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Expert Committee on Birth Defects

Proceedings

of the 10th Rare Disease Day Symposium

6th March 2026

Lady Ridgeway Hospital for Children, Colombo

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Rare Disease Forum of the Sri Lanka College of Paediatricians

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2026 ACADEMIC PROGRAMME

10TH RARE DISEASE DAY SYMPOSIUM

Time	Topic	Speaker
08:20 am	Welcome Address: President, Rare Disease Forum	Professor Ruwanthi Perera
08:30 am	Keynote Address: Founder President, Rare Disease Forum	Professor Harendra De Silva
08:40 am	Address: President, Sri Lanka College of Paediatricians	Professor Pujitha Wickramasinghe
08:50 am	Address: Co-chair, Expert Committee on Birth Defects of the SLMA	Dr Kapila Jayarathne
09:00 am	Genetic Metabolic Liver Disorders: A Lifelong Journey Starting in Childhood	Professor Meranthi Fernando
09:25 am	Inherited Metabolic Disorders Presenting with Recurrent Rhabdomyolysis: A Diagnostic Approach	Dr Arthavan Selvanathan
09:50 am	Early Diagnosis and Timely Intervention: Navigating Congenital Cytopenias	Dr Shanika Vitharana
10:15 am	Birth Defects: The Extent of Developmental Genetics' Responsibility	Dr Vindya Subasinghe
10:40 am	Tea Break	
11:00 am	Free Papers	
12:00 pm	Recognising Dysmorphic Features: A Practical Guide for Paediatricians	Dr Elizabeth Wall
12: 30 pm	Role of Patient Support Groups in Rare Diseases	Ms Kirsty Hoyle
12:50 pm	Vote of Thanks: Secretary, Rare Disease Forum & SLCP	Dr Imalke Kankananarachchi
01:00 pm	AGM & Lunch	

Prof Meranthi Fernando
Consultant Paediatrician

Dr Arthavan Selvanathan
Consultant Metabolic Paedatrician

Dr Shanika Vitharana
Consultant Haematologist

Dr Vindya Subasinghe
Consultant Clinical Geneticist

Dr Elizabeth Wall
Consultant Clinical Geneticist

Ms Kirsty Hoyle
CEO, MSUK

PRESIDENT'S MESSAGE

Professor Ruwanthi Perera
Professor in Paediatrics
University of Sri Jayewardenepura

Rare diseases may be individually uncommon, but collectively they contribute significantly to morbidity and mortality. Many affected families face considerable challenges in obtaining timely diagnosis, appropriate treatment, and long-term care, particularly in settings with limited resources. Although Sri Lanka has made some progress in early detection and management in recent years, much more remains to be done. Early diagnosis is crucial to improving outcomes during the critical “golden period” of growth and development, and it also enables appropriate genetic counselling for parents regarding future pregnancies.

The Rare Disease Forum of the Sri Lanka College of Paediatricians was initiated in 2015 with the aim of improving the quality of life of children with rare disorders. This year marks the 10th anniversary of the Forum, and we are proud of the significant milestones achieved during this journey.

Globally, advances in genomics, targeted therapies, and strong patient advocacy have led to improved outcomes for many rare conditions. However, our country continues to face challenges in awareness, access to specialised care, and sustained policy-level support. Strengthening initiatives such as newborn screening programmes, research collaborations, and national patient registries is essential to bridging these gaps.

In 2026, to further strengthen local support systems, the Rare Disease Forum collaborated with Metabolic Support UK to launch the Metabolic Support Sri Lanka initiative, aimed at supporting children with inherited metabolic disorders. In addition, with the support of the International Gaucher Alliance, the Forum successfully secured access to costly enzyme replacement therapy for two children with lysosomal storage disorders.

Every step forward brings renewed hope to affected children and their families. Let us continue to work together—health professionals, policymakers, researchers, and advocates—to ensure that no individual faces a rare disease alone.

SECRETARY'S REPORT

Dr Imalke Kankananarachchi
Senior Lecturer in Paediatrics
University of Ruhuna

- **9th Rare Disease Day-2025**

The Rare Disease Forum of the Sri Lanka College of Paediatricians commemorated the 9th Rare Disease Day on 7th March 2025 at the Lady Ridgeway Hospital. The programme featured 30 abstract presentations and six plenary lectures, highlighting current developments in the field of rare and metabolic disorders.

The event was enriched by the participation of international experts, including Dr. Anil Jalan and Dr. Ketki Kudalkar from Mumbai, India, who attended in person, and Dr. Chris Hendriksz, who joined virtually. The local faculty comprised Prof. Jayamini Senevirathna, Dr. Rajiva De Silva, and Dr. Vindya Subasinghe. The meeting was well attended, with over 120 participants.

- **New Office bearers of the Rare Disease Forum 2025-2026**

Prof. Harendra de Silva continued to serve as the Patron of the Rare Disease Forum of the Sri Lanka College of Paediatricians. Prof. Ruwanthi Perera and Dr. Imalke Kankananarachchi were reappointed as President and Secretary of the Forum, respectively. Dr. Vindya Subasinghe and Dr. Dananjaya Vithanage were appointed as Treasurer and Assistant Secretary, while Dr. Navoda Atapattu assumed the role of Vice President. Prof. Ranula Ranawaka was appointed as the Editor of the Forum.

- **Symposium of Mitochondrial disorders – 13th November 2025**

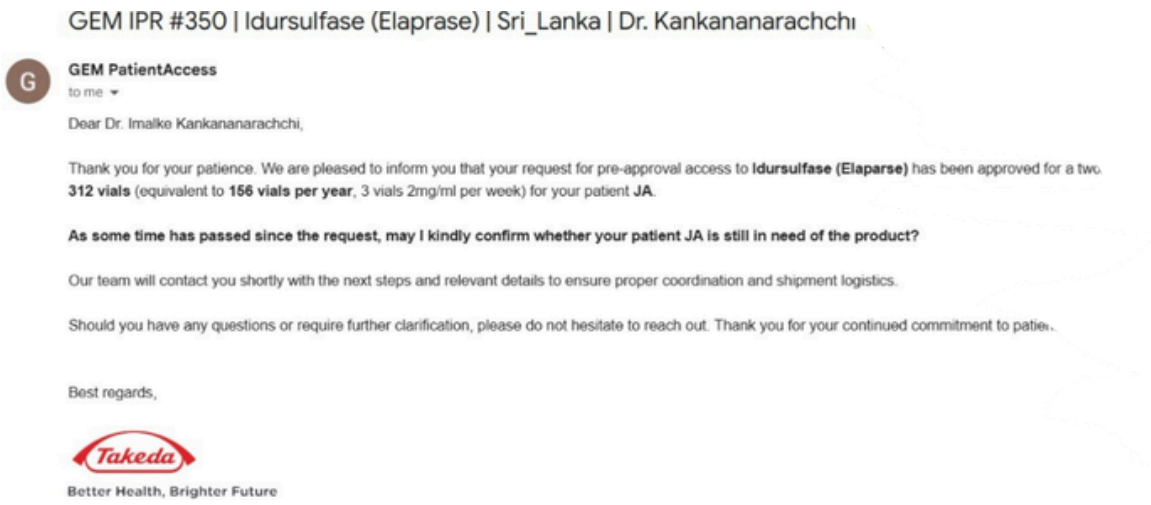
The Rare Disease Forum of the Sri Lanka College of Paediatricians organized a Symposium on Mitochondrial Disorders at the Main Auditorium of the Lady Ridgeway Hospital on 13th November 2025. The guest faculty included Dr. Anil Jalan from NIRMAN, as well as Dr. Pyara Rathnayake and Dr. Vindya Subasinghe from Lady Ridgeway Hospital. The symposium was well attended, with over 150 participants joining both in person and virtually.

The event was enriched by the participation of international experts, including Dr. Anil Jalan and Dr. Ketki Kudalkar from Mumbai, India, who attended in person, and Dr. Chris Hendriksz, who joined virtually. The local faculty comprised Prof. Jayamini Senevirathna, Dr. Rajiva De Silva, and Dr. Vindya Subasinghe. The meeting was well attended, with over 120 participants.

- **Charitable access to Enzyme Replacement therapy of children with Lysosomal Storage Disorders**

The Rare Disease Forum of the Sri Lanka College of Paediatricians, in collaboration with the International Gaucher Alliance, facilitated access to enzyme replacement therapy (ERT) for a child with Gaucher disease as pre-treatment prior to planned haematopoietic stem cell transplantation (HSCT).

The medications were formally handed over to Dr. Shanika Vitharana by the local agent of Sanofi on 1st August 2025. The child received ERT for a period of two months; however, at the conclusion of therapy, the family declined consent to proceed with HSCT.
Another child with Mucopolysaccharidosis type II was approved to receive Intravenous Idursulfase a period of 2 years by the Takeda pharmaceutical company.



GEM IPR #350 | Idursulfase (Elaprase) | Sri_Lanka | Dr. Kankananarachchi

G **GEM PatientAccess**
to me

Dear Dr. Imalke Kankananarachchi,

Thank you for your patience. We are pleased to inform you that your request for pre-approval access to **Idursulfase (Elaparse)** has been approved for a two **312 vials** (equivalent to **156 vials per year**, 3 vials 2mg/ml per week) for your patient **JA**.

As some time has passed since the request, may I kindly confirm whether your patient JA is still in need of the product?

Our team will contact you shortly with the next steps and relevant details to ensure proper coordination and shipment logistics.

Should you have any questions or require further clarification, please do not hesitate to reach out. Thank you for your continued commitment to patie.

Best regards,

Takeda

Better Health, Brighter Future

- **Diagnostic Pathways and Sustainable Solutions for Gaucher Disease & Inherited Metabolic Disorders in Sri Lanka- 15th January 2026**

A meeting was held on 15/01/2026 involving the following individuals to

- Dr. Nishant Kumar Singh – Medical Lead, India & Southeast Asia, Sanofi
- Prof Chris Hendriksz– Former NHS clinician; Lead, African Diagnostic Network
- Dr. Imalke Kankananarachchi – Secretary, Rare Disease Forum, Sri Lanka
- Dr. Vindya Subasinghe – Treasurer, Rare Disease Forum, Sri Lanka
- Vesna Aleksovska – IGA representative and patient advocate
- Dr Alaa Hamed - Global Head of Medical Affairs Rare Diseases, Sanofi

The meeting was convened to address the disruption of diagnostic testing for Gaucher disease and related inherited metabolic disorders in Sri Lanka following the discontinuation of Sanofi-supported dried blood spot (DBS) testing and the emergence of new regulatory barriers. Historically, Sanofi-supported DBS testing enabled the diagnosis of most Gaucher patients; however, recent regulatory changes requiring DBS kits to be registered as medical devices, combined with customs clearance issues, have halted imports. As a result, patients are forced to seek costly private testing abroad, leading to delayed diagnoses and compromised access to timely treatment, particularly for neuronopathic cases and family screening.

During the discussion, Sanofi clarified the regulatory context and ongoing efforts toward kit registration, while models from Africa were presented as potential frameworks for sustainable, research-based diagnostic collaboration. Participants agreed on a phased approach: in the short term, to continue DBS testing using locally available filter paper and explore temporary financial support; in the medium term, to pursue formal DBS kit registration and re-establish compliant diagnostic pathways; and in the long term, to develop research collaborations to enable funded diagnostics, assay validation, and local capacity building, including exploration of genetic-first strategies. Immediate next steps include stakeholder coordination, documentation sharing, exploration of funding mechanisms, and follow-up meetings focused on regulatory and research pathways toward a sustainable solution.

- **Metabolic Support Sri Lanka- 4th March 2026**

The inauguration of Metabolic Support Sri Lanka was held at the Lady Ridgeway Hospital Main auditorium on 4th March 2026 at 9 am with the participation of children with Inherited Metabolic Disorers and their families. Dr Nalinda Jayatissa, Hon Minister of Health and Mass Media, Dr. Rajesh Sambhajirao Pandav, WHO representative of Sri Lanka, Ms Kirsty Hoyle- CEO, Metabolic Support UK, Mr Jonathan Gibson, Campaigns and Communications Lead- MSUK and Ms Sarah Jones Community Lead at Metabolic Support UK joined the event.

Professor Ruwanthi Perera, president of the rare disease forum, welcome the gathering and Prof Pujitha Wickramsinghe, president of SLCP addressed the gathering.Dr Imalke Kankananarachchi, secretary of SLCP made a presentation on the current situation of Inherited Metabolic Disorders in Children and Dr Dedunu Dias, Director Medicine Suppliers division talked about the access to drugs used in rare disorsers. Prof Pujitha Wickramasinghe handed over the list of medications that are not available in Sri Lanka to the Ministry of health. In parallel to this, there were several media briefngs in sri lanks attaned by local and overses delegates

SPEAKER ABSTRACTS

Genetic metabolic liver disorders; a lifelong journey starting in childhood

Professor Meranthi Fernando
Professor in Paediatrics & Consultant Paediatrician
Special Interest in Paedatric Hepatology & Liver Transplantation
Colombo North Centre For Liver Diseases,
University of Kelaniya, Sri Lanka

Liver diseases in childhood differ fundamentally from those in adults with regard to etiology, clinical presentation, management, and long-term outcomes. A defining characteristic of paediatric liver disease is its strong genetic basis. With the notable exception of biliary atresia, most chronic liver disorders presenting in early life are inherited or genetically determined, thereby placing them primarily within the domain of paediatric practice.

Genetic–metabolic liver diseases comprise a heterogeneous group of disorders affecting carbohydrate, protein, lipid, bile acid, or copper metabolism and transport. These include galactosaemia, glycogen storage diseases, tyrosinaemia, urea cycle disorders, fatty acid oxidation defects, progressive familial intrahepatic cholestasis, and Wilson disease, among others. Although these conditions initiate a lifelong disease trajectory beginning in infancy or childhood, many affected children remain undiagnosed or are misdiagnosed, particularly in low- and middle-income countries.

Even when diagnosis is achieved, timely and appropriate management may be limited by inadequate awareness, scarcity of trained specialists, and restricted access to advanced diagnostic modalities, especially genetic testing. Importantly, some disorders require relatively simple yet life-saving interventions, such as early dietary modification in galactosaemia or copper chelation therapy in Wilson disease. Others necessitate disease-specific pharmacotherapy or liver transplantation to prevent mortality or improve long-term quality of life.

Most genetic–metabolic liver diseases follow an autosomal recessive inheritance pattern. In the Sri Lankan and broader South Asian context, consanguinity contributes to familial clustering, with multiple affected siblings observed in some families. This underscores the importance of family screening, genetic counselling, and informed reproductive planning following identification of an index case.

Clinically, these disorders may present with prolonged neonatal jaundice, neonatal or later-onset acute liver failure, hepatomegaly, recurrent hypoglycaemia, persistently elevated transaminases, chronic liver disease, unexplained hepatosplenomegaly, or metabolic crises.

Improving outcomes requires increased awareness among frontline clinicians, strengthened early detection strategies, establishment of centralized referral pathways, and expanded access to diagnostic and therapeutic services. Strategic allocation of healthcare resources is essential, as many of these conditions have favorable outcomes with timely intervention, making early diagnosis and appropriate management both life-saving and cost-effective.

Inherited Metabolic Disorders Presenting with Recurrent Rhabdomyolysis: A Diagnostic Approach

Arthavan Selvanathan[1]
[1]Paediatric Metabolic Physician, Sydney Children's Hospitals Network, Australia
[2]Discipline of Child and Adolescent Health, Faculty of Medicine and Health, University of Sydney Australia

Rhabdomyolysis is a rare but potentially life-threatening condition, which occurs due to breakdown of skeletal muscle. Widespread release of the contents of muscle cells results in myoglobinuria, hyperkalaemia and increased creatine kinase, among other biochemical abnormalities. In some instances the myoglobinuria can accumulate in the renal tract and cause acute kidney injury. Most cases have complete recovery with supportive management: however those with severe or recurrent rhabdomyolysis can be at risk of complications.

Most children who have rhabdomyolysis have a viral trigger (viral myositis) or have it as a result of physical trauma: in these groups the risk of recurrence in this group is low. However, a small proportion of patients (3-5%) have rhabdomyolysis secondary to inborn errors of metabolism. This group is particularly crucial to be aware of for two reasons: the risk of recurrence in such patients is high, and also there often may be a need for disease-specific management in order to optimize outcomes.

This presentation will aim to assist the general paediatrician in understanding more about the metabolic causes of rhabdomyolysis. An initial case vignette will be followed by information on diagnostic clues for inborn errors of metabolism underpinning rhabdomyolysis, as well as relevant investigations and treatment strategies in both the short- and long-term.

Clinically, these disorders may present with prolonged neonatal jaundice, neonatal or later-onset acute liver failure, hepatomegaly, recurrent hypoglycaemia, persistently elevated transaminases, chronic liver disease, unexplained hepatosplenomegaly, or metabolic crises.
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Early Diagnosis and Timely Intervention: Navigating Congenital Cytopenias

Dr Shanika Vitharana
Consultant Haematologist, Bone marrow transplant unit,
Lady Ridgeway Hospital for Children

Congenital cytopenias represent a heterogenous group of rare inhetited bone marrow failure syndromes (IBMFS) characterized by persistent single or multilineage cytopenias,immune dysregulation and variable risk of malignant transformation.
Vigillant clinical suspicion, advanced genomic testing and functional assays are required for an accurate diagnosis of these rare conditions. However, acess to these testing remains a challenge in resource limited setting. This will encompass missed or delay in diagnoses leading to poor outcomes in this group. However, a correct genetic diagnosis is crucial as the management stratergies, transplant approach and disease survallence differ subtantially.

