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Posters

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Tonsillectomy in a patient with PFAPA and 7p22 deletion syndrome

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Syndrome is a rare cause of regular, repeated episodes of fever, sore throat and swollen neck glands in children although the exact etiology of PFAPA syndrome is unknown; one hypothesis suggests that the presence of infections and aberrant cytokine regulation are successive steps in a single ethiopathogenic process. The management of PFAPA is now considered to need the cooperation of ear, nose and throat (ENT) specialist. The role of tonsillectomy in literature is controversial. Some studies reported high success rates with tonsillectomy, further investigations are needed with larger numbers of patients. However, it is uncertain whether adenoidectomy combined with tonsillectomy adds any additional benefit to tonsillectomy alone. A 2-year Caucasian/Azerbaijan girl demonstrated repeated fever episodes with high levels (90-200mg/L) of C-reactive protein (CRP) since 6 months. In neonatal period she was observed because of microcephalus, slight developmental delay and growth retardation, muscle hypotonus and dysmorphic phenotype (broad forehead, hypertelorism, micrognathia and retrognathia, fluffy eyebrows, long and tight eyelashes, long filtrum, narrow lips). She was consulted by genetic and diagnosed with 7p22 microdeletions. During a period of 10 month she was hospitalized 6 times with the high fever, cervical/adenitis and sore throat Abdomen ultrasound, chest X-ray, EKG and EHHOKG were normal. Blood test for ANA, HIV, EBV, Borreliosis, tuberculosis, urine test and urine culture revealed no pathology. Cervical ultrasound showed increased lymphoid nodules with normal structure. There were no episodes of neutropenia. Procalcitonin level and blood culture were repeatedly negatives. Brain MRI with spectroscopy was performed to exclude intracranial pathology because of congenital problems. The patient was under ENT supervision to exclude the presence of otitis media during the high fever episodes. Adenoid hypertrophy was considered.

PFAPA was diagnosed by a pediatrician (repeated episodes of fever with aphthous pharyngitis, cervical/adenitis and high CRP levels, absence of neutropenia). Sequencing of genes was performed to exclude MEFV, MVK, TNFRSF1A, IL1RN and other gene abnormalities, using Illumina TruSight One expanded panel (6700 genes). No monogenic fever syndrome was revealed.

Prednisolone treatment 1mg/kg per os was used twice with excellent but temporary effect. Adenoidectomy was performed. The tissue of adenoids and tonsils has all signs of chronic inflammation. After the surgical treatment in a period of 7 months the patient was ill 4 times with no high fever (gastroenteritis, conjunctivitis, rhinopharyngitis and varicella with acute otitis media) and just once needed antibiotic treatment.

Biography

Andrei Antonov has completed his Medical School in 2001 from S. M. Kirov Military Medical Academy, Saint-Petersburg, Russia and then postgraduate studies in ENT from Tartu State University, Estonia. Currently he works as Head of ENT department in Tallinn Children Hospital and also in Tartu University ENT Clinic and North Estonia Medical Centre, Tallinn.

Notes:

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Case report of malignant infantile osteopetrosis presenting with persistant infantile hypocalcemia

