eBarani Peds Magazine

OFFICIAL QUARTERLY MAGAZINE OF

Indian Academy of Pediatrics

Tirunelveli - Tuticorin - Tenkasi Branch

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Editor in Chief Message

Dear IAPans,

Greetings from TTT Chapter of TNSB,

It's indeed a great moment for IAP TNSB and IAP - TTT Chapter in particular to start the e-journal of IAP-TTT Chapter in the name of e Barani Peds. It gives me great pleasure to be the Editor of this e-journal.

For the past 25 years, we are having "Difficult case Discussion Forum" - local IAP Forum for the 3 southern districts of our state. The cases presented in this forum are very rare of high quality. This particular e-journal will take these case discussions to other chapters of IAP - TNSB and other states.

I'm sure all the practicing Pediatricians in the State, the young Pediatricians in particular will be greatly benefitted.

I have to appreciate the great efforts of the President of TTT Chapter Dr.V.Arunachalam and the Secretary of the chapter Dr.T.R.Ajay Prakaash and all the Pediatricians in the Editorial Board who put in lot of work to bring out this e-journal in good shape.

I'm sure this great initiation by our chapter will continue and will be taken as a good precedence by other chapters as well.

Dr.S.Raju.,MD

President IAP – TNSC Message

Dear President Dr Arunachalam

I am very happy and glad that IAP TTT Branch is going to release the e Barani Peds the official magazine of your branch.Its a excellent initiative taken by dynamic team Dr Ajay Prakaash, Dr Manickavasagam other office bearers and ably guided by senior leaders Dr Thirumalai Kolundu, Dr Raju and all. A newsletter is a platform where members can share academic knowledge and expertise. It also helps members to encourage one another and to develop talents to improve their clinical practise. I hope that this is one of few newletters in IAP and will definitely boost the image of your branch at the state and national level.

I wish the magazine to achieve the goal of strengthening the organisation and individual excellence in their future.

Best wishes to all

Dr K U Suresh Balan

President

IAP TNSC

President IAP – TTT Message

Dear Members,

In the ever-evolving field of pediatric medicine, staying updated with the latest advancements and best practices is of utmost importance. Our E BaraniPeds is committed to providing you with valuable insights, expert opinions, and practical tips from the pediatric community.

In this edition, we have covered a wide range of articles from various Paediatric institutions. Our dedicated team of editors and professionals has worked tirelessly to bring you comprehensive pieces on rare case discussions, and financial articles for doctors.

This year our branch hosts State conference Tamira Pedicon. Warm invitation to you for the upcoming Tamira Pedicon. Expecting support, cooperation and 100% registration from our TTT branch.

To confirm your participation and access more details, please visit our conference website

Warm regards

Dr V Arunachalam

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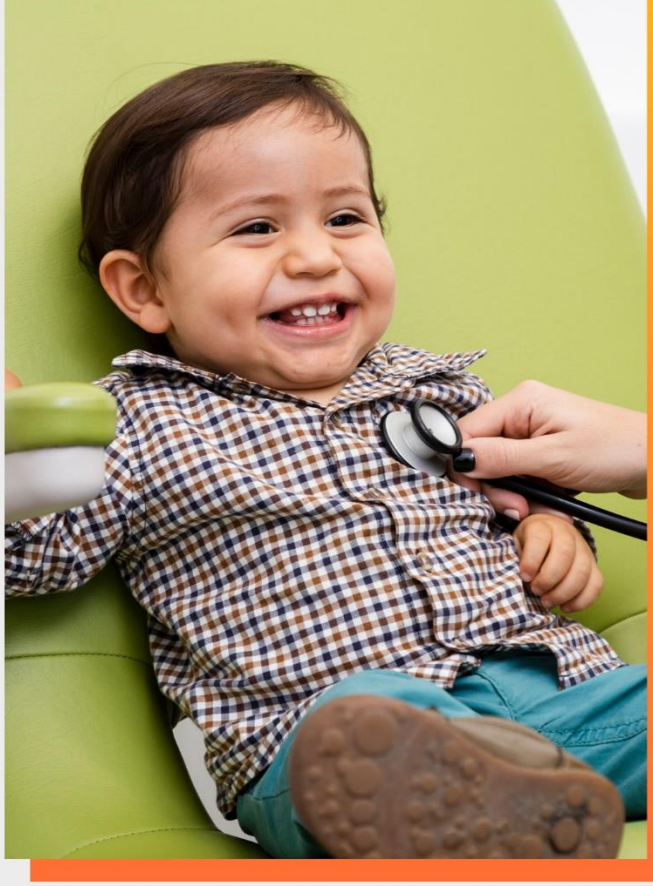

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An interesting case of DSD

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Abstract

5 alpha – reductase 2 deficiency is an autosomal recessive condition caused by a mutation in SRD5A2 gene, located on the short arm of chromosome 2, a gene encoding the enzyme 5 alpha – reductase type2. Patients present with ambiguity of the genitalia, with a clitoral like phallus, bifid scrotum, pseudo vaginal perineoscrotal hypospadias and a rudimentary prostate. In the under virilized male, there can be any degree of sexual ambiguity. Thus phenotypes range from predominantly male genotype to complete female phenotype, through varying degrees of ambiguous genitalia. A detailed physical examination, imaging investigations, karyotyping, laboratory investigations and genetic analysis are useful for diagnosis. This newborn is presented for the rarity of the presentation and the dilemma in the Gender Identity and Sex-of-rearing.

Introduction:

5 alpha- Reductase 2 deficiency condition is rare, affects only genetic males, and has a broad spectrum. 5 alpha-Reductase 2 is expressed in specific tissues and catalyses the transformation of Testosterone to DHT. Decreased production of DHT in utero results in marked ambiguity of external genitalia of affected males. Biosynthesis and peripheral action of testosterone are normal. Phenotype most commonly associated with this condition results in males who have a small phallus, bifid scrotum, urogenital sinus with perineal hypospadias and a blind vaginal pouch. Testes are in the inguinal canals or labioscrotal folds. Mullerian structures are absent; Wolffian structures are present. It is important to distinguish this from PAIS. Detailed physical examination, imaging to make an anatomical description of the external and internal genitalia, karyotyping and lab investigations are useful for diagnosis.

Case report:

We report a case of newborn born of LSCS at a tertiary care hospital with DSD. No significant family history and antenatal history.

Examination:

Testes was palpable on both sides of labia majora, stretched phallus length: 1.2 cm. No genital hyperpigmentation noted. Anogenital ratio >0.5. systemic examination was normal. (fig 1,2)

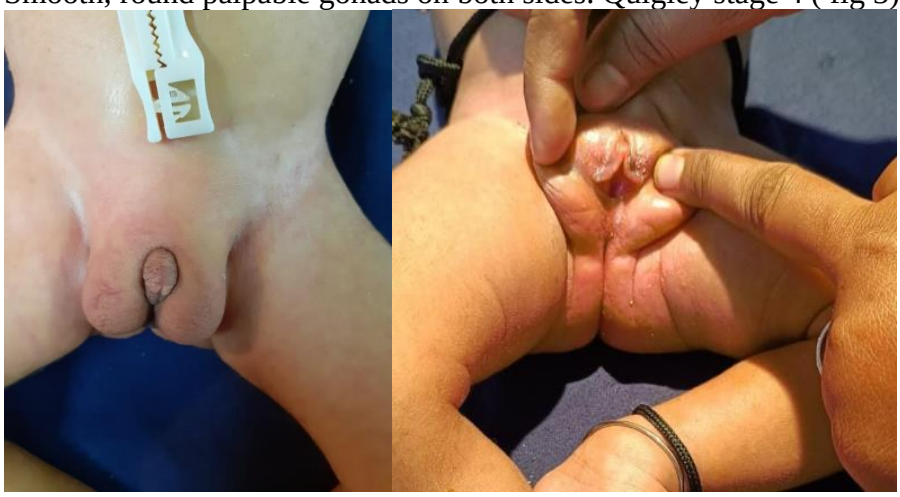
External genitalia:

Anogenital ratio >0.5

Number of genital openings: 2 [perineal urethral and anal opening]

Phallus length:1.2 cm

Smooth, round palpable gonads on both sides. Quigley stage 4 (fig 3)



Quigley staging

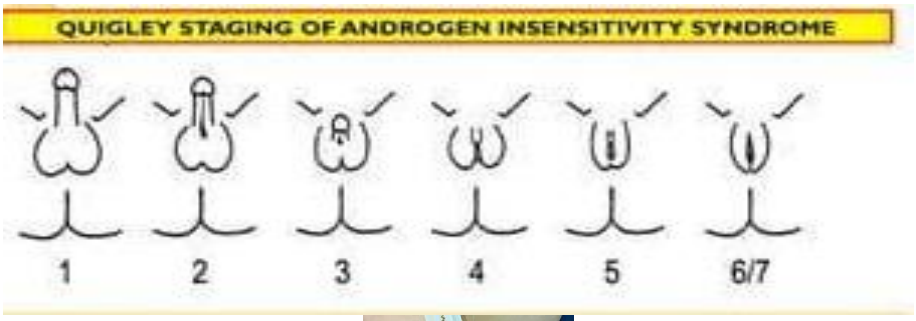
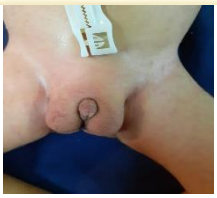


Fig 3

QUIGLEY STAGE -4



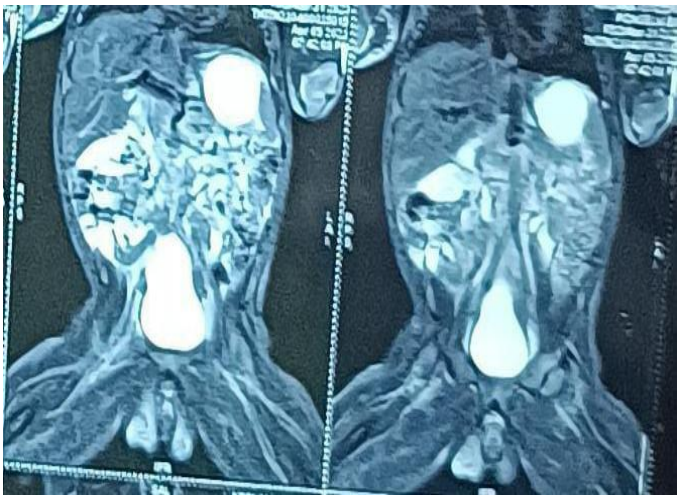
Investigations:

Table 1

PARAMETERS	PATIENT'S VALUE	REFERENCE RANGE	INTERPRETATION
Serum Na+	141 mEq/dl	135-145 mEq/dl	Normal
Serum K+	4.4mEq/dl	3.5-4.5mEq/dl	Normal
17 Hydroxy Progesterone	4.39 ng/ml	<6.3 ng/ml	Normal
Karyotype	**46XY**		**Male**
Total Serum Testosterone	88.12 ng/dl	20-64 ng/dl	**Increased**
5 alpha DHT	257.67 pg/ml	<1200 pg/ml	**Normal**
LH	2.63	0.1-4 IU/L	Normal
AMH	>24.5 ng/ml		**Increased**
T/DHT	3.4		

Imaging:

- USG- Oval testis like structure 8*4cm in both thickened labia majora
- MRI- Ambiguous external genitalia with normal appearing testis and prostate; absent mullerian structures. (fig 4)



Genetic analysis:

Whole exome gene sequencing: Homozygote for a likely pathogenic variant in the SRD5A2 gene associated with pseudo vaginal Perineo-scrotal hypospadias. (fig 5)

The index patient is:

Homozygote for a Likely Pathogenic variant in the *SRD5A2* gene associated with PSEUDOVAGINAL PERINEOSCROTALHYPOSPADIAS; PPSH.

Carrier Status:

No Pathogenic or Likely Pathogenic variants were detected in the Carrier gene list.

Secondary Findings (AGMG gene list):

No Pathogenic or Likely Pathogenic (Class 1/2) variants were detected in the ACMG gene list.

Management:

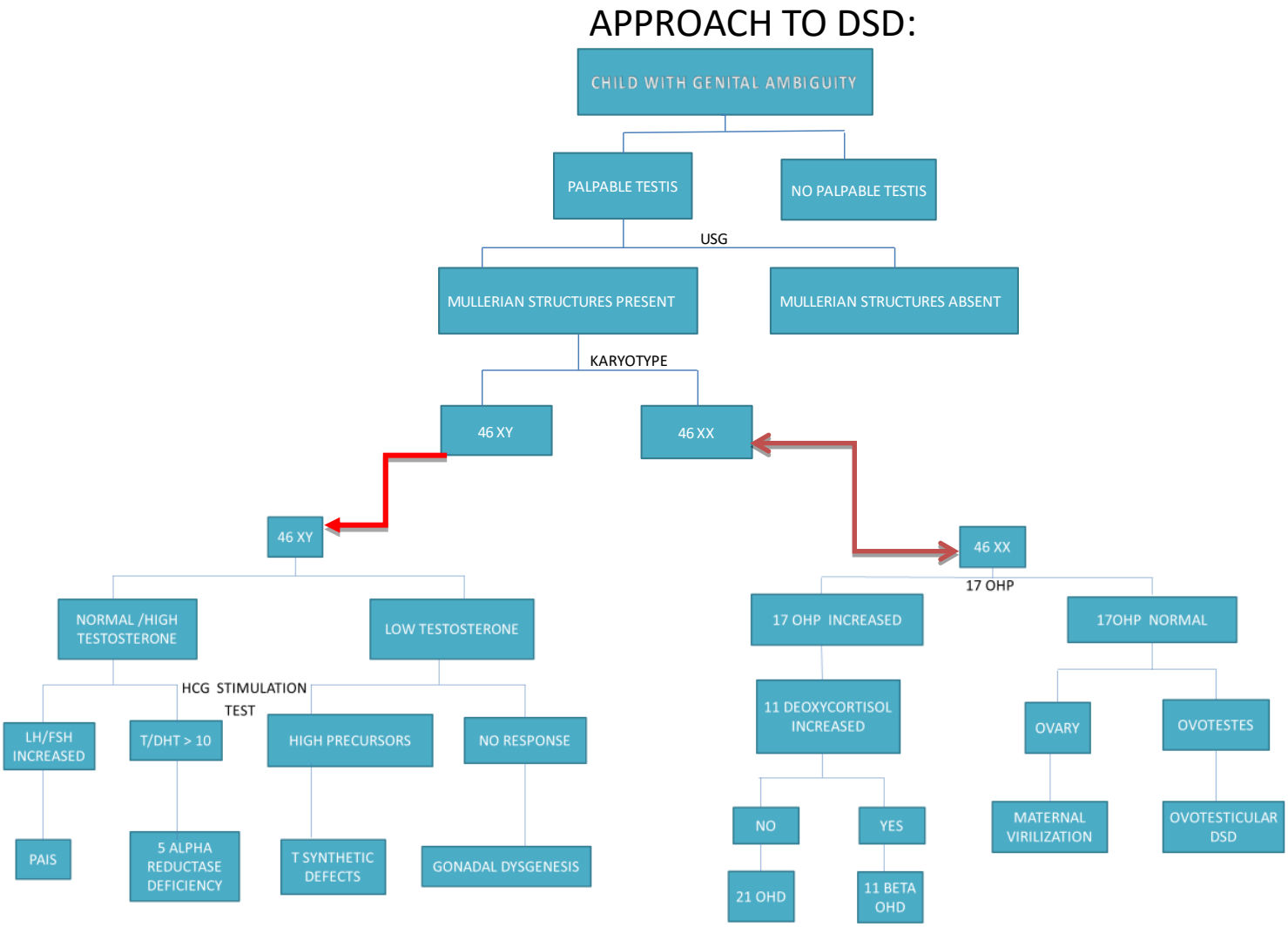
In this case, the newborn had wolffian structures ; mullerian structures were absent. Confirmed by imaging investigations. Karyotyping also done. After making all biological assessments (table 1,) , genetic analysis was done to confirm the diagnosis. Discussion about the test results was told to the parents and decision to rear as male or female was given to the parents. Surgical and medical management was discussed with the parents. Infants with this condition should be reared as males whenever practical(table 2). Treatment of male infants with DHT results in phallic enlargement.