This presentation highlights the approach to congenital cytopenias , and the importance of early recognision in the era of advanced therapies such as, Haematopoietic stem cell transplantation(HSCT) and gene tharapy.
Improving awareness among clinicians, strengthening diagnostic pathways, and ensuring timely access to definitive therapy and multidisciplinary care are essential to improving survival and long-term outcomes in children with congenital cytopenias.

Birth Defects: the extent of developmental genetics 'responsibility

Dr Vindya Subasinghe
Consultant Clinical Geneticist
Lady Ridgeway Hospital for Children
Member, Expert Committee on Birth Defects, SLMA

Birth defects, also referred to as congenital disorders, are structural or functional abnormalities present at birth, including metabolic disorders. They represent a significant global health concern, contributing substantially to neonatal and infant morbidity and mortality. According to global estimates, congenital anomalies account for approximately 7% of neonatal deaths worldwide, with an even greater proportional impact in regions where overall child mortality rates are low. In Sri Lanka, birth defects remain a major contributor to infant mortality despite improvements in overall infant survival rates.

Beyond mortality, birth defects impose long-term consequences including chronic morbidity, intellectual disability, psychosocial burden on families, and increased healthcare costs, ultimately affecting national productivity. Clinical genetics plays a critical role in understanding the developmental origins of these conditions through accurate diagnosis, identification of causative factors, prognostic counseling, and formulation of long-term management plans. Genetic evaluation also enables families to understand disease causation and recurrence risks, supporting informed reproductive decisions.

The etiology of congenital malformations is multifactorial, involving genetic mechanisms, developmental disturbances, and environmental influences such as teratogens. Comprehensive assessment of affected infants is therefore essential to identify underlying mechanisms and associated anomalies. Strengthening diagnostic genetics, preventive strategies, and multidisciplinary care is vital to reduce the burden of birth defects and improve outcomes for affected children and their families.

Recognising Dysmorphic Features: A practical guide for paediatricians

Dr Elizabeth Wall
Consultant Clinical Geneticist, UK

The technical definition of ‘dysmorphology’ is ‘the recognition and study of birth defects and syndromes’. The term was first used in the 1960s. In reality it is an art; the art of diagnosis. It requires a combination of methodical analysis, pattern recognition, collaboration and experience.

In this talk I will discuss an approach to dysmorphology, including:

- How to recognise and describe dysmorphic features
- To identify a likely developmental cause of the dysmorphic feature (e.g. malformation, deformation, disruption, dysplasia)
- Some useful patterns to recognise
- What to do when you get stuck

Useful free resources:
Describing a dysmorphic feature: Elements of morphology
https://elementsofmorphology.nih.gov/index.cgi

HPO terms: https://hpo.jax.org/

Generating a differential diagnosis: https://pubcasefinder.dbcls.jp/

Role of Patient Support Groups in Rare Diseases

Ms Kirsty Hoyle
CEO, Metabolic Support, UK

Patient groups play a vital role in building equitable, effective and sustainable healthcare systems. The voices, stories and everyday experiences of people living with a rare disease are a vital part of the healthcare ecosystem but too often they are unheard or unsought. Patient groups have a unique opportunity to find, share and amplify these voices so that no one is alone.

We are far more than support networks; we are organised communities of lived experience, insight and expertise. Patient groups provide trusted information, peer connection and advocacy — but they also transform individual stories into collective evidence that can influence research, policy and service design. Healthcare systems often focus primarily on clinical endpoints and measurable outcomes. Patient groups broaden that perspective. They articulate what truly matters to communities: quality of life, dignity, timely diagnosis, access to care, continuity of support and the realities of navigating complex, and often inequitable, systems. By gathering lived experience at scale, patient organisations provide context that data alone cannot capture, strengthening the relevance and impact of research and decision-making.

Meaningful patient engagement depends on strong, well-resourced patient groups that are involved early and treated as equal partners. Their insight helps shape research priorities, inform drug development, and improve service planning. Moving beyond tokenism requires recognising patient organisations as strategic partners with expertise and accountability. When their voices are embedded from the outset, healthcare becomes more responsive, more inclusive and better aligned with the needs of the communities it serves.

ABSTRACTS OF ORAL & POSTER PRESENTATIONS

EXPLORING THE CLINICAL SPECTRUM OF PAEDIATRIC SIALURIA: A TWO-CASE SERIES WITH GENETIC CORRELATION

Dinithi Sewmini Gamage Naotunna, Vindya Subasinghe, Pyara Ratnayake

Introduction:
Sialuria is an ultra-rare inborn error of sialic acid metabolism with less than 20 reported cases, caused by a gain-of-function mutation in the GNE gene. This leads to overproduction of sialic acid and an increase in urinary free sialic acids. The clinical manifestations are highly variable.
We present two patients with a heterozygous missense mutation in the GNE gene who had global developmental delay and MRI scans showing cerebral atrophy, along with a negative urine organic acid profile.

Description:
Case 01- This was an eight-year-old boy with microcephaly, tall stature, facial dysmorphism (low-set ears, thin upperlip), and pectus excavatum. His tone was normal.
Case 02- This was a four-year-old girl, the first child of non-consanguineous parents She had hypotonia with some features of autism spectrum disorder. She was not dysmorphic and had normal head circumference.
Supportive management included nutritional optimisation and regular developmental monitoring. Both patients demonstrated stable growth and developmental progress, with no significant deterioration.

Discussion:
These cases illustrate the phenotypic variability that may be seen in paediatric sialuria and highlight the importance of considering sialuria in children with unexplained developmental delay. Early genetic confirmation aids in avoidance of unnecessary investigations and genetic counselling.

CLINICAL PROFILE AND TREATMENT MODALITIES OF EPILEPSY COMPLICATED WITH CONTINUOUS SPIKE WAVE IN SLEEP (CSWS) IN CHILDREN IN A TERTIARY CARE PAEDIATRIC HOSPITAL IN SRILANKA

E.P.L.J.G. Lappen, W.M.W. Sandipanai, Pyara Ratnayake

Introduction:
Developmental epileptic encephalopathy with spike- wave activation in sleep (DEE - SWAS) is a rare condition making up 0.5-0.6% of all epilepsy. As it is associated with cognitive deterioration, early identification and treatment is important.

Description:
Study explores clinical presentations, EEG localisation, aetiology, treatment modalities and seizure outcome of DEE-SWAS

Progress:
24 children, with ages ranging from of 1 month -12 years were studied with mean age of 8.2 ± 3.8 years (2.25 -14.75). Majority were males (62.5%). Spike localisation was centro temporal (30%), occipital (30%), fronto temporal (12.5%) and parietal (12.5%). First diagnosis of epilepsy was at 4.4 years ± 3.9 (1 month – 11 years). First diagnosis of DEE-SWAS was at 6.7 ± 3.9 years (1 month -11 years). Mean duration from epilepsy diagnosis to DEE- SWAS was 2.57± 2.1 years (0-6). 16.7% had uncontrolled epilepsy with multiple daily seizures. However 50% have well controlled epilepsy with 30% of them being on monotherapy. First presentation of DEE-SWAS was seizures (75%), behavioural problems (25%), learning difficulty (12.5%) and speech problems (8.4%). Steroid was the most commonly used treatment modality (75%). Majority were on clobazam (56.5%), sodium valproate (52.2%), levetiracetam (39.1%) and ethosuximide (17.1%). 31% had structural pathologies among them 16.7% had perinatal vascular insult.

Discussion:
Presentation and aetiology of DEE-SWAS has a wide spectrum. Seizures were well controlled in majority. Steroid is the most commonly used treatment modality.

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Discussion:
Presentation and aetiology of DEE-SWAS has a wide spectrum. Seizures were well controlled in majority. Steroid is the most commonly used treatment modality.

SUCCESSFUL ALLOGENEIC HEMATOPOIETIC STEM CELL TRANSPLANTATION IN A CHILD WITH MIRAGE SYNDROME HARBOURING A SAMD9 VARIANT OF UNCERTAIN SIGNIFICANCE (C.3892G>A; P.GLY1298ARG)

Gunawardana B M, Dissanayake R, Vidanagama D, Senadheera N, Vitharana S

Introduction: MIRAGE syndrome is a rare multisystem disorder caused by heterozygous gain-of-function mutations in the SAMD9 gene, characterised by myelodysplasia, recurrent infections, growth restriction, adrenal hypoplasia, genital anomalies and enteropathy. Progressive bone marrow failure is a major cause of morbidity and mortality. Haematopoietic stem cell transplantation (HSCT) remains the only curative therapy for haematological manifestations, although outcomes are frequently complicated by infections, graft-versus-host disease (GvHD) and organ dysfunction

Description: We report a female child with intrauterine growth restriction, recurrent severe infections, adrenal insufficiency and gastrointestinal dysfunction, consistent with a clinical diagnosis of MIRAGE syndrome. She developed progressive pancytopenia secondary to bone marrow failure with myelodysplasia.

Investigations (if any): Bone marrow examination demonstrated hypocellularity with myelodysplastic features and cytogenetic evidence of monosomy 7. Targeted genetic testing identified a heterozygous SAMD9 variant of uncertain significance (c.3892G>A; p.Gly1298Arg), supporting SAMD9-associated marrow failure

Progress: Given progressive marrow failure and high-risk cytogenetics, she underwent matched sibling donor allogeneic HSCT using myeloablative conditioning. Neutrophil and platelet engraftment occurred promptly with achievement of full donor chimerism. Post-transplant complications included cytomegalovirus reactivation, lower gastrointestinal acute GvHD and gastrointestinal dysmotility. Despite these challenges, she remains clinically stable at one year post-transplant with 100% donor chimerism and flow-cytometric evidence of immune reconstitution

Discussion: This case demonstrates that successful HSCT is achievable in MIRAGE syndrome despite significant transplant-related complications. Early referral for transplantation should be considered in SAMD9-related marrow failure, particularly in the presence of monosomy 7. Meticulous pre-transplant optimisation, vigilant infection surveillance and early escalation of GvHD therapy are essential to improving outcomes in this high-risk population

CASE REPORT: EARLY DIAGNOSIS OF MAPLE SYRUP URINE DISEASE IN A NEONATE WITH HYPERAMMONEMIA AND SEIZURES

W.A.C.M.S.Wijerathne, A.J.R Osman Smith , N.Manike , M.Atapattu , A.Samarathunga

Introduction:
Maple syrup urine disease (MSUD) is a rare autosomal recessive disorder affecting branched-chain amino acid metabolism. Early diagnosis is crucial as delayed treatment can lead to irreversible neurological damage and death. This case emphasizes the importance of metabolic screening in neonates with unexplained neurological symptoms.

Description:
A 7 day old term male neonate, born via normal vaginal delivery to non-consanguineous parents, presented with increased tonicity of all four limbs and two day history of poor feeding. Physical examination revealed hypertonia without other remarkable findings. Notably, the characteristic maple syrup urine odour was absent.

Investigations :
Laboratory investigations showed elevated serum ammonia at 152 µmol/L. Arterial blood gas analysis, blood glucose, and electrolytes were normal with no metabolic acidosis. Plasma amino acid analysis revealed markedly elevated leucine + isoleucine (1304.177 µmol/L), leucine/phenylalanine ratio (26.63), leucine/alanine ratio (16.69), and valine (496 µmol/L), confirming MSUD diagnosis.

Progress:
The infant was immediately started on branched chain amino acid restricted formula, supportive care, and anticonvulsants for seizure control. He showed gradual clinical improvement with normalization of amino acid levels over subsequent weeks.

Discussion:
This case demonstrates that MSUD should be considered in neonates presenting with unexplained hypertonia, seizures, and hyperammonaemia, even without the pathognomonic urine odour, which may be absent in early presentations. The absence of metabolic acidosis does not exclude the diagnosis. Early recognition through comprehensive amino acid profiling and prompt initiation of dietary management are essential for preventing irreversible neurological sequelae and improving long-term outcomes in this potentially fatal metabolic disorder.

MOSAIC TURNER SYNDROME WITH TRIPLE X/MONOSOMY X PATTERN IN AN ADOLESCENT : CASE REPORT

W.A.C.M.S. Wijerathne, A.J.R.O Smith , Neel Kannangara

Introduction:
Turner syndrome is a genetic disorder affecting females, characterized by complete or partial absence of one X chromosome. Occurring in approximately 1 in 2,500 female births, it presents with short stature, delayed puberty, and various manifestations. Mosaic patterns account for 15-20% of cases.

Description:
A 13-year-old girl presented with short stature and delayed menarche. Height measured 135 cm (below 3rd percentile), weight 28 kg, with absent secondary sexual characteristics at Tanner stage 1. Physical examination revealed webbed neck, low posterior hairline, shield chest, and cubitus valgus. Cardiac examination and lymphedema assessment were normal.

Investigations:
Laboratory investigations showed elevated follicle-stimulating hormone and luteinizing hormone levels, indicating primary ovarian dysgenesis. Cytogenetic analysis revealed mosaic karyotype 47,XXX(22)/45,X(11), demonstrating both triple X and monosomy X cell lines. Echocardiography and renal ultrasound showed normal findings.

Progress:
Growth hormone therapy and oestrogen replacement therapy were planned to address short stature and induce pubertal development.

Discussion:
This case demonstrates Turner syndrome with rare mosaic karyotype 47,XXX/45,X . The patient exhibited predominantly Turner syndrome phenotype with characteristic physical features and ovarian dysgenesis. The manifestations of triple X syndrome might not manifest early years of life, where it is known to cause premature ovarian failure. The case emphasizes the importance of comprehensive clinical evaluation and cytogenetic analysis in growth disorders, as phenotypic expression may not correlate with karyotypic complexity. Early diagnosis through systematic evaluation enables timely hormonal interventions to optimize growth potential and support appropriate pubertal development in affected individuals. Furthermore, definitive diagnosis will be helpful in the counselling and future surveillance for anticipated implications of a particular condition.

CLINICALLY DIAGNOSED WISKOTT–ALDRICH SYNDROME IN TWO BROTHERS: A CASE REPORT FROM A RESOURCE-LIMITED SETTING

B.R.R.N.Gunaratne, W.D.M.S.Dayananda Bandara, W.D.T.H.K.Gunatilake, S.M.Jeewandara, W.A.D.D.Wickramaarachchi

Introduction:Wiskott–Aldrich syndrome (WAS) is a rare, X-linked primary immunodeficiency characterised by the triad of eczema, thrombocytopenia and recurrent infections. Early diagnosis is essential to reduce morbidity and guide surveillance for complications. In resource-limited settings, however, diagnosis often relies on clinical recognition.

Description: A 5-year-old boy presented with severe, extensive infected eczema complicated by sepsis needing ICU care for one month at TH Kurunegala. He had a background of recurrent skin infections and poor response to conventional topical treatment. Clinical examination revealed widespread eczematous lesions with secondary infection and features of systemic illness. Persistent thrombocytopenia was identified, raising suspicion of an underlying immunodeficiency. Based on the classical clinical triad, a diagnosis of Wiskott–Aldrich syndrome was made.

Investigations (if any):Full blood count demonstrated persistent thrombocytopenia. Other routine investigations were performed to exclude secondary causes of thrombocytopenia and sepsis. Genetic testing for WAS could not be performed due to unavailability. Family screening identified thrombocytopenia in a younger male sibling.

Progress:The sibling was initially asymptomatic but subsequently developed eczema during follow-up, supporting the diagnosis of WAS in both brothers. Both children were managed with aggressive treatment of infections, meticulous skin care and regular follow-up. They continue to experience intermittent episodes of infected eczema.

Discussion:This case highlights clinical diagnosis of Wiskott–Aldrich syndrome without genetics. Recognition of the classical features and early family screening are vital for appropriate management, counselling and long-term surveillance in resource-constrained settings.

FATAL RECURRENT CHYLOTHORAX IN A PREVIOUSLY WELL CHILD: TRAUMA UNMASKING AN UNDERLYING LYMPHANGIOMA

B.R.R.N.Gunaratne, P.Vasikaran, K.D.J.M.K.Galahitiyawa, K.K.Karunaratne, C.De Silva, S.Ilangamage, N.Kitulwatta

Introduction:Chylothorax is an uncommon but potentially life-threatening cause of pleural effusion in children. While most cases are secondary to trauma or surgery, underlying lymphatic malformations such as lymphangiomas can predispose to severe, recurrent disease. We report a previously healthy child in whom minor blunt trauma unmasked extensive lymphatic abnormalities, culminating in fatal recurrent chylothorax.

Description:A previously well 5½-year-old child sustained blunt trauma to the chest and shoulder. Initial evaluation revealed a displaced right clavicular fracture with bilateral pleural effusions. The child was referred to a tertiary centre for progressive respiratory compromise.

Investigations (if any):Imaging confirmed bilateral pleural effusions. Contrast-enhanced CT showed chylous ascites, loculated pleural effusions (right > left), and compressive atelectasis of the right lower lobe. Lymphoscintigraphy revealed multiple intra-abdominal tracer accumulations consistent with lymphangioma and diffuse mediastinal tracer uptake, indicating extensive lymphatic leakage.

Progress:Initial management included bilateral pleurodesis and intravenous octreotide, allowing discharge after clinical improvement. One month later, the child was readmitted with worsening respiratory distress. Despite repeat pleurodesis, intercostal drainage, continuous octreotide infusion, and intensive care support, the child’s condition deteriorated, culminating in fatal respiratory failure.

Discussion:This case highlights the catastrophic potential of extensive lymphatic malformations presenting as treatment-resistant chylothorax. Minor trauma may unmask underlying lymphangiomas. Management is challenging when both conservative and surgical measures fail, underscoring the importance of early recognition and multidisciplinary planning.