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Malignant infantile osteopetrosis is a rare autosomal recessive disorder with a incidence of 1 in 2,50,000 children, presenting in infancy. It is a bone dysplasia characterised by reduced resorption of bone and diffuse symmetrical sclerosis due to impaired function of osteoclasts. This leads to obliteration of marrow cavity by bony overgrowth resulting in inability of bone marrow to participate in hematopoiesis. It presents with pancytopenia resulting in abnormal bleeding, easy bruising, progressive anemia and failure to thrive. Hepato splenomegaly due to extra medullary erythropoiesis, delayed dentition, cranial nerve palsies (deafness, blindness), hydrocephalus and seizures due to hypocalcemia. We describe the case of a 4 day old male neonate a third born to a 2nd degree consanguinously married couple, who presented with fever and two episodes of convulsions from day 4 of life vitals were stable with mild hypotonia on examination. Suspecting neuro infection due to late onset sepsis, a lumbar puncture was done which showed features of pyogenic meningitis. Routine investigations revealed hypocalcemia and calcium correction was given. Antibiotics, anticonvulsants were started and calcium maintenance dose was given. Child had two further convulsions in NICU. Repeat Lumbar puncture was normal and MRI brain was normal. Investigations continued to show persistent hypocalcemia despite adequate maintenance dose of calcium. Other workup for persistent hypocalcemia were normal except for chest X-ray which showed sclerotic changes. X-ray of other bones also showed diffuse sclerosis, bone in bone appearance. Later DNA analysis was sent which showed mutation of TCIRG1(+), homozygous with autosomal recessive inheritance specific for malignant infantile osteopetrosis. Osteopetrosis remains as a rare cause by clinicians unrecognized for neonatal hypocalcemia, which often results in diagnostic confusion and delay. This is important as early intervention with curative hemopoietic stem cell transplantation before optic nerve compression can help preserve the eyesight and improve the survival of the infant.

Biography

Vivetha Elango has completed MBBS in the year 2016. She is pursuing her postgraduate in MD pediatrics at Kempegowda Institute of Medical Sciences Bangalore. This is currently the first paper that has been done.

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Effectiveness of nutritional supplement in growth and development of children aged 2-12 years

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Objective: To determine the effect of 6 months consumption of nutritional supplement on growth and development of children aged 2-12 years.

Background: Young children in India suffer from some of the highest levels of stunting, underweight and wasting observed in any country in the world. The levels of over-nutrition are also on a rise. Prevention of child malnutrition require diets providing adequate energy and essential nutrients to promote catch-up growth, strengthen resistance to infection and support normal mental, physical and metabolic development.

Methods: This is an observational randomized controlled arm study, where a nutritional supplement (Groviva®) is given to 776 children, aged 2-12 years, for 6 months along with normal diet. Anthropometric parameters (height, weight and BMI) is assessed at baseline, 3 and 6 months. The z score for height, weight and BMI is used to analyze the results using Khadilkar growth chart 2009.

Results: A total of 763 subjects are included in the analysis. Each child is grouped according to age bracket (2-4yrs; 5-7yrs; 8-10yrs; 11-12yrs). After consumption of nutritional supplement for 6 months, z-scores for height, weight and BMI shown improvement in almost all age groups, compared to baseline. The improvement is significant in weight and BMI z-score. The standard deviation scores from expected increase in mean of weight, height and BMI is well within the permissible range. No adverse events is observed.

Conclusion: This study showed that 6 month intake of nutritional supplement by children provided a significant improvement in anthropometric parameters, with no adverse event.

Biography

Jasjit Singh Bhasin is a Sr. Consultant & HOD in Pediatric Department at BLK Super Speciality Hospital, New Delhi and the President of Indian Academy of Pediatrics (IAP), Delhi. He has 34 years of experience in Pediatric field. He completed MBBS from University of Delhi in 1980 and MD - Pediatrics from G B Pant Hospital/Moulana Azad Medical College, New Delhi in 1984. He is a member of Indian Medical Association (IMA) and Indian Academy of Paediatrics (IAP). His speciality interest are childhood asthma, nutrition and growth, infectious diseases.

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Short and long-term efficacy and safety of pediatric prolonged-release melatonin for insomnia in children with autism spectrum disorder

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Objective: To present results from an international multicenter study on the efficacy and safety of pediatric-appropriate prolonged release melatonin minitablets (Slenyto®) in children and adolescents with Autism Spectrum Disorders suffering from insomnia.

