BENCHMARK MEDICAL TRUST™

# BMT Newsletter

July 2018 • Vol. No. 1

# Doctor's Day 2018

Welcome to Benchmark Medical Trust's (BMT) first newletter! We hope that you find these contributions from our esteemed members of interest, and we invite you to become a member as well. Happy Doctor's Day.

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*Transforming Healthcare*

## IN THIS ISSUE

# Appendicular Perforation in appendicitis

CASE REPORT BY
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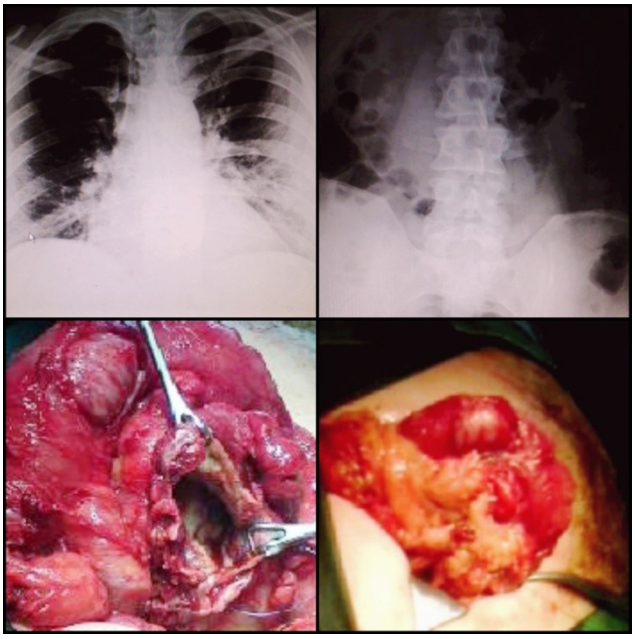
**Appendicular perforation at the base of caecum is a operative challenge in acute appendicitis.**



## BACKGROUND:

The most acute common surgical condition of the abdomen, acute appendicitis is linked with lack of fibre in the diet,viral infection,faecaliths or un known etiology. Perforated appendicitis is associated with higher mortalility rate of more than 5%.

We report a case of 30 yrs old female who presented with pain in right lower abdomen since 10 days associated with vomiting on and off, who was conservatively treated for suspected appendicitis in a clinic and refered since condition did not improve.There was no significant past medical , surgical & recent travel history.

Physical examination revealed PR 100, Temp 99.6f, BP 100/70mmhg, RR 20/mt. Abdomen was not distended but diffuse tenderness noted.There was a mass extending from RIF to Rt hypochondrium and bowel sounds were sluggish. Pre operatively inravenous fluroquinolones, cephalosporins and metrogyl started .Patient underwent laparotomy under spinal anaesthesia and was drained with wash.On inspection there was 2x 2 cm caecal perforation at the level of the base of appendix and appendix seen in bits.Thorough wash was given with saline and betadine solution omental patch was used to cover the caecum closing the abdomen with absorbable sutures to subcutanous tissues & staples to skin with corrugated rubber drain in situ.Patient was kept NPO for 5 days ,the drain was removed at 5thday,oral diet started on 6thday day .The patient passed flatus and motion on 6th day and discharged to home on 7th day after suture removal.Histopathology report revealed inflamed appendix with abscess formation and no malignancy noted.

## DISCUSSION:

Perforation of caecum is uncommon DDX for acute appendicitis.Rt hemicolectomy is the conventional approach for caecal permforation,closure by omental patch as in this case carries lesser postoperative complications rate like infection or septicemia..

## CONCULSION

Prompt diagnosis and early surgical referral can prevent acute appendicitis complications.

# Inborn Errors of Metabolism

## – Please don't miss the boat!!!

*For further details contact:*

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*Uppilipalayam*
*Coimbatore-15*

*Phone 04222575666*
*7373146666*

*geneomm@gmail.com*

Mrs.A and Mr.D were referred to us for genetic workup. Mrs.A has an ongoing pregnancy of 13-14 weeks.They are second degree consanguineous couple. A three generation pedigree was drawn and we found no major abnormalities in it. In her previous pregnancy the antenatal, perinatal and immediate postnatal period were uneventful. The baby was thriving well, had normal developmental milestones till 6 months of age.