"WHEN EXCESSIVE NEONATAL WEIGHT LOSS UNMASKS A RARE RENAL DISEASE: NEONATAL POLYCYSTIC KIDNEY DISEASE"

Piumi Madawala, Prrana Sivakumar, Chamodi Nissanka, Yashoda Athauda, Sangeetha Wickramarathne, Sachith Mettananda

Introduction:
Neonatal polycystic kidney disease (PCKD) is an important cause of early chronic kidney disease and carries significant morbidity and mortality, reported up to 30%. Severe renal dysfunction in the neonatal period often indicates intrauterine onset. We report a rare case presenting with renal failure and failure to thrive.

Description: A term male infant born to non-consanguineous parents had normal antenatal scans except for third-trimester foetal growth restriction. Postnatally, subtle dysmorphic features (low-set ears, micrognathia) were noted. Despite breastfeeding, he developed excessive weight loss of 16% by day 3 and 18.5% by day 15, with intermittent non-bilious vomiting but normal urine output.

Investigations (if any): Severe renal impairment was detected (creatinine 384 µmol/L, urea 66 mg/dL) with metabolic acidosis (pH 7.22, bicarbonate 12 mmol/L). Ultrasound showed bilateral nephromegaly with multiple renal cysts. Liver imaging and sepsis screening were normal.

Early-onset renal failure, nephromegaly, cystic changes, and failure to thrive supported congenital renal pathology. The neonatal presentation with marked renal enlargement and absence of family history favoured autosomal recessive PCKD (ARPKD). Parental ultrasounds were normal. Hepatic fibrosis was not evident but may evolve later.

Progress: Management was supportive, including correction of acidosis, fluid-electrolyte monitoring, nutritional optimisation, and nephrology input. Parents received counselling regarding prognosis, long-term renal outcomes, and potential need for renal replacement therapy. Genetic counselling and PKHD1 testing were advised.

Discussion: Weight loss beyond 10% after the first week should prompt evaluation. Neonatal PCKD should be considered in infants presenting with early failure to thrive and renal dysfunction.

MANAGEMENT CHALLENGES IN A NEONATE WITH VEIN OF GALEN MALFORMATION AND REFRACTORY CARDIAC FAILURE

U.M. Ruwanpathirana, J.R. Fonseka, G.Sooriyasena,S.Amarasekara

Introduction: Vein of Galen malformation is a rare congenital cerebral Arteriovenous malformation with low resistance intracranial shunting which results heart to generate a high cardiac output in order to maintain systemic perfusion and eventually leads to a decompensated state with increased pulmonary hypertension.

Description: We report a day-one male neonate referred for medical management of hypoxic ischaemic encephalopathy. He was tachycardic with a hyperdynamic precordium, a machinery systolic murmur and significant pre- and post-ductal oxygen saturation differences. He showed signs of right heart failure with warm, well-perfused bounding pulses. Cranial assessment revealed a bruit and bulging fontanelle. During treatment, he developed refractory convulsions. Therapeutic hypothermia was initiated for neuroprotection. Severe pulmonary hypertension was treated with intravenous milrinone, while dopamine and dobutamine provided inotropic support.

Investigations: USS brain done on day one , revealed a cerebral venous malformation (1.6*1,7*2.1cm) related to posterior thalamic region and parenchymal haemorrhages in the vicinity .
2D Echo revealed prominent SVC flow, large PDA and dilated internal jugular veins with features of high output heart failure and severe pulmonary hypertension(TR PG 60 mmhg)

Progress: His Bicêtre score was less than 8.0 suggesting instability for surgical interventions. Despite extensive medical management of heart failure and pulmonary hypertension neonate succumbed to death following severe pulmonary haemorrhage on Day ten of life

Discussion: Clinical recognition in this exceptionally rare, antenatally undetected cases may be difficult, necessitating meticulous bedside examination and evaluation. Poses significant challenges in managing the complications and definitive treatment would be staged endovascular embolization of the shunt. No definitive consensus reached worldwide related to the timing of neurosurgical interventions

BENIGNANT RARENESS WITH A DRAMATIC FAÇADE: ACUTE HAEMORRHAGIC OEDEMA OF INFANCY PRESENTATION IN UNUSUAL AGE

U.M.Ruwanpathirana, W.M.M.R Wickramasinghe,K.H.P.Madushani,A.DeSilva

Introduction:
This self-limiting illness runs a benign course and is a type of small vessel cutaneous vasculitis which usually present in age group of 04 to 24 months. Though aetiology is unknown usually present with a preceding infection, Vaccination or medication use. Present as Classical triad of fever, oedema and purpura predominantly over face and extremities. In contrast to the dramatic eruptions the infants are systemically stable.

Description:
We report a one-month-old infant, previously thriving well, who presented with an acute onset of generalized non blanching purpuric lesions, “target-like” in appearance, predominantly over the trunk and face, without mucosal involvement or oedema. Extremities were spared. The infant was afebrile and clinically well despite the striking skin findings. There were no bleeding manifestations, systemic signs suggestive of infection, or hepatosplenomegaly and nor he was icteric. There was no history to suggest possible congenital infections. Dermatology opinion confirmed the diagnosis.

Investigations
Inflammatory markers were negative along with normal platelet counts
ECG revealed no conduction blocks or arrythmia

Progress: lesion diameter and colour intensity increased initially, but after two weeks had a self-limited resolution without any scarring or complications.

Discussion:
This rare and often under recognized disease presented in the subject infant in an unusual age of one month without oedema, fever or any associated prior illness. Given the early neonatal onset and prominent facial involvement in this well infant neonatal systemic lupus erythematosus (SLE) was considered in the differential diagnosis. Other differentials would be sepsis , congenital infections , thrombocytopenia and coagulopathy.

NUANCES OF ATYPICAL KAWASAKI DISEASE IN INFANCY

U.M.Ruwanpathirana, K.H.P.Madushani , W.M.M.R Wickramasinghe , H.G.H.Udara,T.Munasinghe

Introduction: Kawasaki disease is an acute self-limiting febrile vasculitis of unknown aetiology which is commonly seen in children six months to five years of age. It is characterized by inflammation of medium sized arteries with a predilection for coronary arteries and the most devastating complication is , coronary artery dilatation and aneurysm formation.

Description: We report a three-month-old female infant previously thriving well presenting with a high-grade fever for over five days ,a generalized erythematous macular rash , subtle oral mucosal redness and dry, cracked lips. No strawberry tongue observed. Other major criteria like lymphadenopathy , conjunctival suffusion or extremity changes were not observed. Infant appeared acutely ill
Investigations Biochemical findings were not strongly supportive of the diagnosis, with only a marginally elevated CRP of 40 mg/L and hemoglobin of 10.4 g/dL, providing partial evidence suggestive of an atypical presentation. However, the 2D Echocardiogram demonstrated marginal dilatation(Z score − 2.0-2.5) of LMCA,LAD and RCA consistent with early coronary involvement.

Progress: These findings were suggestive of atypical Kawasaki and treatment started with IV immunoglobulin and a course of oral aspirin and oral steroids. With treatment infant recovered completely along with normalization of inflammatory markers and 2D echo at four weeks showed dilated RCA only.

Discussion: Kawasaki disease in children less than six months is rare and the clinical presentations can be subtle and nonspecific. Therefore, is a diagnostic challenge and need to maintain high index of suspicion in prolonged febrile illness with no clear source of infection. Laboratory and echocardiographic evaluation are essential for diagnosis.

SEVERE NEONATAL-ONSET PROPIONIC ACIDAEMIA PRESENTING WITH RECURRENT METABOLIC CRISES

U.M.Ruwanpathirana, I.Kankanarachchii,W.M.M.R.Wickramasinghe

Introduction:
Propionic acidaemia a rare organic acidaemia , is an autosomal recessive disease from deficiency of mitochondrial enzyme propionyl-CoA carboxylase, leading to accumulation of propionic acid and toxic metabolites.

Description:
A male infant, third child born to consanguineous parents, showed reduced activity since birth. On examination, he was drowsy and moderately dehydrated. There was no acidotic breathing, hypotonia or organomegaly. He was initially managed as presumed sepsis with intravenous antibiotics, but with minimal improvement. Basic investigations including venous blood gas and cranial imaging were unremarkable. There was a sibling death in the family with suspected metabolic disease, warranted metabolic evaluation.

Investigations (if any):
Acyl carnitine profile revealed elevated propionyl carnitine C3 = 17 .06 µM (0.2-4.0 µM), C3 /C2 = 2.88 µM (0.02-0.2 µM), C3 /C16 =8.97 µM (0.12-2.6 µM) . Urine screening for organic acids and blood for aminoacidopathies confirmed the diagnosis .

Progress: At one month, re-presented with a febrile respiratory illness followed by drowsiness and reduced feeding. Metabolic acidosis, pancytopenia (Hb 6.4 mg/dL) and marginal hypoglycaemia indicated a metabolic crisis, managed with fluid and electrolyte resuscitation. A transient neurodevelopmental regression was noted, with gradual improvement

Discussion: Clinical features usually manifest in the neonatal period and exhibit a wide clinical spectrum ranging from catastrophic metabolic decompensations in early infancy to lenient late onset presentations in childhood. Neonates present with nonspecific symptoms mimicking neonatal sepsis, encephalopathy or dehydration, creating diagnostic challenges. Parental consanguinity or unexplained sibling deaths are important clues in clinical suspicion and initiating early treatment would prevent catastrophic permanent impairments.

A GENETICALLY CONFIRMED PATIENT WITH FIBRODYSPLASIA OSSIFICANS PROGRESSIVA

Ranasinghe MM, Viknarajah S, Subasinghe SMV, Gunaratne SA, Ranasinghe MM, Vijitha V, Kumara WTGCR, Nadarajah S

Introduction Fibrodysplasia Ossificans Progressiva (FOP) is a rare, severely disabling autosomal dominant genetic disorder caused by gain-of-function mutations in the ACVR1/ALK2 gene, situated on chromosome 2q23–24. The incidence is noted to be 1 in 2 million and known to affect 2500 people globally. This is characterized by congenital malformations of the great toes and progressive heterotopic ossification within soft tissues, ultimately resulting in restricted mobility.

Description: A 18-month-old male child with enlarging bony hard lumps in the scalp and scapular region in the background of congenital hallux valgus deformity.

Investigations (if any): Blood film showed features of iron deficiency anaemia, Inflammatory markers and bone profile was normal.
Bone marrow aspirate and trephine biopsy – showed reactive marrow with no malignant infiltration.
CECT identified multiple ill-defined scalp lumps in the occipital region and a scalp fluid collection with erosion of the frontal bone in the frontal region, thickening and heterogenous density of right posterior neck and chest wall muscles with inflammatory processes involving the muscles and scalp.
Histopathological examination of an excised lesion confirmed early-stage heterotopic ossification favouring a diagnosis of FOP.
Heterozygous c.617G>A substitution in exon 6 of the ACVR1/ALK2 gene was identified from sequencing analysis of purified ACVR1/ALK2 exon 4 and exon 6 amplicons.

Discussion:
This is the first genetically confirmed case of FOP in Sri Lanka
The first drug shown to control the growth of new bone is Palovarotene.
Flare-ups are being treated with anti-inflammatory medications and conservative management with preventive strategies.

HOMOZYGOUS NONSENSE MUTATION IN THE IL12RB1 GENE CAUSING MENDELIAN SUSCEPTIBILITY TO MYCOBACTERIAL DISEASE

W D V Thilakarathna, W M M R Wickramasinghe, A S Modaragama, K Karunarathna, L G C P Godakanda, C S Wijetunge, D N Nelson

Introduction: Mendelian susceptibility to mycobacterial disease (MSMD) is a defect in the innate immune system characterized by mutations in the IL-12 or IFN-γ pathway. It is inherited by autosomal recessive, dominant, or X-linked recessive patterns, with complete or partial functional impairment, predisposing to non-tuberculous mycobacterial and other infections.

Description: A 2-year-old boy, firstborn to healthy, non-consanguineous parents, presented with a right inguinal lump with pus discharge and fever for one-week. He had a history of recurrent abscesses involving the left axilla (at 4 months), the left parotid (at 9 months), and the right inguinal area (at 15 months), requiring intravenous antibiotics and surgical drainage. His birth was uncomplicated. BCG was given at birth, with no adverse reactions. There was no history of other systemic infections or constitutional symptoms. Growth parameters were normal. Examination revealed right-sided cervical lymphadenopathy (1.5*1.5 cm), Right thigh abscess with pus discharge. Systemic examination was normal.

Investigations (if any): He had neutrophilic leukocytosis, anemia, reactive thrombocytosis, and elevated inflammatory markers. Renal and liver functions, chest X-ray, ultrasound abdomen, and blood cultures were normal. HIV screening was negative. Pus culture from the abscess was positive for Pseudomonas, coliforms, and corynebacteria. Cervical lymph node biopsy revealed non-caseating granulomatous lymphadenitis, and Gene expert of the biopsy detected Mycobacterium species. Immunoglobulin levels and lymphocyte subsets were normal. The nitro blue tetrazolium test was negative. Genetics detected a homozygous nonsense mutation in IL12RB1 gene.

Progress: The child responded well to intravenous antibiotics and antitubercular therapy.

Discussion: Clinical features of MSMD depend on the site of involvement. Diagnosis requires identification of pathogenic mycobacteria, immunological testing, and genetics. Management includes treatment of active infection, long-term antimicrobial therapy, cytokine replacement therapy with interferon gamma, and hematopoietic stem cell transplantation.

CAVERNOUS SINUS THROMBOSIS WITH MRSA SEPTICAEMIA COMPLICATED WITH EXTENSIVE BI FRONTAL CEREBRAL INFARCTIONS FOLLOWING INFECTED ACNE IN DANGER AREA OF FACE

T D N Warnasuriya, Sahani Dias,Ishara Akuratiya,Navoda Kalpani,Isuru Fernando, N.W.D.N Ayeshani, W.M.D.M Peris,I ndika Prasanna, Niluka Gamage, Malinda Dissanayake, W.K.G Hemantha, Pradeep Gamage, Samantha Deshapriya, Thilina Munasinghe, Upeksha Liyanage

Introduction:
Cerebral venous sinus thrombosis is rare in the paediatric age group, with an estimated incidence of 0.67 per 1,00,000 children per year.. Cavernous sinus thrombosis may occur as a complication of infectious and noninfectious processes. Septic thrombosis of the cavernous sinuses most commonly follows infections of the middle third of the face due to Staphylococcus aureus.

Description: A 13 year old previously well boy, admitted with severe heaedache,vomiting and fever for 3 days.There was rapid deterioration of Glass glow coma scale and left eye proptosis, which was worsened over few hours noted. He underwent a urgent Bifrontal decompresive craniectomy.There was a recent history of incision and drainage of left cheek infected acne.

Investigations (if any Non Contrast Computed Tomography (NCCT) Brain- Left sided cavernous sinus thrombosis early cavernous sinus thrombosis in right side
Blood culture - Methicillin Resisstant Staphylococcus Aureus

Progress: He was treated with appropriate IV antibiotics, anticoagulation with S/C enoxaparin and rehabilitation started. He was offered a specially designed wheel chair and sent for rehabilitation and showed slow recovery.

Discussion: Septic thrombosis of the cavernous sinuses most commonly follows staphylococcal infections of the middle third of the face. Orbital symptoms like chemosis and proptosis with headache are constant. The free anastomosis between the venous system of upper face to cavernous sinus predispose to infected septic embolism leading to this complication.Treatment is mainly palliative with antibiotics,anticoagulation and rehabilitation.

A CASE REPORT: RARE PHENOTYPE OF COMBINED INFANTILE SPASMS AND MYOCLONIC SEIZURES ASSOCIATED WITH KCNQ2 MUTATION.

Lukshiga S, Thilina M K T, Jasinghe E, Rathnayake P

Introduction: KCNQ2 mutations are classically associated with neonatal epileptic encephalopathy, though atypical later-onset presentations are increasingly recognised.

Description: We report a rare case of KCNQ2-related epilepsy presenting at four months of age with flexor spasms, persistent polymyoclonus, and developmental regression, in the absence of neonatal seizures. Epileptic spasms, continuous low-amplitude myoclonic movements involving the trunk and head and nystagmus were observed.

Investigations (if any): Metabolic screening and neuroimaging were normal. EEG demonstrated an initial burst-suppression pattern evolving into hypsarrhythmia, with background fast activity noted as an unusual feature. Genetic analysis identified a heterozygous pathogenic KCNQ2 variant (c.431G>A).

Progress:
Treatment with prednisolone following the UK Infantile Spasms Study (UKISS) protocol resulted in complete resolution of spasms, myoclonus, and nystagmus, with marked EEG improvement and catch-up in developmental milestones. Low amplitude polyspikes persisted on follow-up EEGs despite sustained seizure control.

Discussion:
This case highlights an unusual clinical and electroencephalographic presentation of KCNQ2-related epilepsy, characterised by late-onset infantile spasms with persistent low-amplitude polymyoclonus, nystagmus, and distinctive EEG background abnormalities. To our knowledge, persistent interictal myoclonic movements with nystagmus have not been previously reported in this setting. These clinical features, together with background fast activity and regular EEG attenuations, may raise suspicion of an underlying KCNQ2 mutation prompting early genetic evaluation. The parallel response of spasms, myoclonus, and nystagmus to steroid therapy suggests these manifestations may be epileptic in nature and raises the possibility that steroids could favourably influence disease expression in selected KCNQ2-related phenotypes.