Type of DSD	Gender assignment
45XO Turner's syndrome	Female
47 XXY	Male
46 XX DSD, congenital adrenal hyperplasia	Female
46 XY DSD	
5 alpha reductase deficiency	Male
17 beta HSD deficiency	Male/female
Complete gonadal dysgenesis	Female
Partial gonadal dysgenesis	Male/female
Complete androgen insensitivity syndrome	Female
Leydig cell hypoplasia (LH receptor defect)	Female
Ovotesticular DSD	Male/female
Partial androgen insensitivity syndrome	Male/female
Hypospadias	Male
Hypopituitarism/ Hypogonadotropic hypogonadism	Male
Isolated micropenis	Male
Cloacal exstrophy	Male/female

Table 2

Conclusion:

Any undervirilized male newborn, it is mandatory to make an anatomical description of the external and internal genitalia as completely as possible [pictures, imaging]. Second step is to investigate the quality of testicular synthesis [from Leydig and Sertoli cells] and equally important to evaluate the action of testosterone on its target organs , external genitalia in particular. The undervirilized male presents a complex and difficult problem for the clinician. Decision to rear an undervirilized neonate as male or female depends on several factors (fig 6). There is no perfect choice of sex of rearing and the most reasonable outcome should be considered.



Bibliography

- Nelson textbook of paediatrics 21st edition, chapter 606, Disorders of sex Development
- Nicolino M, Bendelac N, Jay N, Forest MG, David M. Clinical and biological assessments of the undervirilized male. BJU Int. 2004 May;93 Suppl 3:20-5. doi: 10.1111/j.1464-410X.2004.04705.x. PMID: 15086438.

STRIDOR: NOT JUST CROUP

Krishna Maternity Home & Pediatric centre

ABSTRACT:

Stridor is a variable, high-pitched sound resulting from turbulent airflow due to partial airway obstruction. Inspiratory stridor generally results from obstruction above the glottis.Stridor can be acute or chronic. Acute causes can be infectious or non infectious. Chronic causes could be within (intrinsic) or outside airway (extrinsic) .Overall, the most common cause of stridor in pediatric population is foreign body aspiration .Among the infectious causes most common is croup. This article emphasises the importance of considering alternative diagnoses other than croup when children presenting stridor does not respond to conventional therapy.

CASE REPORTS

CASE 1 :

A 11-month-old, previously well female child presented with history of high grade fever for 1 day followed by sudden onset of difficulty in breathing. There was no history suggestive of foreign body aspiration. On admission, child was lethargic, febrile (103F), tachycardic (heart rate 180–190/min), borderline saturations (Spo2 85%–89% in room air), suprasternal retractions and severe inspiratory stridor. Considering possibility of croup, she was started on epinephrine nebulizations and one dose intravenous steroid(dexamethasone) was administered. As child continued to be hypoxemic, she was transferred to PICU and started on high flow nasal cannula oxygen along with empirical antibiotic (ceftriaxone).Her saturations improved, however stridor and work of breathing continued to worsen in spite of the above measures.Chest X-ray and X-ray neck lateral view was also noncontributory. Blood investigations (table 1) revealed elevated inflammatory markers with polymorphonuclear leukocytosis. Investigations were suggestive of possible bacterial etiology, probably bacterial tracheitis.

In view of worsening respiratory status, endotracheal intubation in operating room along with bronchoscopy under anesthesia was planned.

Table 1: Laboratory investigations

Investigation	Values
CBC	Hb - 9.0 g/dl TLC - 20,440 (neutrophils - 75%, lymphocytes - 22%, monocytes - 3%) Platelet - 5.06 lakhs/cu mm
CRP	65 mg/L
Blood culture	Sterile
Tracheal aspirate culture	Moraxella catarrhalis

Rigid bronchoscopy was done which showed thick purulent fibrinous strands in the supra-glottic area, causing partial airway obstruction with extensive

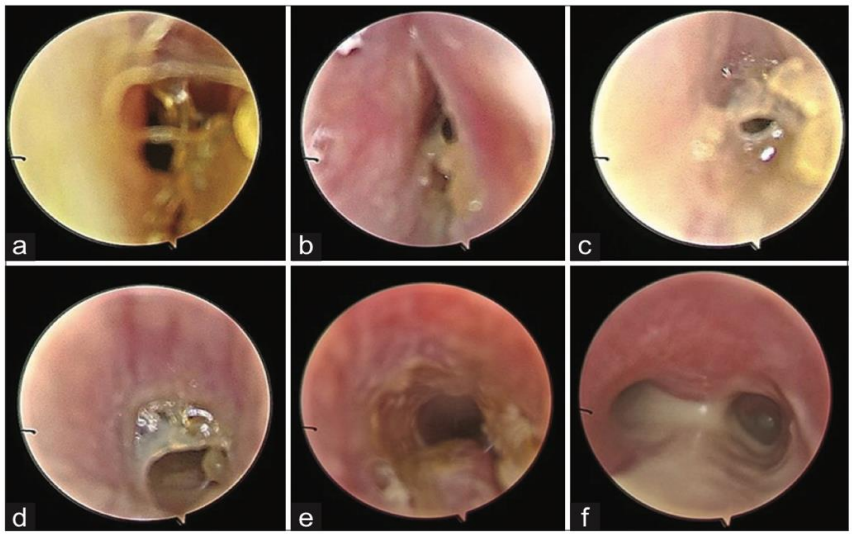


Figure 1: (a-c) bronchscopy images showing purulent fibrinous adhesions in supraglottis and glottis region. (d-f) Purulent exudates in the trachea extending upto carina and main bronchi

purulent material in the main bronchus [Figure 1]. Lavage was done and material sent for culture sensitivity.

Child was intubated in the operating room and was continued on mechanical ventilation in PICU. Bronchial lavage culture showed growth of Moraxella catarrhalis, which was sensitive to cephalosporins. Fever gradually subsided and repeat sepsis markers showed decreasing trend. Child was mechanically ventilated for 72 h.

Respiratory secretions reduced and child was extubated to high-flow nasal cannula. There was minimal post-extubation stridor which responded to steroids and epinephrine nebulizations. Child was weaned off oxygen over the next 48 hours. Antibiotics were continued for a total of 14 days (intravenous followed by oral) and child was asymptomatic on follow-up.