At six months, baby had an acute febrile illness related to upper respiratory infection, suddenly became lethargic, had seizures and went in for unconsciousness. There was marked acidosis and baby succumbed eventually in two days inspite of intensive care. The urine organic acid was sent during the hospital stay and the results came later , was suggestive of Beta-ketothiolase deficiency. The family was counseled about the recurrence risk and was adviced to take genetic consultation before next pregnancy.

Inspite of the information to take genetic evaluation BEFORE CONCEPTION, they came back with an ongoing pregnancy of 13-14 weeks. This really has complicated the situation as we are left with a very short time to do carrier mutation testing for Beta-ketothiolase deficiency and then proceed with prenatal testing as the index child's genetic mutation is not known. If the carrier screening would have been done before conception, we woud have this pregnancy approached in a well planned manner.

We started evaluating the couple, did carrier screening for them to know the mutation and we successfully found out the same. But by the time we finished this evaluation pregnancy has progressed and it was 24 weeks. Hence the family was informed to get delivered at a tertiary care setup and to bring the baby for postnatal evaluation.

*Carrier testing for beta-ketothiolase deficiency. Clinical history: This individual is consanguineously married to a third degree relative. Her first child was normal till six months of age and later died due to acute metabolic crisis. Organic acid profile was suggestive of beta-ketothiolase deficiency.*

*TEST DETAILS*
*This test covers the gene, ACAT1, associated with beta-ketothiolase deficiency*

*RESULT*
*This individual harbours a known, heterozygous pathogenic variant. p.Met193Arg. caused by substitutions in exon 6 of the ACAT1 gene.*

*KEY FINDINGS*

| *Gene* | *Variation* | *Zygosity* | *Clinical significance* |
|---|---|---|---|
| *ACAT1* | *chr11:108009767T >G c.578T> G p. Met193Arg* | *Heterozygous* | *Pathogenic* |

## CLINICAL INDICATIONS

*Family history -- The couple lost a child with clinical indications suggestive of beta-ketothiolase deficiency. The spouse harbors a heterozygous variant, c.578T>G (p.Met193Arg), in the ACATI gene.*

## TEST DETAILS

*Strand ® Mutation Specific Test:*
*To detect the variant, c578T>G(pMet193Arg), in exon 6 of the ACATI gene (RefSeq id: NM_000019).*

## RESULTS

*Positive for the tested variant, c.573T>G(p.Met193Arg), in the ACATI gene.*

## INTERPRETATION SUMMARY

*The individual is heterozygous for the pathogenic variant in the ACATI gene (Fig.1 ).*
*The beta-ketothiolase deficiency (caused by variations in the ACATI gene) is inherited in an autosomal recessive pattern; which means, the parents o fan affected child usually carry one copy of the defective gene.*

## ABOUT BETA KETOTHIOLASE DEFICIENCY

Beta-ketothiolase deficiency is an inherited disorder in which the body cannot effectively process an aminoacid, isoleucine. This disorder also impairs the body's ability to process ketones, hence ketoacidosis occurs. The signs and symptoms of beta-ketothiolase deficiency typically appear between the ages of 6 months and 24 months. Affected children experience episodes of vomiting, dehydration, difficulty breathing, lethargy and, occasionally, seizures. These episodes, which are called ketoacidotic attacks, sometimes lead to coma. Ketoacidotic attacks are frequently triggered by infections, periods without food (fasting), or increased intake of protein-rich foods.

Coming back to our patient, the Newborn was tested postnatally for the mutation found in the parents and unfortunately baby was found to be affected.

For the Index child we have adviced

•Regular checkups and immunizations with the primary pediatrician

•To continue Breast feeds ( breast feeds are relatively safer for infants with this disorder)

•Estimation of Carnitine levels and carnitine supplementation as they are prone for secondary carnitine deficiency

•To review with us during weaning, for Dietician advice

•Advice regarding Crisis management

-When the baby gets acute febrile episodes, avoid catabolic states, control fever, adequate oral fluids, avoiding fasting and dehydration

-If presents with crisis, correct hypoglycemia, acidosis and dehydration along with supportive management and also contact us for further guidance.