A CASE OF PITUITARY STALK INTERRUPTION SYNDROME PRESENTING WITH GROWTH FAILURE AND RECURRENT HYPOGLYCAEMIC EPISODES

T.D.N Warnasuriya, Rajith Jayasanka,N.W.D.N Ayeshani, Sahani Dias, B.S Liyanage, C.L.Chandanayake, Dimarsha de Silva, Upeksha Liyanage

Introduction: Pituitary stalk interruption syndrome (PSIS) is a rare congenital defect with estimated incidence of 0.5 per 100000 live births with uncertain underlying etiology. Patients usually present with anterior pituitary hormone deficiencies with accompanied midline structural abnormalities.

Description: A 2 years and 6 month old boy, born to non-consanguineous healthy parents with uncomplicated birth history presented with recurrent hypoglycaemic episodes since age of 18 month with 5 episodes of hypoglycaemic convulsions. His both height and weight were affected and well below -3 Standard deviation and mid facial hypoplasia noted. He had ketotic hypoglycaemia without hepatomegaly and serum ammonia was normal. His random cortisol level and short synacthen test was normal. He was found to have very low Insulin- Like Growth Factor -1(IGF-1) and growth hormone levels. Other pituitary hormone deficiencies were excluded.

Investigations (if any): MRI brain- absent anterior pituitary and infundibulum with ectopic posterior pituitary at floor of third ventricle is suggestive of pituitary stalk interruption syndrome
Bone age – significantly delayed
IGF 1- 1.74ng/ml(13-143)
Glucagon stimulation test- all values < 1mcg/l, indicating growth hormone deficiency

Progress: He was started on growth hormone therapy soon after the diagnosis and started gaining height and became free of hypoglycaemic episodes.

Discussion: Diagnosis of PSIS primarily relies on pituitary magnetic resonance imaging (MRI), revealing characteristic features such as hypoplastic anterior pituitary, interrupted or absent pituitary stalk, and ectopic posterior pituitary.Treatment is tailored to the hormonal deficiency detected.

SJOGREN LARSSON SYNDROME: A RARE NEURO ICHTHYOTIC SYNDROME - A CASE REPORT

T. Ninuja, S.K. Arulmoli

Introduction: Sjogren Larsson syndrome is a rare autosomal recessive neurocutaneous disorder characterized by ichthyosis, spasticity and neurodevelopmental impairment. Early recognition is essential to facilitate timely supportive interventions and appropriate genetic counselling. We report a case highlighting the evolving neurological manifestations of this rare condition in infancy.

Description: A 1year and 2 month old girl was referred for evaluation of global developmental delay. She was the second child of consanguineous parents, born at term following an uncomplicated antenatal and perinatal period. She had been followed up in the dermatology clinic for ichthyosis. Development was age appropriate until 6month. She experienced her first brief afebrile seizure at 9months, after which developmental delay was noted in all domains. Her elder sibling also under evaluation for developmental delay and ichthyosis. On examination, there were no dysmorphism. OFC was normal. Generalized ichthyosis was present. Neurological examination revealed increased tone in the lower limbs with exaggerated reflexes and clonus, while upper limb examination was normal. Ophthalmological assessment showed no evidence of retinitis pigmentosa.

Investigations (if any): Electroencephalography demonstrated generalised slowing. Magnetic resonance imaging of the brain revealed diffuse white matter changes.

Progress: She was commenced on antiepileptic therapy and baclofen for spasticity, and enrolled in a neurorehabilitation programme. Parents were counselled regarding the diagnosis, prognosis and recurrence risk.

Discussion:
This case highlights the importance of considering rare neurocutaneous syndromes in infants presenting with ichthyosis and evolving neurological features. Early multidisciplinary management and genetic counselling are crucial to optimise outcomes and support affected families.

A RARE COMPLICATION OF A COMMON DISEASE - GRADENIGO SYNDROME

K A P H D Kahandawa, D D C Amarasinghe, S Ganeshan

Introduction: Gradenigo syndrome, is a rare complication of otitis media, which presents as a triad of symptoms; Otorrhea, ipsilateral facial pain in the ophthalmic division of trigeminal nerve and ipsilateral lateral rectus palsy.

Description: A 10-year-old boy presented with left eye weakness, otorrhoea and facial pain. He had a history of left ear pain with discharge, left facial pain for one and half months and left eye weakness for one week. There was a history of fever and upper respiratory symptoms preceding otorrhoea. Examination showed isolated left lateral rectus palsy, no signs of meningism, normal fundoscopy and pupils.

Investigations: Inflammatory markers were normal. Full blood count had mild neutrophilia. Contrast Enhanced Computer Tomography of brain on Day 3 showed left otomastoiditis and osteomyelitis of the petrous temporal bone, para-sinusitis and normal bilateral orbits. Magnetic Resonance Imaging on day 10 revealed left sided otomastoiditis complicated with osteomyelitis and abscess formation in apex of left petrous temporal bone.

Progress:He was managed medically with 21 days of antibiotics with neurosurgical reviews. Follow up MRI after 1 month of completion of treatment showed very small collection of residual abscess which was decided to manage conservatively.

Discussion: Gradenigo syndrome is a rare complication of otitis media in an era of potent antibiotics. Its classical presentation is supported by the anatomical relationships of the middle ear to Abducent and Trigerminal nerves. Prompt identification, aggressive early antibiotic treatment in a patient with neurological symptoms and otitis media is essential to prevent further cranial complications.

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SPONDYLO-METAPHYSEAL DYSPLASIA IN A 2-YEAR-OLD SRI LANKAN GIRL

Piumi Madawala, Piumi Madawala, Chanika Lokuhewage, Sudarshana Bandara, Shobhavi Randeny, Sachith Mettananda

Introduction: Spondylo-metaphyseal dysplasia (SMD) represents a heterogeneous group of rare skeletal dysplasias characterized by abnormalities of the spine and metaphyses, resulting in disproportionate short stature. Due to its rarity and subtle early features, diagnosis is often delayed until characteristic clinical and radiological findings become evident in early childhood

Description: We report a 2-year-7-month-old Sri Lankan girl followed up since birth for extreme short stature. She was born at term to non-consanguineous parents with intrauterine growth restriction and antenatal evidence of short femur length. Despite severe disproportionate short stature, her motor and cognitive development were age-appropriate. Clinical examination revealed a short neck, small thoracic cage with pectus carinatum, bowing of the lower limbs with varus deformity, and short stubby hands and feet. There was no family history of skeletal disorders or extra-skeletal involvement.

Investigations (if any): Baseline haematological and biochemical investigations, including bone profile, were normal. Skeletal survey demonstrated generalized platyspondyly, metaphyseal irregularity and widening of long bones, short square iliac wings, and delayed bone age. Urinary mucopolysaccharide screening was negative, excluding storage disorders

Progress: A diagnosis of spondylo-metaphyseal dysplasia was made based on clinical and radiological findings. Genetic testing could not be performed due to financial constraints. The child is under regular follow-up with growth monitoring and parental counselling.

Discussion: This case highlights the importance of recognizing characteristic skeletal and radiological features of SMD for early diagnosis. Early identification enables appropriate surveillance for potential complications such as kyphoscoliosis and cervical spine instability, and provides essential prognostic counselling to families.

COMMON VARIABLE IMMUNODEFICIENCY 12 (NFKB 1 VARIANT) PRESENTING AS AUTOIMMUNE CYTOPENIA

L G C P Godakanda, M B navarathne, S V Mohan, W M M R Wickramasinghe, W D V Thilakarathna, C S Wijetunge, L N G I Ayesha

Introduction: Common variable immune deficiency (CVID) is a heterogeneous disorder presenting with various clinical manifestations, including autoimmune cytopenias, lymphoproliferation, and granulomas. Heterozygous NFKB1 variants are considered the most common monogenic etiologies of common variable immunodeficiency (CVID). (NF-κB) It is crucial in several biological processes, including adaptive immune response, proliferation, cell survival, and inflammation

Description: A 13-yr-old previously well boy, 3rd born to nonconsanguineous healthy parents, presented with recurrent facial abscess, cervical lymphadenopathy, and was accidentally found to have pancytopenia in a full blood count. Later presented with buttock abscess generalized lymphadenopathy & hepatosplenomegaly

Investigations (if any): He had pancytopenia and elevated inflammatory markers. His renal and liver functions were normal. HIV, virology panel and Mantoux test were negative. Chest X-ray normal, USS abdomen showed moderate hepatosplenomegaly with suspicious lymph nodes. Lymph node biopsy was reactive, bone marrow biopsy, trephine biopsy, immunoglobulin levels, Nitroblue Tetrazolium test were normal. No abnormalities were picked in Upper and Lower GI endoscopies. Flow cytometry findings are suggestive of autoimmune lymphoproliferative syndrome. Genetics identified in the NFKB1 gene NM_003998.4 splice acceptor variant (CVID 12)

Progress: Child responds to IVIG, steroids & Immunosuppressive therapy

Discussion: Clinical features are varied depending on the pathogenic variant in CVID.CVID12 is an autosomal dominant complex immunologic disorder with multisystem involvement & associated with autoimmunity. Diagnosis was helped with basic immunologic panel & genetic testing. Management include regular IVIG, steroids & immunosuppressive therapy.

WHEN CUTANEOUS CLUES REVEAL A HIDDEN MULTISYSTEM DISORDER: A CASE OF GOLTZ SYNDROME

B.K.G.S.S. Dharmasiri, U.M. Ruwanpathirana, K.H.P. Madhushani, H.G.H Udara, I. Kankananarachchi

Introduction: Goltz syndrome, also referred to as Focal Dermal Hypoplasia (FDH) or Goltz–Gorlin syndrome, is a rare X-linked dominant multisystem developmental disorder present at birth. It primarily involves abnormalities of the skin, skeletal system, eyes, and craniofacial structures. First described by Robert Goltz in 1962, the condition necessitates coordinated, multidisciplinary management due to the highly variable developmental involvement across affected organ systems.

Description: A 2-year-old girl born to non-consanguineous parents presented with multiple papular, hypopigmented linear lesions along the lines of Blaschko. Dermatological evaluation showed features consistent with focal dermal hypoplasia. Craniofacial anomalies included a narrow nasal bridge, low-set ears, anodontia, and hypoplastic teeth. She also had dystrophic nails and sparse, brittle hair. Skeletal findings included left foot syndactyly, digital hypoplasia, and a lobster-claw deformity of the left hand. Her developmental milestones were normal, though growth parameters were below age expectations. Systemic examination was unremarkable, and the combined clinical features supported a diagnosis of Goltz syndrome.

Investigations (if any): Blood and urine tests were normal. Long-bone X-rays showed bone hypoplasia and osteopathia striata. Hand and foot X-rays revealed absence of the left third digit and soft-tissue syndactyly. Chest X-ray showed 11 rib pairs. Abdominal and brain ultrasounds were normal. Echocardiography showed aortic regurgitation; ophthalmologic findings were normal l

Progress: The child is receiving multidisciplinary supportive care, including dental management, dermatological follow-up, and plastic surgery follow-up for deformity correction.

Discussion: This case highlights the phenotypic variability of Goltz syndrome, highlights uncommon cardiac and rib anomalies, and underscores the need for comprehensive evaluation and multidisciplinary care.

A NEONATE PRESENTING WITH UNILATERAL HYPOPLASIA OF PECTORALIS MAJOR MUSCLE: A RARE CASE REPORT

S U Atapattu, A C Pieris, R P I Wimalaweera

Introduction:
Complete or partial absence of pectoralis major muscle is a rare congenital anomaly having few differential diagnoses. The first most likely diagnosis is Amazone syndrome which is dominant in females, associated with the absence or hypoplasia of ipsilateral breast and multiple rib abnormalities, and considered extremely rare. Next is Poland syndrome which has similar manifestations, except for male predominance and unilateral hand syndactyly. Although the estimated global incidence of Poland syndrome ranges from 1 in 10,000 to 1 in 100,000, there remains a lack of nationally published epidemiological data specific to these conditions. Other differential diagnoses are conditions associated with Poland syndrome like Moebius syndrome and Sprengel deformity.

Description:
A term female neonate with a birth weight of 2.430kg was delivered via elective LSCS due to breech presentation. APGAR score was normal. After 1hour, baby developed respiratory distress and HHHFNCO2 was started.
General examination revealed right-sided 3 accessory nipples and reduced muscle mass in R/anterior chest wall. Cardiovascular examination was normal.

Investigations (if any):
CXR revealed absence of R/S upper ribs (2nd to 5th), hypoplastic 6th and 7th ribs, lower thoracic and upper lumbar hemivertebrae and scoliosis.
Ultrasound revealed a smaller right-sided pectoralis major muscle.

Progress:
Baby was discharged with orthopedic, cardiology and cardiothoracic referrals as outpatient.

Discussion:
The baby being female and in the absence of hand syndactyly, the diagnosis points more towards Amazone syndrome. Further management involves advanced imaging and a multidisciplinary team approach including cardiothoracic, cardiology, orthopedic, developmental pediatrics and plastic surgical specialties. It is crucial to address functional, developmental, aesthetic and psychological concerns eventually.

RARE PHENOTYPIC MANIFESTATIONS OF COMPOUND HETEROZYGOUS RAG1-DEFICIENCY IN A 4 YEAR OLD GIRL

R D Sandamal, T I Kodikara Arachchi

Introduction: The *RAG1(Recombination Activating gene 1)* is essential for V(D)J recombination, the process by which T-cell receptors and immunoglobulins achieve diversity. This case highlights the multi-systemic complications and diagnostic challenges in a paediatric patient with compound heterozygous RAG1 variant.

Description: A 4-year-old girl presented with recurrent infections starting at 9 months complicated with pyrexia of unknown origin, and extensive varicella infection. At 3years of age she developed, sensoneuronal hearing impairment, vitiligo along with recurrent infections and cytopenias.

Investigations (if any): Investigations revealed deficient in T-lymphocytes with preserved B and NK-cell counts. Genetic testing confirmed two heterozygous variants, one likely pathogenic and other one was uncertain significance in the RAG1 gene.

Progress: The clinical course was complicated by episodic Microangiopathic Haemolytic Anaemia ,autoimmune cytopenias and recurrent peritonitis. Surgical intervention was required for ileo-ileal anastomosis of strangulated umbilical hernia repair surgery and an exploratory laparotomy to investigate intraperitoneal cystic lesions. The latter identified a benign lymphatic cyst likely cystic lymphangioma secondary to chronic peritonitis. Current management includes monthly immunoglobulin (0.4g/kg), prophylactic antibiotics, antifungals, alongside with corticosteroids and cyclosporin for haematological stability.

Discussion: RAG 1 null mutations typically result in T-B-NK+ Severe Combined Immunodeficiency, compound heterozygous variants often lead to "leaky" RAG phenotypes. In this patient, the partial recombinase activity likely allowed for the production of some T-cells, but these cells are often oligo-clonal and autoreactive. This explains the paradoxical presentation of immunodeficiency alongside dysregulated autoimmunity. Such cases necessitate a highly integrated, multidisciplinary approach to modulate disease activity and mitigate organ-specific complications. While current management is supportive, hematopoietic stem cell transplantation remains the definitive therapeutic intervention.

A NEONATE WITH GENETICALLY CONFIRMED CHRONIC GRANULOMATOUS DISEASE

Mahesha Madhavi Ranasinghe, Ranasinghe MM, Amarasinghe AAGP, Sivakumar Prranavethya, Rajiva de Silva, Danushka Dassanayake, Tharindi Suriapperuma, Shobhavi Randeny, Rasika Gunapala

Introduction: Chronic granulomatous disease (CGD) is a rare inherited immunodeficiency characterised by neutrophil function defect that carry a high risk of invasive bacterial and fungal infections, most commonly by S. aureus and Aspergillus spp.

Description: A term baby boy who was born to healthy nonconsanguineous parents was evaluated for an unresolved fever at the age of six weeks. His initial infection was at the age of three weeks, diagnosed as MRSA-positive otitis media. He had pyelonephritis and bronchopneumonia, subsequently requiring treatment in intensive care. The examination revealed lymphadenopathy, BCG flare, hepatosplenomegaly, and skin nodules.

Investigations: Initial evaluation revealed elevated inflammatory markers and negative blood cultures. The continuation of fever despite adequate treatment led to second line investigations to exclude an underlying cause. Further revealed a negative Gene Expert for tuberculosis and HIV screening. Contrast-enhanced CT abdomen revealed multiple small, ill-defined hypoechoic areas in the spleen, suggestive of microabscesses.
Flow cytometry of lymphocyte subsets was normal. The Nitroblue tetrazolium test was positive for CGD, and X-linked recessive CGD was confirmed by genetics.

Progress He was commenced on antifungal and antiviral therapy to cover the infections. Splenic abscesses were managed medically. He is currently on antibiotic, antifungal prophylaxis and antituberculosis treatment while considering an early bone marrow transplant.

Discussion: CGD is one of the primary immunodeficiencies that should be suspected with severe, recurrent, or atypical pyogenic or fungal infections or abscesses in early infancy. Early diagnosis allows initiation of prophylactic therapy and definitive management.

A CASE OF PENTALOGY OF CANTRELL, (THORACO-ABDOMINAL SYNDROME); A RARE AND COMPLEX NEONATAL CONDITION

Mahesha Madhavi Ranasinghe, Ranasinghe MM, MM Sagarika Priyadarshani,Padmasiri KEGP, Wickramaratne SS, Weeratunga ND, Shobhavi Randeny

Introduction: Pentalogy of Cantrell (POC) is a rare but severe congenital anomaly due to defective development of the mesoderm during early gestation. This is a constellation of five anterior midline anatomical defects involving thoracoabdominal region, in supraumbilical abdominal wall, lower sternum, anterior diaphragm, and pericardium, along with congenital cardiac malformations. The incidence ranges from 1 in 65,000 to 1 in 500,000 live births while only 200 cases reported worldwide.