CASE 2:

A 10 months old previously well male child presented with cough, cold along with fever x 1 weekHe was treated outside with IV Antibiotics for 2 days. In view of worsening respiratory distress, he was referred to our hospital. On admission, Child was lethargic, Febrile (100.6 deg F) and tachycardic (heart rate 190 -200 /min). In view of toxic appearance, severe inspiratory stridor, with minimal response to epinephrine nebulisations – possibility of bacterial tracheitis was considered. Child was started on HFNC, IV specific spectrum antibiotics (based on the causative agents) and epinephrine nebulisations. Diagnostic Bronchoscopy done (fig 2) showed purulent secretions and inflamed airway upto subglottis.

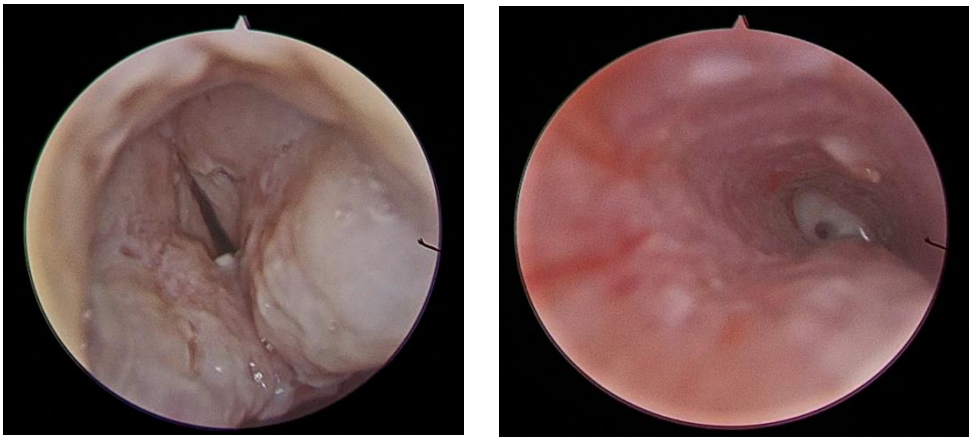


Fig.2 : Bronchoscopy image: upper airway inflammation with purulent exudates

Pus collected ,sent for C/S grew Methicillin resistant Staphylococcus aureus.Child was continued on HFNC, IV antibiotics and nebulisations. Fever spikes and Stridor reduced gradually. He was weaned off oxygen over next 48 hours.Antibiotics were given for a total 14 days (IV f/b oral).

DISCUSSION:

Bacterial tracheitis is characterized by production of mucopurulent exudates with ulceration and sloughing of mucosa.Common causative bacteria include Group A Streptococcus, Staphylococcus aureus, and M. catarrhalis. It affects most commonly affects children between 6 months to 8 years.Alternative diagnoses in children should be considered for children presenting with features of viral croup but do not respond to conventional therapy. The affected children generally appear more toxic with high grade fever. Swallowing of oral secretions is usually preserved and therefore drooling may be absent in bacterial tracheitis unlike in epiglottitis. Recommended first-line empirical antibiotics include third-generation cephalosporin plus vancomycin or clindamycin. Role of corticosteroids early in the disease is controversial.
Early aggressive airway clearance is necessary to prevent grave complications. Diagnostic and therapeutic bronchoscopy, appropriate antibiotics, and supportive care are the essential components for successful outcome.

CASE 3:-

A 40 days old girl infant ,who presented with cough and increased work of breathing since 25 days of life was admitted and treated as acute bronchiolitis outside for 4 days .Baby was on CPAP for initial two days of admission.After two days of discharge, baby again developed increased work of breathing and difficulty in feeding. Child was referred to ER in view of worsening respiratory distress. On admission, Baby afebrile, Irritable, Tachycardic (180 - 190 / min),Saturations (90-95 % with mask O2 5L/min)with suprasternal and subcostal retractions and inspiratory stridor. Cardiac examination was normal.
In view of severe respiratory distress, child was Started on HFNC, IV Piptaz and epinephrine nebulisations. Stridor improved after starting on HFNC. Blood investigations including blood counts, sepsis markers were normal.
Blood gas analysis showed Compensated respiratory acidosis. Chest x-ray was apparently normal. Two dimensional ECHO done to rule out any vascular malformation.

Bronchoscopy under IV sedation and spontaneous ventilation showed Loose aryepiglottic folds with collapsing redundant mucosal fold.
These findings (fig 3) were suggestive of grade 2 laryngomalacia, according to Onley classification of laryngomalacia.

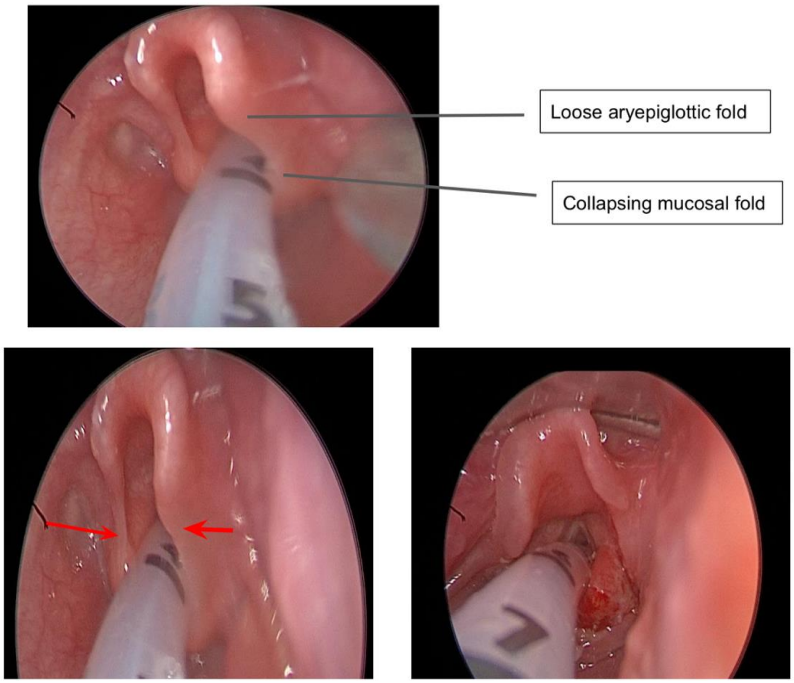


Fig 3: Supra glottoplasty : intra-operative images - showing loose aryepiglottic fold: red arrows: overhanging mucosa- divided and removed.

Supraglottoplasty was done under GA. Both aryepiglottic folds were divided partially and overhanging mucosa was removed using micro debrider.
Child was electively intubated and mechanically ventilated with sedation and paralysis for 48 hours. As stridor improved , baby was weaned off oxygen over next 24 hours. Child had no stridor or retractions on followup.

DISCUSSION:-

Laryngomalacia is the cause of 45-75% of all congenital stridors. It is a dynamic condition where supraglottic structures are weak and they collapse into the airway during the inspiration causing stridor. This stridor typically worsens with feeding, crying, supine positioning, agitation and exercise.Symptoms are usually not present at birth, start within the first 2 weeks of life and resolves by 12 to 24 months. Associated symptoms are swallowing dysfunction, regurgitation and cough. Diagnosis is confirmed by flexible fiberoptic laryngoscopy or bronchoscopy under general anesthesia. Evaluation of airway including assessment of synchronous airway lesions should be done. Mild cases can be followed up till 2 years of age with conservative measures like semi-prone positioning and anti reflux therapy. Supraglottoplasty is indicated if there is severe inspiratory stridor with oxygen dependency associated with feeding difficulty and poor weight gain.