## LESSONS LEARNT

In any child with suspected IEM

•Please do IEM profile as early as possible

•Please store 4-5 ml EDTA sample for DNA store, for mutation analysis later, if family not-affordable now and also till you get the IEM reports this can be a back up sample which can be used for mutation analysis in case of death

•Please remember these conditions have 25% recurrence risk and cannot be found out by routine scans

•Please refer them before next pregnancy as we will have enough time to test the family and hence plan regarding prenatal testing !

## INDICATIONS FOR TEST

*Family history suggestive of beta-ketothiolase deficiency (BKT deficiency).*

## TEST DETAILS

*Strand ® Single Mutation Confirmation Test:*
*To detect the variant, c.578T>G(p.Met193Arg), in exon 6 of the ACAT1 gene(RefSeq id: NM_000019).*

## RESULTS

*Homozygous for the tested variant, c.578T>G (p.Met193Arg). in the ACAT1 gene.*

# Topical Phenytoin In Trophic Ulcer

*For further details contact:*

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*Coimbatore – 641006*

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## INTRODUCTION

Trophic ulceration, common complication of leprosy is disabling, distressing and demoralizing for the patients. Factors such as focal hyposthesia, repeated trauma, autonomic disturbance of vessels, secondary infections and osteomyelitis has always been the major obstacle in the management. Phenytion, an anti epileptic drug can cause enhanced growth of fibroblast and hence promote healing of ulcers of various etiology. Phenytoin is involved in the healing process at several levels including stimulating fibroblast proliferation, enhancing the formation of granulation tissue, decreasing collegenase activity, promoting deposition of collagen, decreasing bacterial contamination and decreasing wound exudates.

## OBJECTIVE

To evaluate the effect of topical phenytoin in treatment of trophic ulcer in leprosy patients.

## METHODS

| | |
|---|---|
| *No of patients* | *: 23* |
| *Treatment* | *: Topical phenytoin powder application once daily* |
| *Duration* | *: 4 weeks* |
| *Inclusion criteria* | *: Trophic ulcer of more than 1-month duration* |
| *Exclusion criteria* | *: Patients with bony deformities* |
| *Parameters* | *: Reduction in surface area of ulcer Bacterial culture of ulcer swab Appearance of granulation tissue Cessation of ulcer discharge* |
| *Investigation* | *: Slit skin smear for AFB, X-ray foot Histopathological examinations after treatment* |

## PREPARATION AND ADMINISTRATION

*- Two tablets of 100 mg powdered and applied every day.*
*- Application to make a thin uniform layer over the ulcer.*
*- Wound covered with gauze.*
*- Phenytoin dressing repeated every day for four weeks.*

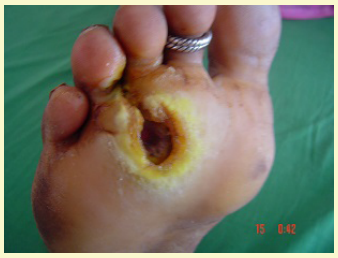
## GRADING

*Grade I : <1cm*
*Grade II : 1-2 cm*
*Grade III : 2-3 cm*
*Grade IV : > 4 cm*
*Grade I : Granulation tissue 25% coverage, Mild discharge*
*Grade II : Granulation tissue 50% coverage, Minimum discharge*
*Grade III : Granulation tissue 75% coverage, No discharge*
*Grade IV : Granulation tissue complete coverage, No discharge*

Comparison of parameters before and after 4 weeks of treatment.