Case: A male neonate was the first-born to nonconsanguineous healthy parents at term with an average birthweight and normal Apgar score following uncomplicated antenatal period. Baby was found to have a congenital diaphragmatic hernia and omphalocele, requiring immediate neonatal intensive care.

Investigations : Chest Xray revealed right sided diaphragmatic hernia which was confirmed by CT imaging showing a defect in the lower sternum. Echocardiogram revealed a large ventricular septal defect and overriding aorta with normal pulmonary valve.

Progress: Surgical repair of the extracardiac defects were done on day 2 of life where a complete POC was confirmed. Surgical findings were supraumbilical omphalocele, ventral defect of the diaphragm along with sternum leading to herniation of the bowel into the anterior mediastinum, and absent pericardium (heart located at the base of the defect).
Postoperative period was complicated with pneumothorax, persistent pulmonary hypertension, and circulatory failure where the baby succumbed to death due to cardiogenic shock by day six of life.

Discussion: Average survival of POC is 37% while that of the complete form is further guarded, mean survival being 36 hours. Management is challenging warranting a multidisciplinary, interprofessional approach.

DOWNS SYNDROME WITH MOYA MOYA DISEASE PRESENTING WITH UNEXPECTEDLY SEVERE DEVELOPMENTAL ABNORMALITIES-CASE REPORT

Karunarathna WACN, Sumanasena SP, Warnakula WAIN, Mannapperuma MBN, Kapila K, Hashan Pathiraja, Aloka Prabath, Wijayasinghe WAPN, Dasunika MS, Nayana Samarasinghe, Mihiri Rubasinghe, Ratnayake P

Introduction:
Moya-moya disease (MMD) is a rare, progressive cerebrovascular disorder with stenosis or occlusion of the intracranial arteries and development of abnormal collaterals. The risk of developing MMD in children with trisomy 21 is significantly higher than in the general population.

Description
An 8-year-old boy with downs syndrome presented with global developmental delay, absent speech, profound bilateral sensorineural hearing loss, and features of autism spectrum disorder. Gross motor milestones, fine motor skills, speech, social interaction, and communication were markedly impaired. He was hyperactive and demonstrated self-injurious behavior. He was fully dependent on caregivers for activities of daily living. There was no history of seizures, transient ischaemic attacks, or developmental regression. Clinical examination revealed cortical visual impairment (stage 2), horizontal nystagmus, hypotonia, broad-based gait. Muscle power was 4/5 with normal deep tendon reflexes and a negative Babinski sign.

Investigations (if any)
Routine MRI for pre-cochlear implant demonstrated MMD with right middle cerebral artery territory cortical laminar necrosis secondary to infarction.
Progress : The child was started on antiplatelet therapy with aspirin.

Discussion:
Moya-moya disease may cause slowly progressive infarctions with minimal or subtle neurological manifestations. This case highlights the importance of considering MMD in children with Down syndrome with unexpectedly severe neuro developmental, visual and behavioural features, even in the absence of clinically apparent cerebrovascular events.

WOLMAN DISEASE: A GENETICALLY CONFIRMED CASE REPORT FROM SRI LANKA

Kayalvily Perinpanayagam, Harshika Wanniachchige, Deepal Perera, Vajira H.W. Dissanayake

Introduction: Wolman disease is a rare, severe autosomal recessive lysosomal storage disorder caused by deficiency of lysosomal acid lipase (LAL) due to pathogenic variants in the LIPA gene. The estimated global incidence is less than 1 in 100,000 live births. It typically presents in early infancy with progressive hepatosplenomegaly, adrenal calcification, cholestatic jaundice, dyslipidemia, and rapid clinical deterioration. Without treatment, the disease is usually fatal within the first year of life.

Description: We report the case of a two-month-old infant born to second-degree consanguineous parents who presented with prolonged jaundice, pallor, and progressive abdominal distension. There were no significant gastrointestinal or metabolic symptoms at disease onset. On examination, the infant was deeply icteric with massive, firm hepatosplenomegaly.

Investigations: Laboratory evaluation revealed pancytopenia, deranged liver function tests, and an atherogenic lipid profile. Abdominal imaging demonstrated bilateral adrenal calcifications. Bone marrow examination showed foamy histiocytes, while liver biopsy revealed features of micronodular cirrhosis. Based on the clinical, biochemical, and radiological findings, a presumptive diagnosis of Wolman disease was made. However, confirmatory enzyme assay and molecular testing could not be performed due to financial constraints.

Progress: Despite supportive management, baby succumbed within one week of hospitalization. Subsequently, a sibling presented with a similar clinical phenotype and was genetically confirmed to have Wolman disease due to a homozygous pathogenic deletion in exon 6 of the LIPA gene.

Discussion: To our knowledge, this represents the first genetically confirmed case of Wolman disease from Sri Lanka. This case underscores the importance of early clinical suspicion, access to genetic confirmation, and appropriate genetic counselling, particularly in consanguineous families.

WOLF HIRSCHHORN SYNDROME- A RARE CHROMOSOMAL DELETION DISORDER

Y H Samarakoon, I A D Chathurangi, A S Modaragama, K A A L Jayanthi, J A A J Jayasinghe, K C S Dalpatadu, D C DeSilva

Introduction: Wolf–Hirschhorn syndrome (WHS) is a rare chromosomal deletion disorder due to a sub telomeric deletion of short arm of chromosome 4 (4p16.3) with female predominance. It is characterized by craniofacial features (“Greek warrior helmet”), growth failure, developmental delay, hypotonia, seizures with multisystem involvement including cardiac, skeletal, ocular, genitourinary depending on size of deletion.

Description: 11-month-old boy, with global developmental delay, failure to thrive, presented with status epilepticus precipitated by fever. He was born at term by vaginal delivery. birth weight- 1.5 kg, length-43cm, OFC- 29cm, and treated for neonatal sepsis and jaundice. At birth, noted to have dysmorphic features with thinned corpus callosum on cranial ultrasound, ostium secundum Atrial septal defect (OS ASD) in echocardiogram and defaulted thereafter. At 10 months, he developed fever provoked convulsions.
His developmental age is around 8 months. Examination revealed occipital flattening, fair skin, sparse hair with virtually absent bi temporal scalp hair, sparse eye lashes and eyebrows, microstomia, micrognathia, bilateral single palmer creases, hypospadias, hypotonia, systolic murmur. Remaining systemic examination was normal.

Investigations (if any): Full blood count, renal, liver, thyroid functions, lumbar puncture normal. CT brain- partial agenesis of the corpus callosum. Electroencephalogram- consistent with structural pathology with more involvement of right hemisphere. Echocardiogram- OS ASD. Ultrasound abdomen- normal. Hearing and Ophthalmological assessments- normal. Karyotyping confirmed 46,XY,del(4)(p16).

Progress: Child is on multidisciplinary follow-up and antiepileptic therapy. MRI brain is awaiting.

Discussion: In WHS karyotyping can detect large deletions, microdeletions require fluorescent in situ hybridisation or chromosomal microarray for diagnosis. Early recognition enables multidisciplinary care and genetic counselling.

NEWLY DIAGNOSED AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME IN A YOUNG CHILD: A DIAGNOSTIC CHALLENGE

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Introduction: Autoimmune Lymphoproliferative Syndrome (ALPS) is a rare inherited disorder of defective lymphocyte apoptosis, most commonly due to mutations in the FAS gene. It is characterised by chronic non-malignant lymphadenopathy, splenomegaly and autoimmune cytopenias, particularly autoimmune haemolytic anaemia (AIHA) and thrombocytopenia. Early recognition is essential to avoid misdiagnosis as malignancy and to guide appropriate immunomodulatory therapy

Description:A previously healthy 3-year-old girl presented with acute onset shortness of breath for three days. There was no history of bleeding, dark urine, previous similar episodes, recent infections or travel. Family and perinatal histories were unremarkable. On examination, she was afebrile, well nourished but icteric and markedly pale, with tachycardia and normal blood pressure. She had multiple firm, mobile, non-tender cervical, axillary and inguinal lymph nodes (up to 2 cm). Abdominal examination revealed massive splenomegaly (10 cm below costal margin) and hepatomegaly (4 cm).

Investigations (if any):She had pancytopenia with haemoglobin 2.4 g/dL, white cell count 1.18 $\times 10^9$/L and platelets 2 $\times 10^9$/L. Direct Coombs test was positive, with elevated LDH (622 U/L), reticulocyte count (20.6%) and indirect hyperbilirubinaemia. ANA and anti-dsDNA were negative. Bone marrow examination showed hypercellular erythropoiesis without malignant infiltration. CECT revealed generalized lymphadenopathy and hepatosplenomegaly. Lymph node histology showed reactive architecture. Flow cytometry demonstrated elevated double-negative T cells. Genetic testing could not be performed.

Progress:She was initially treated with oral prednisolone (2 mg/kg/day) without response, followed by intravenous methylprednisolone pulses (30 mg/kg/day for three days), resulting in marked haematological improvement. Steroids were gradually tapered.

Discussion:ALPS is a chronic, heterogeneous condition often underdiagnosed due to overlapping features with malignancy and autoimmune disorders. Elevated double-negative T cells with autoimmune cytopenias are key diagnostic hallmarks. Early recognition allows timely immunosuppression, reduces steroid dependence and improves long-term outcomes.

A RARE PRESENTATION OF DUAL GENETIC ETIOLOGIES: CONGENITAL MYOPATHY WITH HYPOPARATHYROIDISM

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Introduction:
Dual pathology in a clinical setting is rare. We report a case with 2 genetic conditions; Tubular Aggregate Myopathy (TAM) and hypoparathyroidism.
TAM is an autosomal dominant congenital myopathy caused by variant in STIM1 gene. Genetic & rare diseases information center does not have epidemiological data of TAM
Hypoparathyroidism, caused by variant in PTH gene leads to decreased serum calcium and increased serum phosphate due to insufficient parathyroid hormone. While worldwide data is lacking, prevalence in Japan is reported as 7.2/million.

Description:
A 16-year-old girl, born to nonconsanguineous healthy parents without any family history, evaluated for proximal muscle weakness since age1 ½ yrs. At age 14, she had fractures of right femur and forearm & carpopedal spasms.
On examination, had waddling gait, positive Gower's sign, calf muscle-hypertrophy. The power was 4/5 on MRC scale and reflexes normal. There was no upper limb involvement.
Investigations revealed creatine phosphokinase-3710 IU/L and chronic atrophic changes with fatty replacement in muscles on Muscle MRI.
Initial analysis of Whole exome sequencing (WES) picked up a heterozygous pathogenic variant inSTIM1 gene; c.326A>G (p. His109Arg). Reanalysis of WES had been requested following detection of low calcium (1.93mmol/L), increased phosphate (1.82mmol/l), low ALP (55U/L) and low normal PTH (2.61pmol/L), when a likely pathogenic variant in PTH gene (c.2T>C (p. Met1?)) has been identified.
Calcium and vitamin D supplements started and continued. Brother's biochemical evaluation was normal.

Investigations (if any):-

Progress: Clinically improved

Discussion:
This case report highlights the possibility of 2 rare diseases co existing. Unifying diagnosis is helpful to avoid diagnostic dilemma and provide targeted management and genetic counselling.

EARLY-ONSET ALEXANDER DISEASE IN INFANCY: A GENETICALLY CONFIRMED CASE WITH EMERGING THERAPEUTIC IMPLICATION

Mannapperuma MBN, Karunarathna WACN, Kunasingham K, Sumanasena S

Introduction: Alexander disease is a rare, progressive leukodystrophy caused by pathogenic variants in the GFAP gene, with infantile-onset forms often presenting with macrocephaly, seizures, and developmental delay. Early diagnosis is crucial for prognostication, supportive management, and emerging disease-specific therapies.

Description: We report a 1-year-1-month-old boy, born at term via elective lower segment caesarean section to non-consanguineous healthy parents, with a birth weight of 2.02 kg. He presented with global developmental delay and experienced his first afebrile focal seizure at 5 months of age, characterized by rightward staring without generalized tonic–clonic activity. A second seizure occurred one week later, prolonged for over four hours, requiring intensive care admission. Clinical examination revealed macrocephaly and growth failure. There was no history of developmental regression to date.

Investigations (if any): Baseline metabolic investigations, including acylcarnitine and amino acid profiles, were normal. Neuroimaging demonstrated features consistent with Alexander disease. Genetic testing confirmed a pathogenic GFAP mutation, establishing the diagnosis.

Progress: Seizures are currently well controlled with antiseizure medications, along with adjunctive thiamine and biotin. The child is receiving multidisciplinary rehabilitation, including physiotherapy and developmental interventions, with regular follow-up.

Discussion: This case highlights the importance of considering Alexander disease in infants presenting with macrocephaly, seizures, and developmental delay. Early genetic confirmation enables anticipatory guidance, targeted supportive care, and consideration for novel therapies. The child is currently being evaluated for eligibility in a clinical trial involving Zilganersen (ION373), an emerging antisense oligonucleotide therapy, underscoring the evolving therapeutic landscape in rare neurogenetic disorders.

PRIMARY HAEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS TRIGGERED BY EPSTEIN–BARR VIRUS

Mannapperuma MBN, Sewwandi SDL, Amarasinghe G, Gunapala R

Introduction: Primary haemophagocytic lymphohistiocytosis (pHLH) is a rare, life-threatening hyperinflammatory disorder caused by defects in cytotoxic lymphocyte function. Early diagnosis is challenging, particularly in low-resource settings, due to overlapping features with severe infections and malignancies. Central nervous system (CNS) involvement further complicates recognition and management.

Description: We report a 5-year-old girl, born to second-degree consanguineous parents, who presented with prolonged fever, lymphadenopathy, hepatosplenomegaly, cytopenias, and failure to thrive. She initially fulfilled HLH-2004 diagnostic criteria and was diagnosed with Epstein–Barr virus (EBV)–triggered HLH. Following initial treatment with corticosteroids and antivirals, she achieved remission. Three months later, she re-presented with afebrile seizures and raised intracranial pressure. Neuroimaging revealed cerebellitis with obstructive hydrocephalus, and cerebrospinal fluid was positive for HSV-1.

Investigations (if any): Laboratory evaluation demonstrated hyperferritinaemia, hypertriglyceridaemia, and hypofibrinogenaemia. Bone marrow examination initially showed haemophagocytosis. During relapse, repeat marrow evaluation and genetic testing identified a pathogenic mutation in the UNC13D gene, confirming primary HLH.

Progress: The child required multidisciplinary management including neurosurgical intervention, antivirals, immunoglobulin, corticosteroids, and escalation to oncology-led immunochemotherapy with etoposide and cyclosporin. She experienced another relapse but responded well to re-treatment. She is currently awaiting haematopoietic stem cell transplantation.

Discussion: This case highlights the diagnostic complexity of pHLH, the potential for sequential viral triggers, and the importance of considering HLH in children with relapsing or isolated CNS manifestations. Early multidisciplinary involvement and timely escalation of therapy were crucial to the favourable outcome.

A CASE OF RARE LEUKODYSTROPHY OF ASPA GENE MUTATION

JAYAWICKRAMA RDP, SUMANASENA. S, VIPULAGUNA D, DALPADADU K, DE SILVA DEEPTHI, ATHAPATTU T, RATHNAYAKE P, Karunathilake KGLRAD

Introduction:
Canavan disease is a rare autosomal recessive leukodystrophy caused by mutations in the ASPA gene, causing accumulation of N-acetyl aspartic acid and diffuse white matter degeneration.

Description:
Master A , presented at one year three months of age with developmental regression. He was born at term to non-consanguineous healthy parents with normal development until four months , after which he developed bronchopneumonia complicated by meningitis. Following this , there was progressive loss of milestones.
He had macrocephaly with preserved growth parameters, significant irritability, minimal visual interaction due to cortical visual impairment but showed auditory responsiveness. There was minimal social interactions ,absence of reaching or grasping . Had marked dystonia and hypertonia of all four limbs, partial head control, and was unable to sit with support.

Investigations (if any)
He had normal hematological parameters and negative metabolic screening. EEG demonstrated an immature background for age. MRI brain revealed markedly delayed myelination with involvement of the posterior limb of the internal capsule, posterior corpus callosum, and globus pallidi. Ophthalmological evaluation showed bilateral early optic atrophy.USS abdomen was normal.Whole exome sequencing identified a likely pathogenic heterozygous frameshift termination mutation in the ASPA gene, with autosomal recessive inheritance.

Progress:
The baby was restarted on antispastic medications with gradual dose escalation and enrolled in early intervention including cortical visual impairment–specific therapy and orthotic support. Over follow-up, mild improvement in tone, head control, and social engagement was observed.

Discussion:
This case highlights the importance of early recognition, genetic confirmation, and multidisciplinary rehabilitation in neurodegenerative leukodystrophies, with appropriate family counselling despite limited disease-modifying treatment options.

WHEN SCHOLASTIC DECLINE SIGNALS A METABOLIC DISORDER: NIEMANN-PICK DISEASE TYPE C

H.G.H. Udara, K.H.P. Madhushani, U.M. Ruwanpathirana, B.K.G.S.S. Dharmasiri, I. Kankananarachchi, T.M. Munasinghe

Introduction: Niemann–Pick disease type C (NPC) is a rare autosomal recessive lysosomal storage disorder, commonly due to NPC1 gene mutations. The condition is characterised by progressive neurodegeneration with variable systemic involvement, frequently resulting in delayed diagnosis. We report the case of a young boy with NPC who was referred primarily because of declining school performance.