CONCLUSION:

Inspiratory stridor can be a presenting symptom of various upper airway pathologies – ranging from benign anatomic abnormality to life threatening airway collapse and respiratory failure. History, age at presentation, associated syptoms and response to conventional therapy are useful

in differentiating infective and non-infective causes. Infective causes like bacterial tracheitis require early airway clearance and antibiotics to improve outcomes.

REFERENCES:-

1. Jones R, Santos JI, Overall JC Jr. Bacterial. tracheitis. JAMA 1979;242:721-6.
2. Singh RP, Marwaha RK. Fulminant branhamella catarrhalis tracheitis. Ann Trop Paediatr 1990;10:221-2.
3. 'Tebrueggea M, Pantazidou A, Yau C, Chisti M), Curtis N. Bacterial tracheitis-tremendously rare, but truly important: A systematic review. J Pediatr Infect Dis 2009;4:199.
4. Huang YL, Peng CC, Chiu NC, Lee KS, Hung HY, Kao HA, et al. Bacterial tracheitis in pediatrics: 12 year experience at a medical center in Taiwan. Pediatr Int 2009;51:110-3.
5. Blot M, Bonniaud-Blot P, Favrolt N, Bonniaud P, Chavanet P, Piroth L. Update on childhood and adult infectious tracheitis. Med Mal Infect 2017;47:443-52.
6. Pu S, Xu H, Li X. Supraglottoplasty in neonates and infants: A radiofrequency ablation approach. Medicine (Baltimore). 2018 Feb;97(7):e9850

CASE SERIES: Failure to thrive – Limelight for Cystic Fibrosis

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Introduction

Failure to thrive is not uncommon in Indian children. It is a chronic, sometimes potentially life threatening disorder of infants and children(1). Children are considered as failing to thrive when their rate of growth does not meet the expected growth rate for a child of their age(2). This may be due to various causes from antenatal maternal nutrition and habits to childhood nutrition, diseases and psychological issues(3). These causes leads to failure to thrive either by inadequate intake, inadequate absorption and increased requirement or by excessive loss(3). Here we are presenting a series of 6 cases presented with failure to thrive in infancy and diagnosed to have cystic fibrosis.

Presentation

Case 1, Baby.Mukund Vignesh, 3 months old male child from thenkasi, born by 2nd degree consanguineous marriage was presented with persistent cough for 1 month, white stools for 4 days and failure to thrive with weight loss of 400 grams from birth weight (<3rd centile for age). On examination he was tachypneic without desaturation and CXR showed B/L hyper inflated lungs. Serum electrolytes were normal and stool was white and had plenty of fat globules.

Case 2,Baby.Mohammed Uwais, 4 months old male child from puliangudi born by non consanguineous marriage was presented with vomiting, oily small frequency stools and failure to thrive with weight loss of 500 grams in last 2 weeks. Blood investigations showed low sodium, potassium and chloride ions. Fat globules were plenty in stools. CFTR hot spot mutation done was negative. Suspected Barters syndrome and evaluated.

Case 3, Baby.Haajira Banu, 15 months old female child from pottalputhur born by non consanguineous marriage was presented with fever, cough, breathlessness and failure to thrive with inadequate weight gain. Her sibling died at the age of 18 months with similar illness. On examination she had tachypnea and crepitations. Serum electrolytes were normal and had plenty of fat globules in stool and was pale.

Case 4,Baby.Aliya Mariyam, 8 months old female child from melapalayam born by non consanguineous marriage was presented with small quantity frequent stools and failure to thrive. She was emaciated. Blood investigations were normal. Stool examination showed pale stools and had plenty of fat globules.

Case 5, Baby.Ismail Anas, 4 months old male baby from thenkasi born by 3rd degree consanguineous marriage was presented with recurrent respiratory tract infections, sticky stools and failure to thrive since 2 months of age. History of sibling death for recurrent respiratory tract infections. Examination showed bilateral wheeze and electrolytes were normal. Stool were pale and microscopy revealed with plenty of fat globules.

Case 6, Baby.Ameera 4 months old female child from kayalpattinam born by 3rd degree consanguineous marriage presented with breathlessness with desaturation, semisolid stools for 20 days and failure to thrive with inadequate weight gain. Her sibling died at the age of 8 months with similar illness. Evaluation showed bilateral extensive pneumonia with low sodium and chloride. Plenty of fat globules were present in stool and stool was pale.

Diagnosis

All the 6 cases had some common features. Failure to thrive, Stool issues since early infancy either recognised by parents or not. Oily, white, pale, sticky, small quantity, frequent stools were suggestive of Fat malabsorption. Some parents complaint of diarrhea because of frequent stool passage. History of recurrent respiratory tract infections, occurring early or identified later. Electrolyte abnormalities can be present and family history of Sibling death due to respiratory illness present. Whole exome sequencing done in the suspicious groups and all these cases were positive for cystic fibrosis gene mutations.(Table-1) Sweat chloride could not be done in this region.

	CASE 1	**CASE 2**	**CASE 3**
FEATURES	Malabsorption Recurrent RTI FTT	Malabsorption Recurrent RTI-In later life FTT Electrolyte imbalance Metabolic Alkalosis	Malabsorption Recurrent RTI FTT Sibling death Electrolyte deficiencies
SUSPECTED	Malabsorption Syndrome /CF	Barters Syndrome / Malabsorption Syndrome /CF	Malabsorption Syndrome/CF
WHOLE EXOME SEQUENCING	CFTR-exon 14 c.2215delG-loss of function (deletion) AR-Homozygous	CFTR-exon 24 c.3925C>T-stop gained variation (novel) AR-Homozygous	CFTR-exon 14 c.2052dupA-loss of function (frameshift duplication) AR-Homozygous
	CASE 4	**CASE 5**	**CASE 6**
FEATURES	Malabsorption FTT Recurrent RTI – In later life	Malabsorption FTT Recurrent RTI Sibling death	Malabsorption Recurrent RTI FTT sibling Death
SUSPECTED	Malabsorption Syndrome /CF	Malabsorption Syndrome /CF	Malabsorption Syndrome /CF
WHOLE EXOME SEQUENCING	CFTR-exon 14 c.2052dupA-loss of function (frameshift duplication) AR-Homozygous	CFTR-exon 24 c.3925C>T-stop gained variation (novel) AR-Homozygous	CFTR-exon 22 c.3551A>T-impact sec.protein structure (novel) AR-Homozygous

Table-1

Discussion

CFTR gene (Cystic Fibrosis Transmembrane Conductance Regulator gene) is located in Chromosome 7q31. Which is expressed mostly in Pancreas, Gall bladder, Intestine, Parotid, Minor salivary glands, Epididymis, Palpabral Conjuctiva(4).