| Sno. | Size Of Ulcer | | Culture Report | | Granulation Tissue | |
|---|---|---|---|---|---|---|
| | Before | After | Before | After | Before | After |
| 1 | III | III | +ve | +ve | I | I |
| 2 | IV | II | +ve | -ve | II | III |
| 3 | III | I | +ve | -ve | I | IV |
| 4 | II | I | -ve | -ve | I | IV |
| 5 | III | II | +ve | -ve | I | III |
| 6 | III | II | +ve | +ve | I | II |
| 7 | IV | IV | +ve | +ve | I | I |
| 8 | IV | I | +ve | -ve | II | IV |
| 9 | III | I | -ve | -ve | II | IV |
| 10 | IV | II | -ve | -ve | II | III |
| 11 | III | I | +ve | -ve | I | II |
| 12 | III | I | -ve | -ve | I | IV |
| 13 | II | I | -ve | -ve | I | II |
| 14 | III | I | +ve | -ve | I | IV |
| 15 | IV | I | +ve | -ve | I | III |
| | III | I | +ve | -ve | II | III |
| 16 | III | I | +ve | -ve | I | IV |
| 17 | IV | III | +ve | +ve | I | I |
| 18 | III | I | +ve | -ve | II | I |
| | IV | II | +ve | +ve | I | II |
| 19 | III | I | +ve | -ve | I | II |
| 20 | II | I | +ve | -ve | II | III |
| 21 | IV | II | +ve | +ve | I | I |
| 22 | III | II | -ve | -ve | I | II |
| 23 | IV | I | -ve | -ve | I | IV |

Before



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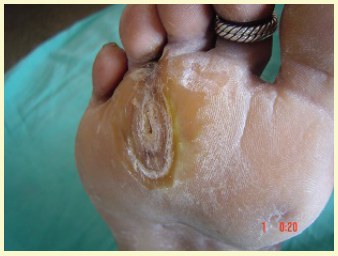
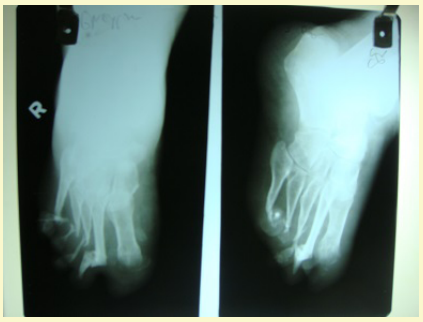
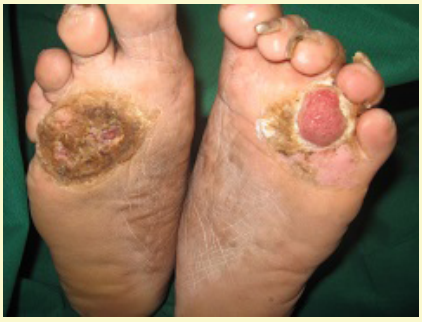




XRay



Control vs Phenytoin

## DISCUSSION

*- Total no of patients: 23 (male: 15, Female: 8)*
*- Patients age range 9-78 yrs. Mean age is 49 years*
*- Five out of 23 patients were receiving MDT-MB treatment*
*- Duration of ulcer range - 2 months to 24 months, with a mean of 9 months.*
*- Post treatment reduction in ulcer size in 86% of cases*
*- Bansal and Mukul compared the wound healing effects of topical phenytoin with normal saline and revealed healing is more in the phenytoin group.*
*- Pus culture report shows bacterial growth in 74% of cases. Post treatment culture report showed negative results in 72% of previous positive cases. Phenytoin decreases bacterial contamination and wound exudate .*
*- Appearance of granulation tissue observed in 85% cases*
*- One female patient aged 72 years with bilateral trophic ulcer treated with phenytoin dressing on one side and saline on otherside showed marked improvement on phenytoin side*
*- Biopsies done in 3 patients showed evidence of collagenization and decreased cellular infiltrate.*
*- Studies done earlier with various preparations of phenytoin showed the most favourable result with powder . So we used phenytoin powder in our patients. No side effects reported.*

# The Menace of MDR

*For further details contact:*

*Dr.KRUPASANKAR*
*MD;Medical Oncology*

*Adayar Cancer Institute, Chennai.*

Practising as a medical oncologist in a tertiary care centre for the past 5 years, the one thing that has caught my undivided attention has been the prevalence of patients presenting with infections caused by community acquired multidrug resistant organisms (MDRO's) which prompted me to write this article.

At our institute, we did a retrospective analysis on the prevalence of MDRO's in the stool of paediatric patients with acute leukemia and correlated it with blood culture positivity and outcomes which was published in Pediatric blood and cancer in July 2017. MDRO was defined as resistance to atleast one antibiotic in 3 or more of the following groups of antimicrobials – Cephalosporins / Beta lactam – Beta lactamase inhibitor / Carbapenems / Fluoroquinolones and Aminoglycosides.