Description: A 7-year-old boy, born to 3rd degree consanguineous parents was referred to paediatrician for further evaluation of poor school performance. His early development was appropriate however subtle neurodevelopmental regression became evident between 5 - 6 years of age. Presenting features included gait abnormalities, recurrent falls, clumsiness and impaired comprehension. There was no history of neonatal jaundice or seizures. Examination revealed microcephaly, dysarthria, vertical supranuclear gaze palsy, mild ataxia, moderate splenomegaly, mild hepatomegaly. Cherry – red spots were absent. Cognitive assessment showed a fluid IQ of 56.

Investigations: Full blood count and EEG were normal. Bone marrow biopsy demonstrated foamy macrophages and sea-blue histiocytes. Biomarkers supportive of NPC were markedly elevated; serum Chitotriosidase - 2424 µmol/hr/ml (reference 9–50) and CCL18 - 219.02 ng/mL (reference 1–72). MRI brain and genetics are awaited.

Progress: The child shows gradual neurodevelopmental regression and is receiving multidisciplinary supportive care. Arrangements are currently underway to initiate Miglustat, a substrate reduction therapy, which is not readily available locally.

Discussion: This case highlights poor academic performance as an early and easily overlooked presentation of NPC and underscores the challenges in timely diagnosis and access to disease-specific therapy in resource-limited settings.

DIARRHOEA-7, PROTEIN-LOSING ENTEROPATHY TYPE: ROLE OF GENETIC TESTING IN PROVIDING AN ALTERNATIVE DIAGNOSIS IN A CASE OF REFRACTORY CROHN'S DISEASE

Mithun Samaranayake, Wathsala Hathagoda, Shaman Rajindrajith, Kapila Panditha

Introduction: Diarrhoea-7, protein-losing enteropathy type (DIAR7) is a rare autosomal recessive disorder characterised by early-onset watery diarrhoea and protein loss, caused by pathogenic variants in the DGAT1 gene. We report a case highlighting the pivotal role of genetic testing in resolving a diagnostic dilemma in an infant initially diagnosed with early-onset Crohn's disease (CD) refractory to conventional therapy.

Description: An 8-month-old previously well girl presented with a two-week history of persistent watery diarrhoea and bilateral lower-limb oedema. She was the third child born to consanguineous parents. On examination, bilateral pedal oedema was noted, while growth parameters and developmental milestones were appropriate for age. Upper gastrointestinal endoscopy and ileocolonoscopy were performed, and histology showed features of colitis. Based on these findings, a diagnosis of early-onset Crohn's disease was considered, and treatment was initiated with intravenous methylprednisolone followed by oral prednisolone, azathioprine, and infliximab. Despite therapy, the oedema persisted and required repeated albumin infusions, raising concern regarding an alternative diagnosis.

Investigations (if any): An 8-month-old previously well girl presented with a two-week history of persistent watery diarrhoea and bilateral lower-limb oedema. She was the third child born to consanguineous parents. On examination, bilateral pedal oedema was noted, while growth parameters and developmental milestones were appropriate for age.
Upper gastrointestinal endoscopy and ileocolonoscopy were performed, and histology showed features of colitis. Based on these findings, a diagnosis of early-onset Crohn's disease was considered, and treatment was initiated with intravenous methylprednisolone followed by oral prednisolone, azathioprine, and infliximab. Despite therapy, the oedema persisted and required repeated albumin infusions, raising concern regarding an alternative diagnosis.

Progress: Genetic counselling was provided to the parents. The patient had received three doses of infliximab prior to the genetic diagnosis. Nutritional assessment was undertaken, and dietary modifications were advised with specialist dietetic input.

Discussion: This case underscores the critical role of molecular diagnostics in infants with treatment-refractory inflammatory bowel disease–like presentations. Early age of onset, consanguinity, atypical clinical features, and poor response to immunosuppressive therapy should prompt evaluation for monogenic enteropathies. Establishing a precise genetic diagnosis allowed for more targeted management and avoided unnecessary escalation of immunosuppressive therapy.

ATYPICAL PRESENTATION OF POLYOSTOTIC FIBROUS DYSPLASIA INVOLVING THE MANDIBLE, SPHENOID BONES AND TEMPOROMANDIBULAR JOINT IN A CHILD: A CASE REPORT

Nisangika Damayanthi, A Padeniya, B R R N Gunaratne, S L Mohotti, P Ajanthan

Introduction: Fibrous dysplasia (FD) is a benign developmental bone disorder caused by post-zygotic activating mutations of the GNAS gene, resulting in replacement of normal bone with fibro-osseous tissue. It presents as monostotic or polyostotic disease, usually during childhood. Craniofacial involvement is common; however, temporomandibular joint (TMJ) involvement is rare and may mimic temporomandibular disorders or primary headache syndromes, leading to misdiagnosis.

Description: A 12 year old Sri Lankan girl presented with a two and a half year history of recurrent frontal headaches occurring weekly, without migrainous features. She was treated as migraine without improvement. Over six months, headaches subsided and were replaced by progressive left sided TMJ pain. Initially responsive to analgesics, the pain became refractory, prompting a provisional diagnosis of trigeminal neuralgia. Examination revealed swelling, tenderness over the left TMJ, and restricted mouth opening. Systemic examination was otherwise normal.

Investigations (if any):Baseline laboratory investigations were unremarkable except for a transiently elevated ESR. TMJ ultrasonography showed reactive cervical lymph nodes without joint effusion. Contrast-enhanced CT demonstrated an expansile ground-glass lesion involving the left mandibular ramus, angle, condyle, and sphenoid bone, consistent with polyostotic fibrous dysplasia.

Progress:Intraoperative findings confirmed cortical bone irregularity, supporting the radiological diagnosis and awaiting bone biopsy report.

Discussion:TMJ involvement in pediatric fibrous dysplasia is rare and may present with headache or facial pain, mimicking migraine or trigeminal neuralgia. This case emphasizes the importance of early imaging in children with atypical or treatment-resistant craniofacial pain to prevent diagnostic delay.

MULTIPLE PROXIMAL SMALL INTESTINAL ATRESIA IN A NEONATE

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Introduction: Small bowel atresia is a common cause of neonatal intestinal obstruction, however, multiple atretic segments involving the proximal small intestine are rare. Such cases are associated with increased surgical complexity and a higher risk of postoperative morbidity, including short bowel syndrome.

Description: A 1-day-old term male neonate, born by elective caesarean section to non-consanguineous parents after ten years of primary subfertility, was admitted soon after birth with antenatally detected small bowel atresia and polyhydramnios. The baby had not passed meconium and developed progressive abdominal distension with bilious gastric aspirates. On examination, he was hemodynamically stable with a distended abdomen, and no external congenital anomalies were identified.

Investigations: Antenatal ultrasonography demonstrated dilated proximal bowel loops. Postnatal abdominal radiography showed multiple dilated proximal bowel loops with paucity of distal gas, suggestive of intestinal atresia. Routine laboratory investigations were within normal limits. Echocardiography revealed a small apical muscular ventricular septal defect, while renal ultrasonography was normal.

Progress: Exploratory laparotomy revealed near absence of small intestinal mesentery, with blood supply arising from the ileocolic artery. Multiple atretic segments extended from the duodenojejunal flexure to the distal ileum, with a narrow but patent sigmoid colon. Excision of atretic segments with primary anastomosis was performed. Postoperatively, the neonate had an uncomplicated course, tolerated enteral feeds, and showed satisfactory growth.

Discussion: Multiple proximal small bowel atresia likely results from intrauterine vascular insults and carry a poorer prognosis due to limited residual bowel. Early surgical intervention, bowel preservation, and multidisciplinary care are essential to optimize outcomes.

MULTIPLE PROXIMAL SMALL INTESTINAL ATRESIA IN A NEONATE

Ayeshani N W D N, Liyanage U B, Liyanage J, Peiris W M D M, De Silva B K T, Karunarathne V S W

Introduction: Small bowel atresia is a common cause of neonatal intestinal obstruction, however, multiple atretic segments involving the proximal small intestine are rare. Such cases are associated with increased surgical complexity and a higher risk of postoperative morbidity, including short bowel syndrome.

Description: A 1-day-old term male neonate, born by elective caesarean section to non-consanguineous parents after ten years of primary subfertility, was admitted soon after birth with antenatally detected small bowel atresia and polyhydramnios. The baby had not passed meconium and developed progressive abdominal distension with bilious gastric aspirates. On examination, he was hemodynamically stable with a distended abdomen, and no external congenital anomalies were identified.

Investigations: Antenatal ultrasonography demonstrated dilated proximal bowel loops. Postnatal abdominal radiography showed multiple dilated proximal bowel loops with paucity of distal gas, suggestive of intestinal atresia. Routine laboratory investigations were within normal limits. Echocardiography revealed a small apical muscular ventricular septal defect, while renal ultrasonography was normal.

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Discussion: Multiple proximal small bowel atresia likely results from intrauterine vascular insults and carry a poorer prognosis due to limited residual bowel. Early surgical intervention, bowel preservation, and multidisciplinary care are essential to optimize outcomes.

EOSINOPHILIC MENINGITIS AND HEPATIC LESIONS IN A CHILD WITH SEVERE EOSINOPHILIA

Ayeshani N W D N, Liyanage U B, Peiris W M D M, de Silva B K T

Introduction: Eosinophilic meningitis is defined as >10 eosinophils/mm3 of CSF or at least 10% leukocytes in the CSF are eosinophils. The most common cause is CNS infection with helminthic parasites.

Description: 10-year-old girl presented with fever, headache and neck pain for four days. On examination, she had neck stiffness, while Kernig's sign was negative. She was conscious, rational, and had no seizures. Neurological examination, including fundoscopic assessment, was normal. Mild splenomegaly was noted. She had significant exposure to household pets and history of raw green salad consumption.

Investigations (if any): Investigations revealed marked peripheral eosinophilia (WBC 35,140/mm^3; eosinophils 24,380/mm^3, 69%). CSF cytology showed mild-to-moderate eosinophilic infiltrates. Stool examination and serology for Toxocara and Toxoplasma were negative. Abdominal ultrasonography showed focal hepatic lesions with splenomegaly. Serum IgE was elevated at 3799 IU/ml. Chest X-ray, contrast-enhanced CT brain, and echocardiography were normal.

Progress: The child was treated with oral albendazole for two weeks under multidisciplinary care of Paediatrician, Neurologist, Haematologist, Parasitology team, with marked clinical and haematological improvement and resolution of hepatic lesions. Bone marrow biopsy was deferred due to favourable response. The most probable diagnosis was parasitic eosinophilic meningitis, likely Angiostrongylus cantonensis.

Discussion: Any tissue-migrating helminth may cause eosinophilic meningitis, most commonly A. cantonensis. Transmission through undercooked snails, prawns or crabs and pet exposure. Infectious non parasitic causes and hyper-eosinophilic syndrome, malignancy also may contribute. Diagnosis is difficult due to limited confirmatory tests. Management is mainly supportive; Antihelminthics and steroids should be used cautiously. Follow-up is essential to exclude hyper-eosinophilic syndrome.

WHEN CLUMSINESS ALARMS; A RARE CASE OF PAEDIATRIC ONSET MULTIPLE SCLEROSIS PRESENTING WITH CEREBELLAR SYMPTOMS

K.H.P. Madhushani, H.G.H. Udara, U.M. Ruwanpathirana, B.K.G.S.S. Dharmasiri, T.M. Munasinghe

Introduction: Paediatric Onset Multiple Sclerosis (POMS) is a rare, demyelinating autoimmune disorder of the central nervous system affecting children under 18 years. It exhibits a relapsing and remitting course of neurological events without encephalopathy, separated in time and space. POMS presenting with cerebellar symptoms frequently leaves a challenge for diagnosis.

Description: We report an 8-year-old girl referred to neurologist due to a left sided progressive intention tremor. She had experienced an undocumented episode of transient left lower limb weakness two months earlier. Examination revealed left sided cerebellar signs with otherwise normal upper limb, lower limb and cranial nerve neurology. No bladder and bowel dysfunction or features of encephalopathy were detected.

Investigations: Cerebrospinal fluid (CSF) analysis was otherwise normal with elevated CSF Ig G index but CSF oligoclonal bands were negative. MRI brain revealed multiple periventricular, juxtacortical white matter lesions with left middle cerebellar peduncle lesion showing features of dissemination in time and space compatible with Multiple sclerosis. MRI Pan spine and Visual Evoked Potentials were normal.

Progress: Child was treated with intravenous methylprednisolone pulses, intravenous immunoglobulin and intravenous Rituximab. Fingolimod, a disease modifying treatment for POMS was started later in the course due to initial local unavailability. She showed significant clinical improvement with minimal residual neurological deficits and has remained relapse-free to date.

Discussion: POMS should be considered early in children presenting with progressive cerebellar symptoms without encephalopathy. Early diagnosis and timely initiation of disease-modifying therapy are crucial for favourable neurological outcomes.

SECOND CHILD WITH IMERSLUND-GRÄSBECK SYNDROME IN THE SAME FAMILY PRESENTING WITH CHRONIC CONSTIPATION

Sivakumar Prranavethya, M. M. Sagarika Priyadarshani, K. E. G. P. Padmasiri, Ranasinghe M. M, Sachith Mettananda

Introduction: Imerslund-Gräsbeck syndrome is an extremely rare autosomal recessive disorder of vitamin B_{12} malabsorption. This syndrome may present with proteinuria in addition to the clinical features of vitamin B_{12} deficiency.

Description: A 2 years and 4 months old girl, the second-born child of non-consanguineous parents, presented with chronic constipation for 6 months with suboptimal response to treatment. Her elder brother being diagnosed with Imerslund–Gräsbeck syndrome at 4 years of age when he presented with lower limb weakness and anaemia. Additionally, both her mother and elder brother have α-thalassemia trait. The physical examination revealed moderate pallor and systemic examinations were normal.

Investigations (if any): Haemoglobin 7.8 g/dL (normal 11.0-14.0), with normal mean corpuscular volume. The blood picture showed the appearance was of thalassemia trait with vitamin B12 or folate deficiency. Serum vitamin B12 of 87 pg/mL (normal: 187-883), red cells folate of 100.8 ng/mL (normal: 126-651), Urine albumin: creatinine ratio of 15.7 mg/mmol (normal: <3).

Progress: She was commenced on intramuscular hydroxocobalamin along with folate supplements. On review after 2 weeks, the child showed significant clinical improvement in constipation, haemoglobin was 10.9 g/dL, and serum vitamin B_{12} levels had normalised (1604 pg/mL).

Discussion: We report a second child with Imerslund-Gräsbeck syndrome, a rare genetic cause of vitamin B12 deficiency from the same family. This case report describes a very unusual presentation of vitamin B12 deficiency as chronic constipation. It also highlights the importance of developing a family screening strategy to identify affected siblings early to prevent irreversible complications of the disease.

EXPOSURE KERATOPATHY UNVEILING FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY: A CASE REPORT

K.A.C.J.J. Rathanayake, H.G.H Udara, K.H.P. Madhushani, T.A.C. Mudalige, T.M. Munasinghe

Introduction: Facioscapulohumeral muscular dystrophy (FSHD) is a slowly progressive myopathy and the third most common muscular dystrophy. It typically presents in late childhood or adolescence, with early involvement of facial and shoulder girdle muscles. Early recognition is essential to delay complications and optimise supportive care.

Description: A 13-year-old girl initially presented to an ophthalmologist with red eyes and was diagnosed with bilateral exposure keratopathy. She was referred for neurological evaluation due to facial weakness. History revealed progressive proximal muscle weakness and difficulty in walking over 2–3 years, along with poor intellectual abilities. Examination showed facial and neck muscle weakness, ophthalmoplegia, asymmetric proximal weakness in upper and lower limbs, significant scapular winging, and scoliosis. Deep tendon reflexes were elicitable. Cranial nerves were otherwise normal, with no sensory or cerebellar involvement.

Investigations: Electromyography revealed myopathic changes in proximal muscles of upper and lower limbs. Thoracolumbar spine X-ray showed significant scoliosis. Serum creatine phosphokinase (CPK) was 110 U/L (reference <145). Echocardiography was normal. Genetic studies are awaited.

Progress: She was started on a supervised low-intensity, high-repetition exercise program with balance exercises and posture maintenance. Environmental modifications are being implemented, and ophthalmology follow-up continues.
Discussion: Facial muscle weakness is an early and subtle feature of FSHD, often first presenting as reduced facial expression. Subtle weakness may go unnoticed and result in secondary complications, as observed in this case. Early recognition and timely supportive management can delay complications and improve functional outcomes in affected children.

FANCONI ANAEMIA COMPLICATED BY MYELODYSPLASTIC SYNDROME WITH MONOSOMY 7: SUCCESSFULLY MANAGED WITH ALLOGENEIC HAEMATOPOIETIC STEM CELL TRANSPLANTATION

Jothiji V, Mayura S, De Silva N, Vidanagama D, Jayasinghe G, Vitharana S

Introduction: Fanconi anemia is an inherited bone marrow failure syndrome associated with severe aplastic anemia, myelodysplastic syndrome (MDS), and acute myeloid leukaemia. Allogenic haematopoietic stem cell transplantation (HSCT) is the only curative option for haematlogical manifestation of the disease with promising outcomes using reduced intensity conditioning regimen (RIC).

Description: We report a case of 7 years old boy with a history of tracheoesophageal fistula and tetralogy of Fallot presented with pancytopenia. Fanconi anemia was confirmed by positive chromosomal breakage studies. Bone marrow reveled MDS with monosomy 7. Given the high-risk cytogenetic abnormality and disease progression, the patient was planned for urgent HSCT. Donor was 22 year old maternal aunt with a 10/10 HLA match. He received RIC with rATG, Fludarabine and Cyclophosphamide. Peripheral blood stem cells (PBSC) were infused with a CD 34 cell dose of 10 x 106 /Kg.

