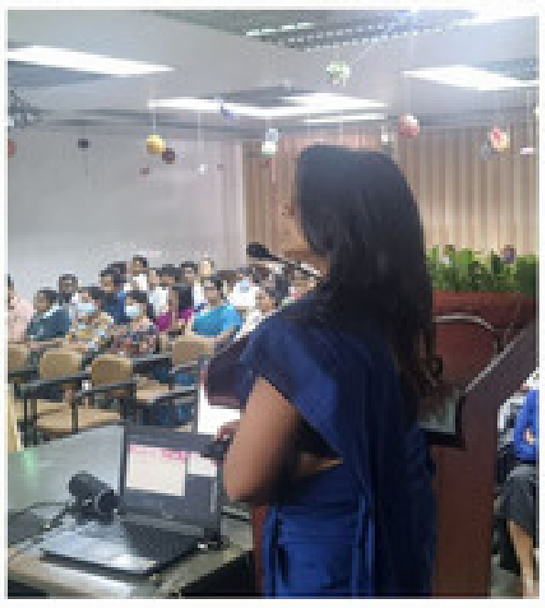


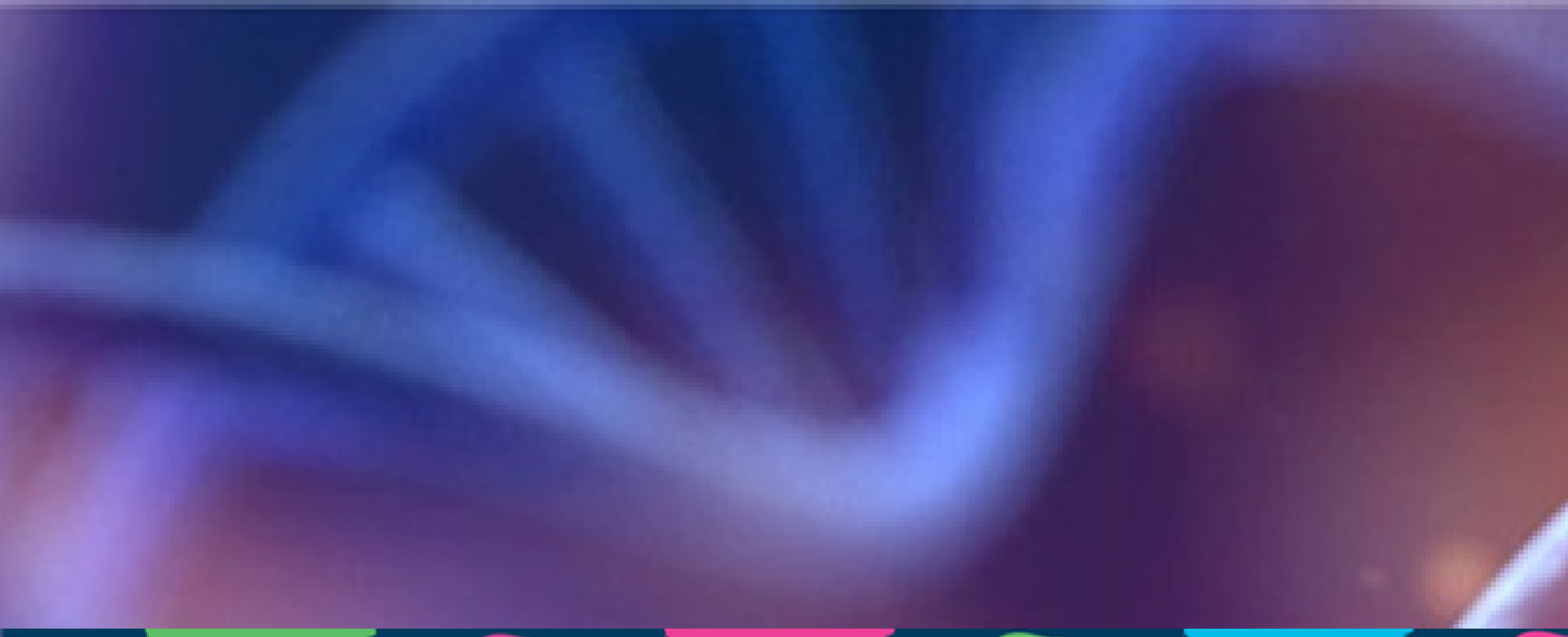






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