To our amazement, at the end of our analysis, we realised that 50 percent of our paediatric patients with acute leukemia presented with community acquired MDRO's in the stool at admission.

Among these patients, we found that one-third of them grew the same organism in the blood also, which was associated with increased mortality among these patients.

Our retrospective analysis went on to show the high prevalence rate of community acquired MDRO's in the stool of newly diagnosed children with acute leukemia and that it was associated with increased positivity of blood cultures and mortality.

To our knowledge, this was the first study from India looking at the concordance between stool and blood cultures in paediatric patients with acute leukemia and further large prospective studies are needed to validate our findings.

Prevalence of community acquired MDRO's has led to increased antibiotic resistance which remains an unmet need and figuring out ways to reduce the prevalence of community acquired MDRO's remains the need of the hour.

*For further details contact:*

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*MD Radiologist*
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# Vascular Neoplasms And Low Flow Malformations Of The Craniofacial Region With Endovascular Management

## STUDY

To assess the effectiveness and safety of endovascular management in vascular neoplasm and low flow craniofacial malformations.

## MATERIALS & METHODS

*Sclerosing agent - Mixture of ethanol & lipidol (radiopacity) 8 : 2*

*Embolising agent – PVA particles*

## METHOD

*- under GA*
*- Aspirate the contents before injecting the sclerosing and embolising agents*
*- Test Injection*
*- Under road map – Direct puncture & Inj. of sclerosing agent*

## CRITERIA APPLIED

*Age*
*Flow characteristics of the vascular malformation*
*Unmanageable symptoms*

## DIAGNOSTIC ALGORITHM

*X-ray*
*Ultrasound*
*MRI*
*Check angio DSA*
*Direct puncture embolisation*

## CONTRAINDICATION

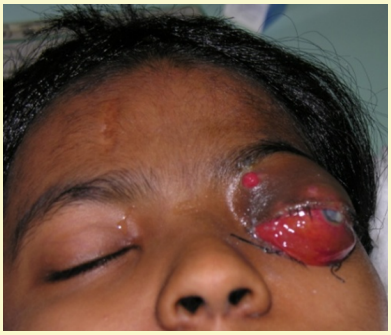
*No particular contraindication however too large a malformation may require multiple sittings of endovascular management*

## PROGNOSTIC GUIDE

*Follow up MRI to assess for change of T2 hyperintensity -> hypo intense areas which is suggestive of good prognosis.*

12 yr Old Girl Presented with painful progressive proptosis of left eye

PRE OPERATIVE



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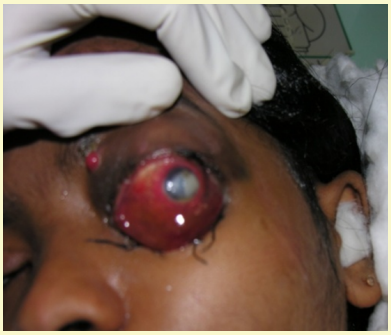
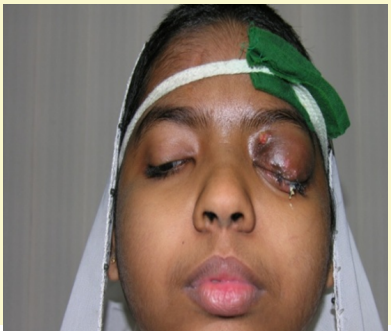
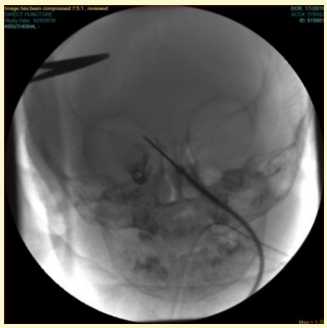
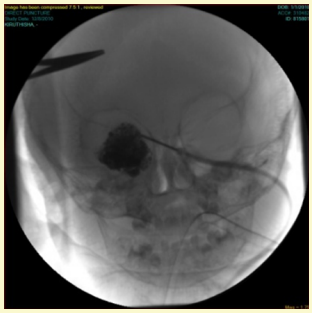
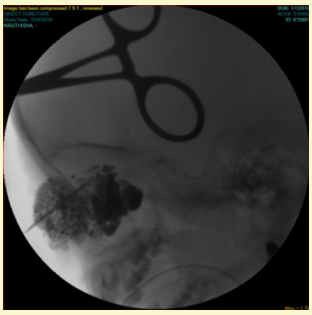
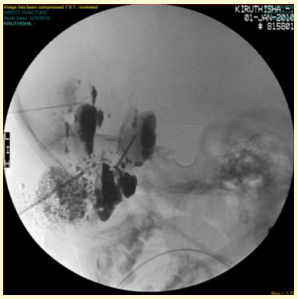
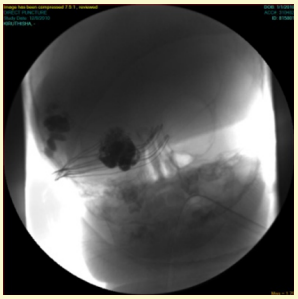