His transplant was delayed due to several episodes of neutropenic sepsis. During the peri and post-transplant period, the patient developed multiple complications, including neutropenic sepsis,typhlitis Clostridium difficile infection, haemorrhagic cystitis, haematochezia and angioedema following to platelet transfusions. All complications were managed promptly with appropriate supportive and targeted therapies. The patient achieved complete donor chimerism by day +30 post-transplant and was subsequently discharged in stable condition.

Discussion: Hematopoietic stem cell transplantation can be successfully performed in children with Fanconi anemia and high-risk MDS despite significant peri-transplant complications, provided timely transplantation and comprehensive supportive care are ensured.

A CASE OF A KETOLYTIC DEFECT PRESENTING WITH RECURRENT EPISODES OF METABOLIC ACIDOSIS

K. H. P. Madhushani, H. G. H. Udara, K. A. C. J. J. Rathnayake, T. A. S. Mudalige, l. Kankananarachchi

Introduction: Defects in ketolysis; Monocarboxylate transporter 1 (MCT1) deficiency, Beta-ketothiolase deficiency and succinyl CoA:3-oxoacid CoA transferase (SCOT) deficiency, are rare inborn errors that lead to toxic ketoacidosis, when the body is under stress. They should not be overlooked in a child presenting with intractable vomiting.

Description: A 3 year and 2 months old girl, born to third degree consanguineous parents, presented with two days history of intractable vomiting, lethargy, poor feeding and excessive thirst. Her history was notable for a prior episode of severe metabolic acidosis during a bronchopneumonia one month back. Examination revealed below average growth parameters, with the child drowsy and ill, though hemodynamics remained stable.

Investigations (if any): Investigations revealed high anion gap metabolic acidosis with pH of 7.1, PCO2 18mmHg, PO2 38mmHg, HCO3- 6mmol/L and an anion gap of 28mEq/L. She exhibited euglycemic ketoacidosis with a random blood sugar of 82mg/dL alongside 4+ of urine ketones. Serum ammonia, amino acid levels, and acylcarnitine profile were normal with markedly elevated levels of 3-hydroxybutyric acid in urine suggestive of a defect in ketolysis. Further genetic studies are awaited.

Progress: During the acute phase, she was started on 0.9% NaCl and 10% dextrose with empirical antibiotics. Clinical status and metabolic markers normalized within 48 hours from treatment initiation. On discharge, advised to avoid prolonged fasting and ketodiets, provided with a ‘sick day’ plan and an emergency management protocol. No relapses have occurred to date.

Discussion: This case emphasizes that in children presenting with ketoacidosis, euglycemia, normal ammonia and normal lactate is a critical indicator of ketolysis defects, where rapid diagnosis and emergency treatment is lifesaving.

A CASE OF A KETOLYTIC DEFECT PRESENTING WITH RECURRENT EPISODES OF METABOLIC ACIDOSIS

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WHEN THE AIRWAY CANNOT BE FOUND: ANTENATAL HYDROPS DUE TO CONGENITAL HIGH AIRWAY OBSTRUCTION SYNDROME

RGDS Rankoth, Sandya Doluweera

Introduction: Congenital High Airway Obstruction Syndrome (CHAOS)is a rare fetal condition caused by complete or near-complete obstruction of the upper airway, most commonly at the level of the larynx. Early antenatal diagnosis is crucial for parental counselling and perinatal planning.

Description: We report a case of CHAOS in a male neonate born to a 30-year-old gravida 3 mother.The pregnancy was unplanned and complicated by maternal anaemia and polyhydramnios .An anomaly scan at 22 weeks of gestation revealed gross fetal ascites, enlarged fluid-filled lungs, thoracic compression, and features suggestive of upper airway obstruction, consistent with CHAOS. Following Multidisciplinary decision and counselling regarding the fatal prognosis, the parents selected the expectant management. At 27 weeks of gestation, the mother developed abdominal pain and preterm labour with maternal tachycardia, prompting emergency caesarean section. The neonate was born floppy, cyanosed, apnoeic, and bradycardic, with APGAR scores of 1 at 1, 5, and 10 minutes. Endotracheal intubation failed due to inability to visualise the laryngeal inlet, and the baby died 10 minutes after birth.

Investigations (if any):

Progress: baby succumbed shortly after birth due to an unmanageable congenital upper airway obstruction.

Discussion: CHAOS is universally fatal without intervention due to heart failure and hydrops.The Ex Utero Intrapartum Treatment (EXIT) procedures, which allow establishment of a secure airway while maintaining uteroplacental circulation, represent the only potential life-saving option reporting minimal survival rates in carefully selected fetuses without severe cormorbidties and extreme prematurity.

A SEVERE CASE OF CONGENITAL ICHTHYOSIS IN A COLLODION BABY

S.Zulaiha Zuhair, P.M.Arshath Ahamed

Introduction: Collodion babies are a rare entity with a myriad of outcomes and prognosis, relating to disorders of cornification. Congenital autosomal recessive ichthyosis (CARI) usually presents at birth, often as a collodion baby. This is a case of a possible CARI with severe ichthyosis.

Description:A baby girl, born preterm following spontaneous onset of labour at 32 weeks to healthy first time parents was noted to have a tight, shiny parchment membrane at birth. Facial distortion was present with bilateral eyelid ectropion, eclabium and fish-mouth appearance. The only risk factor was the consanguineous marriage between parents. On day 3 the membrane started peeling, revealing thick skin with rapid shedding and growth of skin, causing hypernatremic dehydration, and later hypoalbuminemia. Cannulation remained to be difficult due to the severe ichthyosis which progressed over the following days and weeks.

Investigations (if any):

Progress:The baby was managed with dermatology and ophthalmology follow up, emollients and low potency steroid application and eye care. There was good weight gain but the ichthyosis was severe. At 2 months of age, a trial of oral retinoids was started with the parents consent. However there was no significant improvement, and the baby succumbed to an event of severe skin sepsis at the age of 4 months.

Discussion:The severity and prognosis of congenital forms of ichthyosis are dependent on the time of diagnosis and the type of ichthyosis. This was a severe case with poor prognosis despite early detection and multidisciplinary management.

WHAT "NOS" REALLY MEANS: HIDDEN RARE BONE MALIGNANCIES IN PAEDIATRIC CANCER REGISTRIES

A Atchuthan, Perera DLTR, Dissanayake DMHN

Background: In cancer registries, a substantial proportion of rare malignancies are coded using non-specific histological categories such as osteosarcoma, not otherwise specified (NOS) or sarcoma, NOS. While often interpreted as limitations of registry data, the epidemiological meaning of NOS categories in rare paediatric malignancies has not been adequately examined.

Objectives: To evaluate the role of NOS histological categories in the epidemiology of paediatric malignant bone malignancies and to assess whether NOS coding reflects true diagnostic uncertainty or masks an underlying burden of ultra-rare malignant bone entities.

Methods: A population-based descriptive study was conducted using national cancer registry data from Sri Lanka (2011–2022). Children and adolescents aged 0–19 years diagnosed with malignant bone malignancies were identified and classified using ICD-O-3 morphology codes. The frequency, temporal distribution, and proportional contribution of NOS categories were analysed in relation to specified malignant histological subtypes.

Results:Across the 12-year study period, osteosarcoma NOS constituted the single largest histological category of paediatric malignant bone malignancies (n = 410). In contrast, individually specified malignant subtypes occurred at very low frequencies, including chondroblastic osteosarcoma (n = 34), central osteosarcoma (n = 13), fibroblastic osteosarcoma (n = 6), telangiectatic osteosarcoma (n = 4), periosteal osteosarcoma (n = 4), chondrosarcoma NOS (n = 9), and mesenchymal chondrosarcoma (n = 2). Over time, increasing identification of variant osteosarcoma subtypes coincided with a relative decline in NOS-coded cases, suggesting improving histological resolution rather than a change in disease biology.

Conclusions: In paediatric malignant bone malignancies, NOS categories do not simply indicate poor data quality but function as epidemiological markers of hidden biological heterogeneity. Recognition of NOS coding as a signal of unmeasured ultra-rare malignant disease burden is essential for accurate rare cancer surveillance and registry interpretation.

HEREDITARY SENSORY AND AUTONOMIC NEUROPATHY TYPE IV DUE TO NTRK1 MUTATION IN A PRESCHOOL CHILD: A CASE REPORT

Kapila Kunasingam, Mannapperuma MBN, Karunarathna WACN, Dilrangi KAR, Karunathilake KGLRAD, Weerasinghe RG, Hashan Pathiraja, Deepti de Silva, Sumanasena S

Introduction: Hereditary sensory and autonomic neuropathy type IV (HSAN IV), also known as congenital insensitivity to pain with anhidrosis, is a rare autosomal recessive disorder caused by mutations in the NTRK1 gene. It is characterised by impaired pain perception, autonomic dysfunction, and recurrent self-inflicted injuries, resulting in significant morbidity if not recognised early.

Description: We report a 4-year-old girl who presented with longstanding self-mutilating behaviour and recurrent unexplained injuries since early childhood. Clinical examination revealed multiple fresh wounds and healed scars over the limbs, tongue deformity secondary to repeated biting, and complete absence of sweating. There was a history of recurrent episodes of fever suggestive of temperature instability. The child had generalised joint hypermobility with a Beighton score of 7, associated hypotonia, and an initial global developmental delay. Although developmental progress has improved with intervention, she continues to have mild intellectual impairment.

Investigations (if any): Whole exome sequencing identified two heterozygous variants in the NTRK1 gene, one pathogenic and one likely pathogenic, confirming the diagnosis of HSAN IV.
Nerve conduction studies were normal, with no evidence of large fibre neuropathy. In contrast, sympathetic skin responses were absent in both hands and feet, consistent with small fibre autonomic dysfunction.

Progress: Management includes a multidisciplinary approach with injury prevention, wound care, temperature regulation strategies, physiotherapy, and caregiver education. Motor skills have improved, though intermittent self-injurious behaviour persists.

Discussion: This case highlights the varied phenotypic spectrum of HSAN IV and underscores the importance of early recognition and genetic confirmation to enable anticipatory guidance and long-term multidisciplinary care, particularly in resource-limited settings.

INTRADURAL EXTRAMEDULLARY SCHWANNOMA PRESENTING AS REFRACTORY LIMB PAIN IN A 15-YEAR-OLD MALE: A DIAGNOSTIC CHALLENGE

R.R.C Abeysinghe, Madarasinghe M.D, Premarathne P.B.U.S

Introduction: Spinal schwannomas are rare benign nerve sheath tumours, comprising approximately 25% of intradural extramedullary spinal tumours. While most common in middle-aged adults, paediatric cases are exceedingly rare, especially outside the context of neurofibromatosis. This case highlights an adolescent with an atypical presentation of refractory thigh pain secondary to a conus medullaris schwannoma.

Description: A 15-year-old boy presented with a 6-month history of progressive right thigh pain, severe at night and with movement, later accompanied by intermittent lumbar pain. Systemic symptoms were absent. Examination revealed a rigid gait without neurological deficits or spinal tenderness.

Investigations (if any): Routine blood tests were unremarkable. MRI spine revealed a 1.8 cm spherical extramedullary intradural lesion at T12–conus level. Histopathology post-resection confirmed a WHO Grade 1 schwannoma with Antoni A and B areas and Verocay bodies.

Progress: The tumour was completely resected. Post-operatively, the patient's pain resolved significantly. At follow-up, he was mobilising well with no symptom recurrence.

Discussion: This case illustrates that spinal schwannoma, though rare in adolescents, should be considered in the differential diagnosis of unexplained refractory limb pain, even in the absence of neurological signs. Schwannomas typically present with radicular pain that is worse at night or in the morning, as seen in our patient. MRI is the imaging modality of choice, showing well-demarcated, contrast-enhancing lesions. Complete surgical resection is curative and associated with a low recurrence rate (~5%) and good neurological recovery. In sporadic cases like this, long-term prognosis is excellent. Increased clinical suspicion can prevent diagnostic delay and reduce prolonged morbidity in young patients with atypical pain syndromes.

A CASE OF INCONTINENTIA PIGMENTI WITH CLASSICAL CUTANEOUS EVOLUTION IN AN INFANT

H.G.H. Udara, K.H.P. Madhushani, U.M. Ruwanpathirana, B.K.G.S.S. Dharmasiri, T.M. Munasinghe, B.S. Wijenayake

Introduction: Incontinentia Pigmenti is a rare X-linked dominant neuroectodermal disorder caused by mutations in the IKBKG (NEMO) gene. It predominantly affects females and is characterized by characteristic skin lesions progressing through vesiculobullous, verrucous, hyperpigmented, and hypopigmented stages, with potential multisystem involvement

Description: A 4-month-old female infant presented with a history of recurrent, non-healing skin blisters since the neonatal period, followed by progressive skin pigmentation. She had no family history of similar illness. Initial lesions appeared as erythematous vesiculobullous eruptions over the trunk and limbs, which healed poorly and gradually evolved into verrucous lesions then to linear hyperpigmented streaks distributed along Blaschko's lines. There were no associated fever, systemic illness, or seizures. Growth and development were age appropriate. On examination, multiple hyperpigmented whorled patches with residual vesicular and verrucous lesions were noted over the trunk and extremities, and few small patches of alopecia. Nails and oral mucosa were normal. Ophthalmological evaluation showed no retinal abnormalities

Investigations: A skin biopsy from an active lesion demonstrated epidermal spongiosis, hyperkeratosis, acanthosis, and a dermal inflammatory infiltrate rich in eosinophils, consistent with Incontinentia Pigmenti. Genetic testing was not performed due to limited resources.

Progress: Her cutaneous manifestations were gradually evolving but did not show any systemic involvement.

Discussion: This case highlights the importance of recognizing the characteristic evolving cutaneous stages of Incontinentia Pigmenti in infants presenting with recurrent blisters and pigmentation. Early diagnosis enables appropriate parental counselling and long-term surveillance for potential neurological and ocular complications.

SPINAL MUSCULAR ATROPHY TYPE 1: A CASE HIGHLIGHTING THE DIAGNOSTIC ODYSSEY, GENETIC INSIGHTS, AND THE CRITICAL NEED FOR NEWBORN SCREENING IN RESOURCE-LIMITED SETTINGS

Abeysinghe R.R.C, Madarasinghe M.D, Premarathna P.B.U.S

Introduction: Spinal muscular atrophy (SMA) is one of the most common autosomal recessive disorders, affecting ~1 in 10,000 births, with a carrier frequency of ~1 in 50. SMA type 1 (Werdnig-Hoffmann) presents before six months with severe hypotonia and progressive respiratory failure. Approximately 94% of cases are due to homozygous deletion of SMN1 exon 7.

Description: A 40-day-old term male, born via emergency LSCS, required resuscitation, presented with respiratory distress, weak cough, and lethargy. Examination revealed profound hypotonia, areflexia, bell-shaped chest, paradoxical breathing, and frog-leg posture.

Investigations (if any): Electromyography indicated anterior horn cell pathology. Genetic testing confirmed a homozygous deletion of exons 7 and 8 in the SMN1 gene, diagnostic of SMA.

Progress: Despite multidisciplinary supportive care, died from respiratory failure on day 33 of life, reflecting the natural history of untreated SMA type 1.

Discussion: This case underscores the rapid progression of SMA type 1, a key differential in the floppy infant syndrome. The diagnostic delay here highlights a critical gap. Newborn screening (NBS) for SMN1 deletions, now recommended for the uniform screening panel, allows presymptomatic identification. For infants with two SMN2 copies (like this case), immediate treatment with disease- modifying therapies (nusinersen, onasemnogene abeparvovec, risdiplam) is recommended to dramatically alter outcomes. Implementing NBS in regions like Sri Lanka is essential to enable timely intervention and prevent such tragic outcomes.

THINKING BEYOND INTELLIGENCE QUOTIENT, FOR COGNITIVE AND VISUOMOTOR REHABILITATION IN FRAGILE X SYNDROME

Ariyapala P P N N, Vipulaguna D, Jayawickrama R D P, Atapattu K T G, Madushani L

Introduction: Fragile X syndrome (FXS) is a genetic cause of intellectual impairment. Supporting these children needs detailed assessments, particularly focusing on cognition and visuo-motor skills.

Description: The present case report describes the comprehensive neurocognitive profile of a 15- year adolescent boy with Fragile X syndrome. The general intellect was impaired, with an IQ standard score of 65. He had significant difficulty with executive functioning as evidenced by poor cognitive planning ability and working memory. Visual construction ability, fine motor ability and visual memory were also impaired. It should be noted that the adaptive skills of the participant were in below average range but not impaired.

Investigations (if any): Genetic testing confirmed Fragile X syndrome. Berry Spot test is negative. Audiological and ophthalmological assessments were within normal limits, confirming intact hearing and vision.

Progress: A multidisciplinary treatment plan was developed combining cognitive therapy interventions to strengthen cognitive deficits and accommodate significantly impaired abilities. Cognitive rehabilitation techniques were introduced to improve working memory and visual memory. Occupational therapy targeted fine motor abilities and visual-construction ability. Emphasis was placed on building on the verbal abilities of the adolescent to support education and vocational training. The family was counseled on possible future vocational pathways for the child.

Discussion: This case reports highlight the importance of moving beyond general intellect tests and assessing other cognitive domains to identify specific strengths and weaknesses.

“BEYOND THE TEXTBOOK: EARLY-ONSET INFANTILE HYPERTROPHIC PYLORIC STENOSIS IN A SRI LANKAN FEMALE NEONATE”

Piumi Madawala, Piumi Madawala, Anura Jayawardhana

Introduction:
Infantile hypertrophic pyloric stenosis (IHPS) classically presents with gradual onset of non-bilious projectile vomiting around third week of life in the first-born males. It is rare in Asians. Here, we report a 5-day old Sri Lankan female baby; presented with non-bilious non-projectile vomiting due to IHPS.