CFTR gene codes for membrane protein and ion channel which helps in the transport of sodium and chloride ions inside and outside respectively (Chart-1)(5). Mutation in this gene causes dysregulation of epithelial lining fluid, extracellular dehydration and thickened mucous which leads to it's clinical features (Image 1)(6)

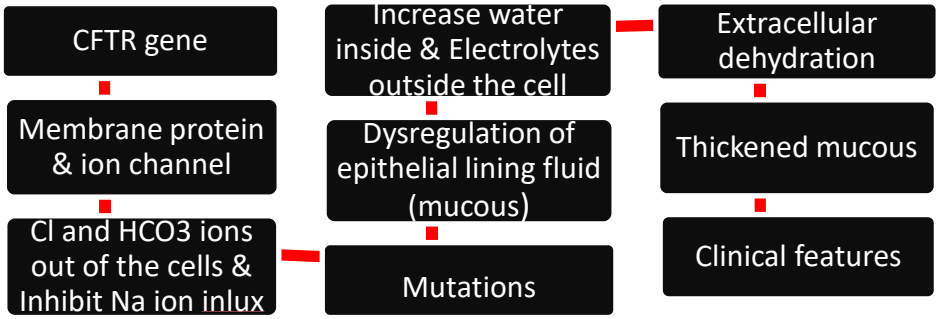

Chart-1

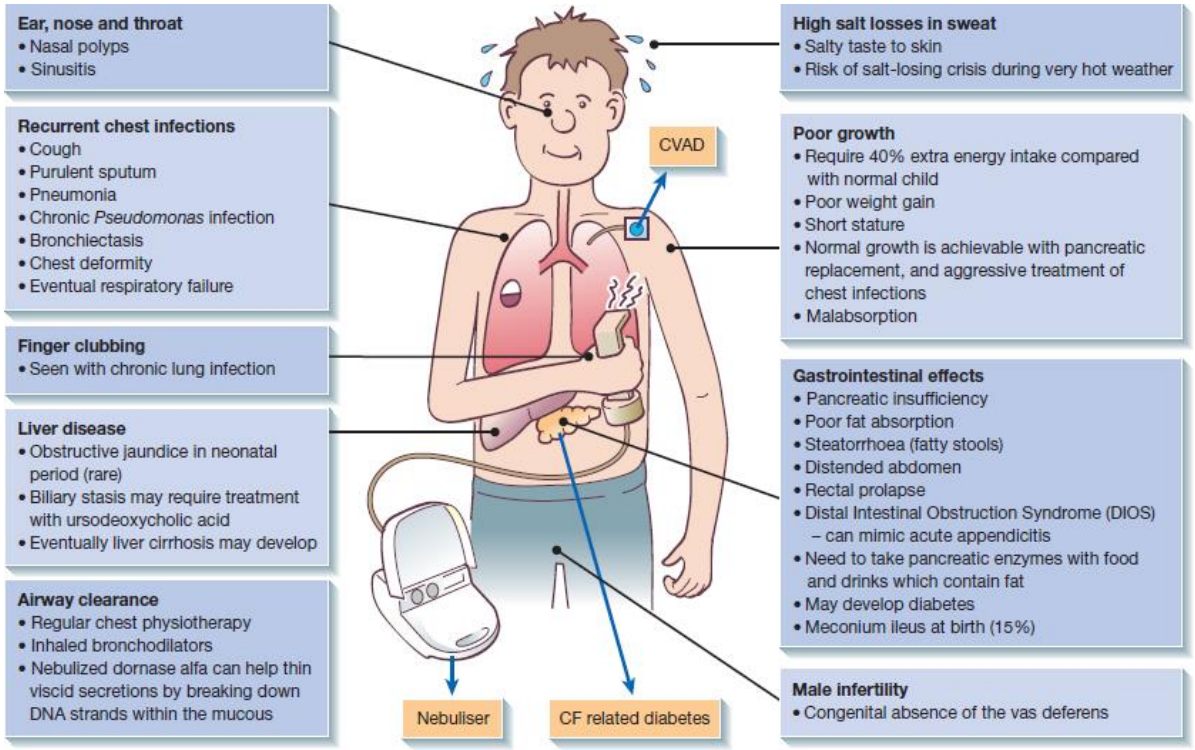

Image-1 (7)

Screening of cystic fibrosis can be done in newborn filter paper screening, sweat chloride test, transepithelial potential difference measurement and low thiocyanate and hypothyocyanate levels in saliva(8). Those who needs screening is mentioned in Table 2.

	INFANCY	CHILDHOOD	ADULT
SINOPULMONARY	- Infection	- ABPA - Sinusitis - Polyposis	- ABPA - Sinusitis - Polyposis - Respiratory failure
GIT	- Meconium ileus - Pancreatic Insufficiency - Rectal prolapse	- DIOS - Intussuception - Biliary Issues	- DIOS - Intussuception - Biliary Issues - Adenocarcinoma
OTHERS	- Dehydration - Hyponatremic hypochloremic metabolic acidosis	- Renal calculi - Hyponatremic hypochloremic metabolic acidosis	- Delayed puberty - Osteoporosis - Renal failure - Arthritis / Vasculitis

Table-2 (7)

Treatment of this condition is mainly symptomatic which consists of Airway clearance technique, Pancreatic enzyme replacement, Fat soluble vitamin supplementation, Probiotics, and some newer drugs(9) Dornase (Breakdown DNA in sputum → Decrease viscosity), Denufosol (Open alternate Cl channel → Decrease viscosity), Ivacaftor (CFTR protein enhancement), Trikafta (Combination of Ivakaftor, Tezacaftor, Elexacaftor). These newer drugs are effective against some specific mutations only. But we can prevent the disease by screening both the parents and by genetic counselling.

In our cases all children were presented with failure to thrive, malabsorption especially for fat and recurrent respiratory tract infections. Newborn filter paper screening cannot be used for older infants(10) (>3 months). Sweat chloride test is not available locally in our region. So we did hot spot mutation for cystic fibrosis in one patient but it came negative (It only detects most common mutations). There are >900 mutations in CFTR gene. So we did whole exome sequencing and got the diagnosis. We got 4 rare homozygus mutations among the 6 cases. In the 4, 2 were novel mutations.

Conclusion

Cystic fibrosis is not uncommon in our society. It is not a western country disease anymore. In infancy it usually presents with failure to thrive and malabsorption with or without respiratory illness. By proper diagnosis and treatment we can extend the life and by proper screening and counselling we can prevent the disease.

1. Wright CM. Identification and management of failure to thrive: a community perspective. Archives of Disease in Childhood. 2000;82(1):5-9.
2. Nangia S, Tiwari S. Failure to thrive. The Indian Journal of Pediatrics. 2013;80:585-9.
3. Krugman SD, Dubowitz H. Failure to thrive. American family physician. 2003;68(5):879-84.
4. McCarthy VA, Harris A. The CFTR gene and regulation of its expression. Pediatric pulmonology. 2005;40(1):1-8.
5. Ooi CY, Durie PR. Cystic fibrosis transmembrane conductance regulator (CFTR) gene mutations in pancreatitis. Journal of Cystic Fibrosis. 2012;11(5):355-62.
6. Gibson RL, Burns JL, Ramsey BW. Pathophysiology and management of pulmonary infections in cystic fibrosis. American journal of respiratory and critical care medicine. 2003;168(8):918-51.
7. Behrman RE, Vaughan III VC. Nelson textbook of pediatrics: WB Saunders company; 1983.
8. Farrell PM, White TB, Howenstine MS, Munck A, Parad RB, Rosenfeld M, et al. Diagnosis of cystic fibrosis in screened populations. The Journal of pediatrics. 2017;181:S33-S44. e2.
9. Graeber SY, Mall MA. The future of cystic fibrosis treatment: from disease mechanisms to novel therapeutic approaches. The Lancet. 2023.
10. Rock MJ, Mischler EH, Farrell PM, Wei L-J, Bruns WT, Hassemer DJ, et al. Newborn screening for cystic fibrosis is complicated by age-related decline in immunoreactive trypsinogen levels. Pediatrics. 1990;85(6):1001-7.

Mitochondrial disorders and *RRM2B* mutations – gaining clarity in a wiser way

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Introduction

The knowledge of the clinical spectrum and molecular genetics is continuously expanding in suspected mitochondrial disorders especially in the last decade. Of note is the **Mitochondrial DNA depletion syndrome (MDS)** with ***RRM2B* mutation**, with 15 infants reported in the literature till date. We report the first Asian child with *RRM2B* mutation and highlight the salient features.