PRE OPERATIVE

POST OPERATIVE

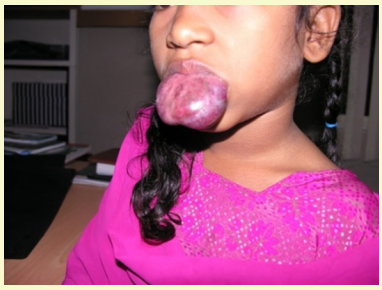


Left orbit - Low flow vascular malformation

7 yr Old presented with swelling over the tongue since birth & increased in size as the child grew up

PRE OPERATIVE



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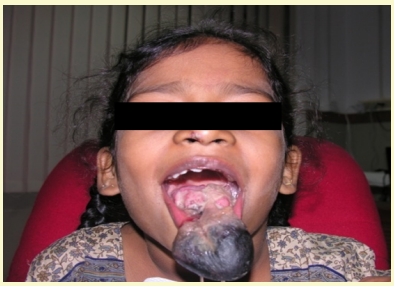

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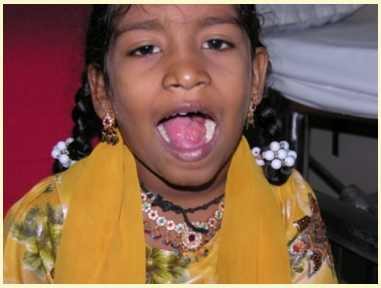

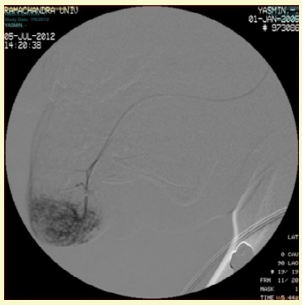

Embolisation of the low flow vascular malformation in the anterior 1/3 of tongue

## RESULTS

*A series of 11 patients with venolymphatic malformations were treated by endovascular management.*
*Treatment response was excellent for macrocystic lesions, good for combined lesions and fair / good for microcystic lesions.*
*None of the patients experienced adverse effects related to the treatment.*
*In a follow up period of 8 months 2 patients experienced lesion recurrence in a setting of concomitant infection.*

## CONCLUSION

*Low flow vascular malformation are challenging and difficult group of entities that can be successfully managed with interventional techniques.*
*Surgery carries high risk of disfigurement and mortality.*
*Endovascular management carries a good prognosis provided there is good imaging facilities.*
*It is important for the physicians who treat these complex patients to be familiar with different approaches, techniques, sclerosing & embolising agents that can be used, so that the patients can be offered the best available treatment at an earlier stage without causing any permanent functional & asthetic damage.*

# Blended Oxygen & Babies

*For further details contact:*

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*dr.r.nikhil@gmail.com*

Over the last few decades, the standard practice of initial use of 100 % oxygen, whenever supplemental oxygen is needed has been challenged, as increasing evidence has shown that hyperoxia due to oxygen supplementation may result in tissue & organ injury, especially the newborn retina and lungs.

Blended oxygen is delivered by a simple device called oxygen blender, which helps mixing medical air and hospital grade oxygen to deliver gas with 21% oxygen to 100 % oxygen at an accurate, controlled manner through any interface such as cannula/mask/bag & mask device/t-piece resuscitator.