Description:
A 5-day old female baby, who was born at term with birth weight of 2560g, discharged 48 hours after birth following normal neonatal examination and establishment of breast feeding, presented on fifth day of life with frequent non-bilious, non-projectile vomiting soon after feeds. She had 13% weight loss with some dehydration and mild jaundice on examination. The classic olive shaped pyloric mass was palpated in the mid epigastrium after an episode of vomiting.

Investigations (if any): Progress of gastric peristaltic wave across the abdomen was demonstrated following a test feed. Blood gas revealed hypochloraemic, hyponatraemic and hypokalaemic metabolic alkalosis. pH – 7.51, HCO3 – 36, Cl – 90, Na – 130, K – 3.1. Additionally, hyperbilirubinemia was noted on biochemistry. Serum Indirect bilirubin was 204 mmol/L. X ray abdomen showed distended gastric air bubble. USS confirmed the diagnosis; pyloric thickness was 4mm.

Progress: After initial stabilisation of hydration and electrolyte abnormalities, baby underwent successful pyloromyotomy on day 7 of life.

Discussion: This case highlights the need for a higher index of suspicion for IHPS despite the absence of “typical risk factors”. Our objective was to emphasize the need to consider pyloric stenosis as a possible differential when encountering infants with vomiting in the first week of life.

WHEN THE SCALP TELLS A STORY: ADAMS-OLIVER SYNDROME

Surangani SGY, Dharmasiri BKGSS, Jayasanka KTR, Kumuduni DGK, Withanaarachchi K

Introduction: Adams- Oliver syndrome (AOS) is a rare multiple congenital anomaly syndrome with variable genetic heterogeneity, characterised by aplasia cutis congenita of the scalp and terminal transverse limb defects. The degree of organ involvement and early recognition is important for appropriate multidisciplinary management and will predict the prognosis.

Description: We report a newborn baby boy delivered at 37 weeks of gestation by normal vaginal delivery to fourth-degree consanguineous parents with a birth weight of 2255g and not required resuscitation at birth. Pregnancy was complicated with maternal chronic hypertension superimposed with preeclampsia. No family history of similar condition. Physical examination revealed a large scalp and skull defect over the vertex consistent with aplasia cutis congenita, hypoplastic terminal limbs, and cutis marmorata telangiectasia congenita, suggesting AOS.

Investigations (if any): Initial laboratory evaluation showed coagulopathy and hepatic involvement which improved later. 3D Computed Tomography skull revealed a large bony defect in the vertex of the skull vault. The echocardiography revealed biventricular hypertrophy. The ultrasonography of abdomen was normal.

Progress: The neonate is managed in a multidisciplinary approach. Intravenous antibiotics was started for presumed sepsis and supportive care. Enteral feeds were tolerated well. Scalp lesions are managed with regular wound care and awaiting flap repair by the plastic and neurosurgical teams. Limb anomalies did not require urgent surgical intervention.

Discussion: AOS is diagnosed according to clinical criteria which are fulfilled in this baby. Prenatal diagnosis may improve outcomes and facilitate parental counselling.

CLINICAL DIAGNOSIS OF RUBINSTEIN–TAYBI SYNDROME WITH DEVELOPMENTAL DELAY: IMPORTANCE OF MULTIDISCIPLINARY MANAGEMENT IN A 6-YEAR-OLD BOY: A CASE REPORT

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Introduction: Rubinstein–Taybi syndrome (RSTS) is a rare neurodevelopmental disorder characterised by distinctive craniofacial features, broad thumbs and toes, and cognitive impairment. Early clinical recognition is critical to initiate multidisciplinary care and to anticipate potentially serious complications, particularly in settings where access to genetic testing is limited.

Description: A six-year-old boy was evaluated for short stature, moderate learning disability, and global developmental delay. He demonstrated characteristic dysmorphic features, including typical facial appearance, narrow palate, beaked nose, microcephaly, bilateral exotropia and broad thumbs and toes. A significant perioperative event of apnea and bradycardia occurred following general anaesthesia. He is the first child of non-consanguineous parents, with no family history of congenital or neurodevelopmental disorders.

Investigations (if any): Genetic confirmation was not available due to financial constraints. Routine laboratory and imaging studies were unremarkable. The diagnosis of Rubinstein–Taybi syndrome was established clinically based on classical phenotypic features and developmental profile.

Progress: A coordinated multidisciplinary management plan was implemented involving community paediatrics, speech and language therapy, physiotherapy, occupational therapy, and ophthalmology. With regular follow-up, the child has demonstrated meaningful developmental gains, most notably in expressive and receptive language skills.

Discussion: This case emphasises the pivotal role of clinical diagnosis in identifying rare syndromes in resource-limited settings. Recognition of RSTS enables anticipatory guidance, targeted developmental interventions, and heightened awareness of anaesthetic risks. Structured multidisciplinary care can significantly improve functional outcomes, underscoring the value of early community-based intervention.

PHACE SYNDROME UNMASKED BY GIANT FACIAL HAEMANGIOMA IN A NEONATE

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Introduction: PHACE syndrome is a rare neurocutaneous disorder defined by Posterior fossa malformations, large segmental Haemangiomas, Arterial anomalies, Cardiac defects, Eye abnormalities, and Sternal clefts. It is most commonly associated with large cervico-facial infantile haemangiomas. Early recognition is critical due to the risk of serious neurological, cardiovascular, and visual complications, and because prompt intervention may improve outcomes.

Description: A 20-day-old neonate, born at 33 weeks of gestation to non-consanguineous parents, presented with progressively enlarging facial haemangiomas. The lesion was first noted on day 4 of life and progressed to involve the right trigeminal nerve distribution (V1–V3), including the palate, with associated lip ulceration, and additional left-sided V1 involvement. There were no seizures, bleeding, or respiratory compromise. On examination, he had large anterior and posterior fontanelles, and normal general movements. There were no sternal clefts. Ophthalmological evaluation revealed no retinal anomalies.

Investigations: Neuroimaging with ultrasonography demonstrated posterior fossa abnormalities consistent with a Dandy–Walker malformation. Echocardiography did not show any coarctation of the aorta but a small ostium secundum Atrial septal defect. MRI and MRA of Brain are awaited to assess associated arterial anomalies.

Progress: The haemangiomas have progressed with worsening lip ulceration and feeding difficulty. Oral steroid was commenced, and the infant is under close multidisciplinary follow-up.

Discussion: This case highlights the importance of early recognition of PHACE syndrome in neonates with large facial haemangiomas and underscores the need for systematic evaluation and timely multidisciplinary management.

KLIPPEL-TRENAUNAY SYNDROME

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Introduction: Klippel-Trenaunay syndrome (KTS) is a rare congenital disorder that cause abnormal development of capillary, venous and lymphatic systems. It is usually due to sporadic mosaic mutation of PIK3CA gene and is one of the PIK3CA related overgrowth spectrum disorders.

Description: A 6-months-old baby girl, born to non-consanguineous parents presented with progressively enlarging swelling of left gluteal region associated with intermittent fever. The overlying skin had bluish discolouration and was warm and tender. The mass was soft in consistency, ill-defined, partially compressible, pseudo-fluctuant and non-pulsatile. There was associated hemihypertrophy of the left lower limb.

Investigations (if any): Magnetic resonance imaging of the region revealed arteriovenous malformations involving superior and inferior gluteal veins confirming the diagnosis of KTS in correlation with clinical findings.

Progress: Child recurrently presented with worsening swelling and secondary infections leading to significant impairment in gross motor development, including inability to sit and is currently awaiting surgical intervention.

Discussion: Klippel-Trenaunay syndrome is a rare complex malformation disorder characterized by capillary malformations, varicosities and soft tissue and bony hypertrophy. These findings are most often unilateral and affects limbs. The presence of two out of three signs is enough to obtain the diagnosis. The severity of the vascular distortion and the degree of overgrowth segment have a relevant prognostic effect. Usually, it has a benign course but may cause life threatening complications. Diagnosis is primarily clinical, supported by Doppler ultrasound and MRI to define vascular involvement. Management is multidisciplinary, focusing on symptom control, compression therapy, and selective interventional procedures.

CONGENITAL SYPHILIS IN AN ERA OF ELIMINATION

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Introduction: Congenital syphilis is a vertically transmitted infection caused by Treponema pallidum, with a high transmission risk resulting in fetal loss, perinatal mortality, and congenital infection.

Description: A term male neonate was delivered vaginally to a mother with no antenatal care. The baby had normal birth parameters and no clinical features of congenital syphilis. On day one of life, he was started on intravenous crystalline penicillin for suspected neonatal sepsis. Five days postpartum, the mother was diagnosed with early latent syphilis and treated with intramuscular benzathine penicillin.

Investigations (If any): The neonate's serum VDRL titre on day 5 of life was 1:32, identical to the maternal titre measured on the same day. Cerebrospinal fluid analysis (CSF) revealed reactive VDRL with mild pleocytosis, elevated proteins, and normal glucose levels. Full blood counts, inflammatory markers, liver and renal functions, and cultures were unremarkable. Neuroimaging, long bone radiographs, hearing and ophthalmological assessments were normal.

Progress: The infant was treated for presumptive congenital syphilis with intravenous crystalline penicillin. Serological and clinical follow-up, including VDRL, is planned.

Discussion: In an asymptomatic neonate, a fourfold or higher VDRL titre compared with maternal titre supports the diagnosis of congenital syphilis. In this case, neonatal titres were likely influenced by early antibiotic exposure. A Reactive CSF VDRL is significantly specific for neurosyphilis. This case highlights the importance of comprehensive neonatal evaluation and treatment when maternal syphilis is untreated. Despite the elimination of mother-to-child transmission of syphilis in Sri Lanka, sporadic cases continue to occur, emphasising the need for ongoing vigilance and early maternal screening.

NEONATAL CHOLESTASIS AS A SENTINEL MANIFESTATION OF NIEMANN PICK TYPE C

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Introduction: Niemann-Pick type C (NPC) is an autosomal recessive lysosomal storage disorder caused by mutations in the NPC1 or NPC2 genes, leading to lysosomal accumulation of cholesterol and glycosphingolipids. The disease exhibits marked phenotypic heterogeneity, with age-dependent presentations ranging from severe neonatal liver disease to progressive neurological deterioration in childhood.

Description: A two-month-old male infant, born to third-degree consanguineous parents, presented with cholestatic jaundice from the second week of life. He was deeply icteric, affecting all mucocutaneous membranes, without features of obstructive jaundice. There were no signs of congenital infections, dysmorphism, or peripheral stigmata of chronic liver disease. Family history was significant for a sibling death at four months from suspected metabolic liver disease which was later genetically confirmed to be of Niemann pick type C. The infant had marked hepatosplenomegaly but normal growth, development, and neurological examination.

Investigations Laboratory tests showed normal blood counts and liver synthetic function, elevated bilirubin, AST/ALT (10× normal), high triglycerides and cholesterol, and normal renal function and venous blood gas. Ultrasound excluded biliary atresia, and alpha-fetoprotein was markedly elevated.

Progress
Close surveillance for liver failure and early neurodevelopmental therapy initiated

Discussion:
NPC is slowly progressive, with initial visceral involvement that may be transient, eventually leading to neurological impairments such as hypotonia and developmental delay. There is no curative treatment; management focuses on supportive care through a multidisciplinary team, including hepatology, neurology, nutrition, and physiotherapy, to optimize outcomes and quality of life. If progress to liver failure , liver transplantation maybe indicated.

PROLONGED PYREXIA REVEALING CYSTIC INTERSTITIAL LUNG DISEASE IN AN INFANT WITH TRISOMY 21

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Introduction: Prolonged pyrexia in infancy poses a significant diagnostic challenge, particularly in children with Trisomy 21 who are predisposed to infections, immune dysfunction and atypical respiratory disorders. While infection is often the initial consideration, persistent fever despite appropriate antimicrobial therapy should prompt evaluation for non-infectious causes. Interstitial lung disease (ILD) in infancy is rare and often diagnosed after exclusion of other etiologies.
Description:.A 12-month-old male infant with Trisomy 21 due to non-disjunction presented with intermittent high-grade fever for six weeks. He had associated prolonged non productive cough,persistent tachypnoea,hypoxemia with prolonged oxygen dependancy.He had received multiple courses of intravenous antibiotics, including ceftriaxone, vancomycin and meropenem, without sustained clinical improvement. On admission, he remained febrile and tachypnoeic with oxygen saturation of 88-92% on room air requiring supplemental oxygen. No focal infective source was identified on examination.

Investigations (if any):Laboratory evaluation showed neutrophilic leukocytosis with normal inflammatory markers. Blood and urine cultures were repeatedly sterile. Tuberculosis screening was negative.Chest radiograph demonstrated bilateral perihilar haziness without focal consolidation. Echocardiography showed normal cardiac anatomy. High-resolution CT of the chest revealed bilateral cystic interstitial changes consistent with cystic interstitial lung disease.Bronchoscopy was unremarkable and bronchoalveolar lavage did not identify infectious organisms.Comprehensive immune deficiency workup was normal.Infectious etilogies were extensively excluded.Lung biopsy and genetic testing could not be performed due to limited fascilities.

Progress:The child was managed with supportive care including oxygen therapy, chest physiotherapy and nutritional optimisation. Following exclusion of infectious causes, empirical corticosteroid therapy was considered in consultation with a multidisciplinary team.

Discussion:In this case persistent fever and respiratory symptoms were not attributable to infection despite extensive evaluation.Based on clinical features,imaging findings and exclusion of other possible causes ,a diagnosis of cystic interstitial lung disease was made.Early consideration of ILD in similar presentations and early use of HRCT and a multidisciplinary approach are essential to avoid diagnostic delay and guide appropriate management.

NEONATAL PRESENTATION OF OSTEOGENESIS IMPERFECTA: DIAGNOSTIC CHALLENGES OF NEONATAL OI IN THE ABSENCE OF CLASSIC FEATURES IN A RESOURCE-LIMITED SETTING.

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Introduction: Osteogenesis imperfecta (OI) is an inherited connective tissue disorder characterized by bone fragility due to defective type I collagen. Its incidence is approximately 1 in 20,000 live births. Severe forms may present in the neonatal period (type II and III) with fractures and skeletal deformities and are associated with significant morbidity and mortality. We report a neonate diagnosed with OI at 28 days of life.

Description: A 28-day-old term male infant, born to non-consanguineous parents, presented with sudden-onset inconsolable crying while asleep. Examination revealed swelling of the left thigh without external signs of trauma. Blue sclerae were absent. There was no history suggestive of non-accidental injury. Antenatal and perinatal histories were unremarkable. There was no family history of fractures or connective tissue disorders.

Investigations: Radiography demonstrated a mid-shaft fracture of the left femur, bowing of long bones, cortical thinning, and generalized osteopenia, suggestive of OI. Serum calcium, phosphate, and alkaline phosphatase levels were normal. DEXA scan is planned to be done at a later age. Genetic testing was not performed due to financial constraints. The clinical and radiologic features were most consistent with OI type III.

Progress: The fracture was managed with immobilization using gallows traction. Intravenous bisphosphonate therapy with calcium and vitamin D supplementation was initiated. Physiotherapy, rehabilitation, and family counselling were provided. The infant showed satisfactory fracture healing and improved mobility.

Discussion: Neonatal OI should be suspected in neonates with limb bowing or fractures after minimal trauma, even without blue sclerae or family history. Unlike non-accidental injury, fractures occur in the setting of generalized osteopenia. Diagnosis is particularly challenging in low-resource settings due to limited imaging and genetic testing. Early recognition, multidisciplinary management, and initiation of bisphosphonates at 2–6 weeks of life can improve outcomes.

PAEDIATRIC MALIGNANT BONE TUMOURS AS ORPHAN CANCERS: EPIDEMIOLOGICAL EVIDENCE FROM SRI LANKA (2011–2022)

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Background: Paediatric malignant bone tumours are clinically aggressive cancers associated with substantial morbidity and mortality. Despite this, they occur at very low population frequencies and remain under-represented in clinical trials and therapeutic development. Whether paediatric malignant bone tumours fulfil formal criteria for classification as orphan cancers at the population level has not been well explored in low- and middle-income countries.

Objectives: To evaluate the epidemiological characteristics of paediatric malignant bone tumours in Sri Lanka from 2011 to 2022 and to assess whether these tumours meet criteria for orphan cancer designation based on incidence, histological heterogeneity, and research neglect.

Methods: A population-based descriptive study was conducted using national cancer registry data. Children and adolescents aged 0–19 years diagnosed with malignant bone tumours were identified. Primary analyses were histology-based using ICD-O-3 codes, with supplementary site-based classification using ICD-10 C40–C41. Age-specific, sex-specific, and temporal patterns were examined.

Results: Across the 12-year study period, paediatric malignant bone tumours demonstrated consistently low incidence, with annual case numbers well below thresholds typically used to define rare diseases. Osteosarcoma and Ewing sarcoma accounted for the majority of cases, while numerous malignant histological subtypes—including telangiectatic, periosteal, and chondroblastic osteosarcoma, chondrosarcoma variants, and other rare malignant entities—occurred sporadically. A pronounced adolescent peak (10–19 years) and male predominance were observed. Many histological subtypes individually met ultra-rare disease criteria.

Conclusions: Paediatric malignant bone tumours in Sri Lanka fulfil key epidemiological characteristics of orphan cancers: very low incidence, marked histological heterogeneity, and limited therapeutic innovation. Recognition of paediatric malignant bone tumours as orphan cancers is essential to inform research prioritisation, policy development, and international collaboration.

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