Case:

- 4-month-old girl
- presented with poor feeding and lethargy
- non-consanguineous parents
- previous sibling death
- No neurological concerns at birth
- No family history
- Hypotonia with extensor posturing
- Lactic acidosis

The varied neurological and systemic manifestation prompted genetic testing which showed homozygous missense variation in exon 4 of the ***RRM2B* gene** resulting in aminoacid substitution of Histidine for Arginine at codon 121.

Table 1

Clinical phenotypes of different mitochondrial DNA depletion syndromes

Mitochondrial DNA depletion syndromes	Age of onset	Common clinical features
Myopathic		
TK2-related	Infancy—early childhood	Hypotonia and muscle weakness, facial weakness, bulbar weakness (dysarthria and dysphagia), elevated serum creatine phosphokinase
Encephalomyopathic		
SUCLA2- and *SUCLG1*-related	Infancy	Hypotonia and muscle weakness, psychomotor delay, scoliosis/kyphosis, abnormal movement disorders (dystonia, athetoid, or choreiform), sensorineural hearing impairment, epilepsy, growth retardation, lactic acidosis, elevated methylmalonic acid in urine and plasma, cortical atrophy and basal ganglia involvement in neuroimaging
RRM2B-related	Neonatal—infancy	Hypotonia and muscle weakness, psychomotor delay, microcephaly, sensorineural hearing loss, failure to thrive, lactic acidosis

Table 1

Clinical phenotypes of different mitochondrial DNA depletion syndromes

Hepatocerebral		
DGUOK-related	Neonatal	Hepatic dysfunction, psychomotor delay, hypotonia, rotary nystagmus developing into opsoclonus, lactic acidosis, hypoglycemia
MPV17-related	Infantile—childhood	Hepatic dysfunction, psychomotor delay, hypotonia, peripheral neuropathy, lactic acidosis, hypoglycemia, leukoencephalopathy in neuroimaging
POLG-related	Early childhood	Hepatic dysfunction, epilepsy, psychomotor delay, ataxia, neuropathy, hyporeflexia and hypotonia evolving into spastic paraparesis, stroke or stroke-like episodes, myoclonus, choreoathetosis, parkinsonism, nystagmus, somnolence, irritability, cortical visual loss, and sensorineural hearing impairment, generalized brain atrophy in neuroimaging
C10orf2-related	Neonatal—infancy	Hepatic dysfunction, psychomotor delay, epilepsy, peripheral neuropathy, hypotonia, ophthalmoplegia, nystagmus, athetosis, ataxia, sensorineural hearing impairment, lactic acidosis, cerebellar cortical atrophy in neuroimaging
Neurogastrointestinal		
TYMP-related	Late childhood —adolescence	Gastrointestinal dysmotility, weight loss, peripheral neuropathy, ptosis, ophthalmoplegia, elevated thymidine and deoxyuridine in plasma, leukoencephalopathy in neuroimaging

Discussion:

Mitochondrial disorders are traditionally viewed as difficult to diagnose but have more clarity in recent years. MDS are associated with a variety of neurological and systemic manifestations and can be classified into myopathic, encephalomyopathic, hepatocerebral, or neurogastrointestinal. MDS 8A is characterized by neonatal hypotonia, lactic acidosis, and neurological deterioration.

Conclusion:

Screening for mitochondrial disorders when the clinical suspicion is high, prior to neuroimaging or muscle biopsy is rewarding in resource limited setting lacking a structured metabolic pathway.

References:

(1) El-Hattab AW, Scaglia F. : review and updates of genetic basis, manifestations, and therapeutic options. Neurotherapeutics. 2013 Apr;10:186-98. (2) Suomalainen A, Isohanni P. Mitochondrial DNA depletion syndromes–many genes, common mechanisms. Neuromuscular Disorders. 2010 Jul 1;20(7):429-37. (3) Keshavan N, Abdenur J, Anderson G, Assouline Z, Barcia G, Bouhikbar L, Chakrapani A, Cleary M, Cohen MC, Feillet F, Fratter C. The natural history of infantile mitochondrial DNA depletion syndrome due to RRM2B deficiency. Genetics in Medicine. 2020 Jan;22(1):199-209.

‘Eyes that look and the Eyes that see’ -common neurological manifestations with uncommon inherited metabolic disorders.

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OBJECTIVES

Neurologic manifestations often masquerade underlying inherited metabolic conditions. We highlight four children with common neurological presentation who had rare underlying inherited metabolic disorders in a single center in one year.

Neonatal Seizures:

31-week preterm born to consanguineous parents with multiple previous abortions and preterm deaths presented with refractory seizures and hypotonia. Genetics showed (second Asian child with) novel nonsense homozygous variant in the ***ALDH4A1 gene*** causing **Type 2 hyperprolinemia (1)**.

Floppy infant:

4-month girl born to non-consanguineous parents presented with hypotonia, extensor posturing poor feeding and metabolic acidosis. Genetics showed (first Asian child with) homozygous missense variation of the ***RRM2B gene*** causing **mitochondrial DNA deletion syndrome (2)**.

Developmental Delay:

1-year girl born to non-consanguineous parents presented with developmental delay, peripheral hypotonia and feeding difficulty. Genetics showed ***POMK+ mutation*** causing **limb girdle muscle dystroglycanopathy**, so far reported in two patients (3).

Epilepsy:

11-year boy born to consanguineous parents with normal development presented with recurrent seizures followed by vomiting. Genetics showed ***heterozygous PCCA mutation*** (unlike common homozygous mutation) causing **propionic acidemia (4)**.

Conclusion:

All children had hypotonia as a common feature with non-neurological clues. Whole genome sequencing in suspected children can be resourceful and reveal rare diseases in resource limited settings.

References:

(1) Kaur R, Paria P, Saini AG, Suthar R, Bhatia V, Attri SV. Metabolic epilepsy in hyperprolinemia type II due to a novel nonsense ALDH4A1 gene variant. Metabolic Brain Disease. 2021 Aug;36(6):1413-7. (2) Bornstein B, Area E, Flanigan KM, Ganesh J, Jayakar P, Swoboda KJ, Coku J, Naini A, Shanske S, Tanji K, Hirano M. Mitochondrial DNA depletion syndrome due to mutations in the RRM2B gene. Neuromuscular Disorders. 2008 Jun 1;18(6):453-9. (3) Johnson K, Bertoli M, Phillips L, Töpf A, Van den Bergh P, Vissing J, Witting N, Nafissi S, Jamal-Omidi S, Łusakowska A, Kostera-Pruszczyk A. Detection of variants in dystroglycanopathy-associated genes through the application of targeted whole-exome sequencing analysis to a large cohort of patients with unexplained limb-girdle muscle weakness. Skeletal muscle. 2018 Dec;8:1-2. (4) Wang H, Meng L, Li W, Du J, Tan Y, Gong F, Lu G, Lin G, Zhang Q. Combinations of exonic deletions and rare mutations lead to misdiagnosis of propionic acidemia. Clinica Chimica Acta. 2020 Mar 1;502:153-8.

Financial Literacy for Doctors

Doctor's Vulnerability:

Due to our hectic work schedule, lack of time to read financial magazines and reluctance to discuss financial matters with the peers, we seldom possess adequate financial knowledge in personal finance and at the same time we are reluctant to take the services of the qualified financial advisors also.

Moreover we easily succumb to the pressure of the banking and financial sector people and investment agents who are mostly our relatives or friends and take wrong financial calls resulting in huge loss.

In order to avoid this we must get basic training in personal finance to take correct financial decisions and become confident in managing our personal finance. In case we feel it is difficult , then we can engage a financial adviser registered under SEBI to manage our finance.