Current recommendations for the use of supplemental oxygen in neonatal resuscitation:

• For neonates born at ≥35 weeks gestation, resuscitation is initiated with room air (21% oxygen).

• For neonates <35 weeks gestation, resuscitation is initiated with 21% to 30% oxygen.

• The oxygen concentration should be adjusted to achieve targeted SpO2 levels, which are monitored by pulse oximetry.

• The supplemental oxygen concentration should be increased to 100% if chest compressions are initiated.

Beyond resuscitation, for the neonates who receive supplemental oxygen, the target levels of neonatal oxygenation are based on hemoglobin oxygen saturation (SpO2) measured by a simple continuous noninvasive pulse oximeter. An optimal target pulse oximetry saturation range between 90 to 95% for preterm infants provides adequate oxygenation that meets the metabolic needs of the neonate while avoiding high concentrations of oxygen, hyperoxia and hypoxia.

Data from clinical trials suggest that hyperoxia is proven risk factor for Bronchopulmonary dysplasia (BPD) and Retinopathy of prematurity (ROP).

Hence, the need of the hour in all obstetric centers and neonatal intensive care units (NICU) is a blended oxygen source and pulsoximeter.

*For further details contact:*

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*Gastro Enterologist*

*Winnepeg Clinic,*
*CANADA.*

# Hydatid cyst of the liver diagnosed with endoscopic ultrasound

## AIM

We report a case of Hydatid cyst in the left lobe of the liver, juxtaposed to the gastric wall which was diagnosed with endoscopic ultrasound & Fine needle aspiration.

## METHODS

A 58 yo first nation male was investigated for recurrent right upper quadrant pain. CT abdomen showed a stable liver mass for 2 years. MRI of the liver identified an exophytic mass adjacent to the posterior aspect of the left lobe of liver & the lesser curvature of the stomach measuring about 5cm. EUS within the region revealed a 4.1 X 4.1 cm hypo echoic heterogeneous mass. The lesion appeared solid & it was not possible to ascertain which organ the lesion originated from. (i.e. stomach or liver) Using the linear array echo-endoscope, a total of four passes were obtained via FNA. Samples obtained were sent to cytology & pathology.

## RESULTS

Cytology showed abundant amorphous debris, a few cells & hooklets consistent with hydatid cyst. Final Histopathology reports were consistent with Echinococcus multilocularis the kind which is less likely to burst & seed the abdomen.

## CONCLUSION

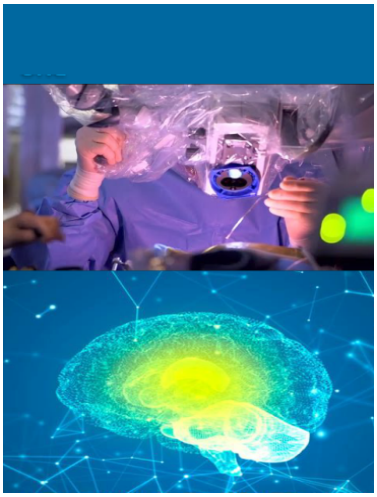
Hydatid cysts are most commonly found in liver (about 75%). Trans-abdominal ultrasonography, CT & MRI are the most common imaging modalities used. Through our case we think EUS can also be used provided it is in the right vicinity.

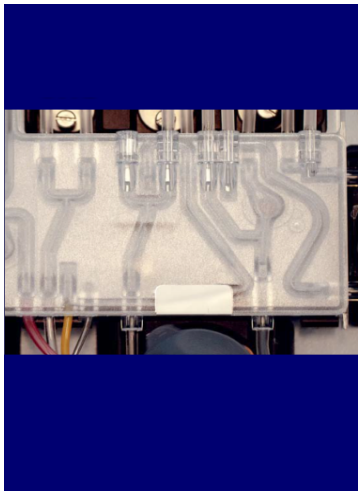
Neuro One

Leaders in Neuro Care

# Technology Meets Expertise

*State of the art technology advances is a key element in Neuro-sciences. We provide the best in technology by bringing together the finest physicians, the best surgical outcomes and utilizing the most up to date technology to ensure the greatest success. Our State-of-art technology include real time Imaging, Galileo EEG, Fluresence Guided* Surgery(FGS), and Plasmapharesis System.





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