Need For Financial Prudence:

Due to extended period of education and heavy inflow of specialist and super specialist in our field, our practice has become increasingly difficult. we settle very late in our life and start to earn only at the age 30 to 35. Marriage and having a child are also delayed because of this. Even then when we start our life, we go for multiple loans to buy a car, house, to set up hospitals or clinic, leading to heavy debt trap which takes away most of our income as interest. To overcome all these things, we must have adequate financial knowledge to optimise our investments to get good returns.

Finance Advice In A Nutshell :

1. Start financial planning and Retirement planning at an early age as soon as we start earning and not very late in life.

2. Save with discipline and invest with innovation.

3. Take Term insurance upto 10 to 15 times of your annual income plus all our liabilities. Don't go for any other insurance. Always remember investment and insurance are entirely different and should not be clubbed.

4. Take Health insurance for Rs 25 lakhs immediately after starting to earn and can do top up with another 25 lakhs. Once we are married, the policy can be a family floater which includes our spouse, children and parents.

5. Save money, equivalent to 6 to 12 months of our expenses in Liquid Mutual funds as emergency money which can be withdrawn within 24 hours. It will earn more interest compared to S/B account. It is better to have money equivalent to 3 months of our expense as cash in hand. Some mutual funds offer ATM cards for Liquid Mutual Funds.

6. Generally there is an advice that percentage of savings equivalent to your age can be in Fixed income instruments like Debt funds, PPF , Post Office Schemes, Bank FDS. Most of the time the returns from these investments doesn't beat the inflation after paying

income tax, resulting in de growth of the value of our money. To beat the inflation, our investment returns should be at least 10%. Mutual funds and Shares can give this returns. Youngsters should keep the fixed income securities investment as minimum. Little older people and retired people can go for this category. For income tax exemption under 80c , best options are ELSS and PPF. Under latest tax regime decide whether you are going to avail tax benefit under 80C. Interest from PPF and Sukanya Samriddhi Yojana which is an investment for girl child are tax free.

7. Gold: only 5% to 10 % of our investment can be in gold and that too only as Exchange Traded Fund (ETF) or Sovereign gold bond which are paper Gold. Buying jewellery has only ornamental value and will not serve as good investment option as we are loosing 20% of our investment on day one itself by way of wastage and making charges.

8. Real estate: After demonetisation and GST introduction , real estate has lost most of its sheen. If at all you want to invest, invest in Plots in developing areas, commercial property. Regarding Flats, buy first flat without any hesitation if you are going to live in it. Think twice before buying second flat as they are going to bring only loss. Most of the time good Mutual fund earns more than the real estate.

9. Investment in Mutual funds are very important as long term investment of 7 to 10 years. They give inflation beating return of 10 to 12%. They should constitute 50% to 70% of our investment .Instead of investing in Multiple Mutual funds, choose 4 or 5 good mutual funds and invest under systematic investment plan (SIP). Our spread in mutual funds can be like this. large cap or index 50 funds1, Large and mid cap fund 1, Mid cap fund 1, Flexi cap fund 1 and ELSS 1. If you want to be a carefree passive investor, you can invest simply in few types of index funds.

10. Investments in shares requires lot of knowledge. When we don't have time to read financial magazines, we can choose best 10 to 20 top performing companies in various sectors and buy 1 or 2 shares every month or during every fall of Sensex by 500 to 1000 points. similarly we can buy Nifty BeEs also during every fall of Sensex. This Will give a decent return of at 12 to 15%. If invested properly, this investment will make us rich . Investing in stocks is wrongly depicted as gamble by few persons, instead careful investing gives unimaginable returns.Unless you are a professional, don’t go for intra day trading or F & O which makes most of the people to loose money.

11. Try to avoid high cost loans. Re organising our loan may save us from loss of money by way of interest . For this we may need the help of a financial adviser.

12. Try to use Debit cards instead of credit cards . Credit cards favours uninhibited and unnecessary spending.

Imagine we attain financial independence at the age 50 or 55, we can enjoy life in travel, reading, writing and spending time with our family and lead a peaceful life.Finally remember this famous quote “ Money without time is only a fake wealth, so try to balance your work, family and hobbies.

Wishing you a healthy and wealthy 2023.

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Women’s Mental Health Article

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"Hello. This is letter from a Woman. She is a mother. She is a doctor.
But before all this, she started as a young child just like others including the boys. She grew into a young girl just like other girls. Adolescence changed everything for her. The way she looks, she thinks and she feels. She faced the challenging society layered with expectations and judgements of different colours in her journey of growing. She identified herself and worked her way to studying into a doctor. All along, she needed to balance herself inside-out. Inside her was nature's biology that seemed more difficult to govern at times. Her biology always ruled her mind. Her voyage into motherhood only added more challenges to this. Now, she is battling both physically and mentally between being a woman, a mother and a doctor."

The prevalence of mental health illness among women doctors has skyrocketed. Recent estimates (international studies) revealed rates as high as 70% for depression, anxiety, sleep disturbances and substance use. Younger women succumbed more to depression and PTSD symptoms. Gender differences in terms of severity of symptoms showed higher values for female doctors irrespective of their marital status and nature of speciality. The steap was noticed especially during COVID outbreak with difficulty in adapting to social and family dynamics. There are studies published on physician burnout being more among women than men in the background of cultural factors.

Mental health of a woman is not merely determined by her natural resilience, intelligence and personality but also strongly impacted by radical biological changes at pivotal ages through her life. It is important to understand and embrace these biological changes alongside a steady support system extending beyond family to our community.

So where to start for a better mental health? When to ask for help?

Answers simply begin with our everyday life.
* Timely meals and sleep primarily regulate the working of a cascade of hormones in our body. Off-timed lifestyle slowly sets in chain of events and the cumulative impact affects mental health significantly.
* Incorporating "Mindfulness" into our daily activities to increase self-awareness helps us to live more in the present. This concept has been adapted from Zen Meditation of Buddhism.

* Self-care, as in me-time, cannot get more relevant than today. Few minutes of disconnection from our routine to rejuvenate ourselves can recharge vital neurotransmitters associated with mental illnesses.
* Choice of activities to relax has become challenging in this world of social media. Unfortunately, time-out or break from screen time is utmost necessary. Screens can turn into evil when we allow their long stay and they take over our lives - affecting across all generations from elders to children.
* Leisure activities can be both physically and mentally engaging but ultimately it must require effort from us - such as arts, sports, reading etc. Novelty always rewards us internally keeping us refreshed and going.
* Interactive time with family and friends play crucial role in knowing when it is time to get more help. The splurge of virtual interaction through social media has removed the need to spend time on relationships. Noticing a dear one at low point or leaning on shoulder of supportive caregiver during rough times always pave a easier route to understand if more professional help is needed.
* Most important of all - Learning the right information. The liberal access to boundless information strewn over internet can only consume lot of inefficient time and energy. It is important to gain knowledge from right resources, i.e qualified professional.
The extensive scientific research around the globe has lead us to evidence-based management of psychiatric illnesses including medications and psychotherapies.

And yes, it is possible - the ultimate goal for a better quality of life with sense of fulfilment.

ACTIVITIES DONE BY
IAP TTT

WHO WORKSHOP ON FEVER WITH RASH SURVEILLANCE

PEDIATRIC CARDIOLOGY & PEDIATRIC NEUROLOGY CME

TENKASI JUNE MEETING

PROGRAM WITH AHA KANNIYAKUMARI

PACT WORKSHOP

CHALLENGING CASE FORUM





TENKASI CRITICAL CARE MEETING



eBARANIPEDS LOGO LAUNCH

BREASTFEEDING WEEK

ORS DAY CELEBRATION

ACTIVE PAEDIATRICIAN AWARD

NUTRITION WEEK CELEBRATION

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