Methods: A 13 weeks double-blind placebo controlled study, followed by a prospective 9-month open-label follow-up study to test the efficacy and safety of Slenyto® in community dwelling patients with ASD suffering from sleep problems. Sleep measures included the validated caregivers' Sleep and Nap Diary (SND) and Composite Sleep Disturbance Index (CSDI) and additional measurements capturing child behavior (Strength and Difficulty Questionnaire, SDQ) and quality of life of parents (WHO-5).

Results: 125 children and adolescents with ASD or a distinct neurogenetic disorder (age 2-17.5 years; 96.8% ASD, 3.2% Smith-Magenis syndrome) treated by Slenyto® (2 or 5mg) demonstrated efficacy and safety in improving total sleep time (TST) (p=0.034), sleep latency (SL) (p=0.011) externalizing behavior (p=0.021) and quality of parents life (p=0.01) over placebo after the 13 weeks double-blind period. 95 patients who completed the 13 weeks double-blind trial (51 Slenyto®; 44 placebo) at final 2/5mg dose, received open-label Slenyto® with optional dose adjustment to 2/5/10 mg/day after 3 months. 41 of the Slenyto® randomized group completed 1 year of Slenyto® and 38 of the placebo randomized group completed 9 months of Slenyto®. Subjects treated continuously with Slenyto® for 52 weeks (N=41) slept on average 62.08 minutes longer (p=0.007), fell asleep -48.6 minutes faster (p<0.001) and had longer uninterrupted sleep duration (89.1 minutes; p=0.001). In addition, quality of sleep improved (p<0.001) and number of awakenings decreased > 50% (p=0.001). Quality of parent's life significantly improved during long-term treatment with Slenyto®. Child's sleep disturbance (CSDI), significantly improved (p<0.001) in all completers regardless of randomization history (N=79). Slenyto® was generally safe; the most frequent treatment related adverse events were fatigue in 5.3% (5 events) and mood swings in 3.2% (3 events) of patients.

Conclusion: Slenyto® is an effective and safe treatment option for short and long-term treatment of children with ASD suffering from insomnia.

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Responding to medical child abuse

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The inclination of Physicians to be investigation-oriented, fascinated by rare conditions, ignorant of abusive behaviours and accepting of reported histories (Donald and Jureidini 1996) has meant that the prevalence of Medical Child Abuse cannot be discounted despite the vast improvements in knowledge and awareness of this issue. It is unfortunate that parents are able to effectively manipulate the fact that clinical practice is based on an assumption of truthfulness and a shared interest in the welfare of the child, but the responsibility lies in the practitioner to be attuned to the possibility of such behavior. The reluctance to offend parents must be done away with if suspicions arise (Morley 1995). Often, the realization that their patient is suffering from MCA comes too late. Given that the single biggest risk factor for MCA is exposure to the medical profession, practitioners must be conscious of their role in their phenomenon. The presentation will thus canvass and summarise significant existing research on this area and in so doing, serve as a useful primer for busy practitioners. specifically, on (1) a diagnosis of the reasons pediatricians might be particularly susceptible to fraudulent behavior on the part of parents (2) discussion on risk factors for MCA (3) practical approaches for detecting cases of MCA in the early stages (4) useful strategies that ought to be employed when conducting medical history examinations (5) where it is a suspected case of MCA, the appropriate management of such cases.

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A quality improvment project on a guide to atrial ECG

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The American Heart Association practice standards for electrocardiographic monitoring in hospitals recommend to record an atrial electrogram whenever tachycardia of unknown origin develops in a patient after cardiac surgery. Most of the rotational junior doctors have general pediatric ckground and their experience in post operative cardiac patients care is minimal. This project was carried out at Glenfeld Hospital Leicester UK to promote awareness of junior doctors about safe and effective use of atrial ECG and also to build up a quick resource to perform atrial ECG especially in emergency conditions. A questionnaire was constructed to survey current awareness of doctors about atrial ECG. Also the participants understanding of existing trust guideline and the need of having a quick resource on method of performing atrial ECG was established. The survey included trainee doctors, Senior House Officers and Registrar, working in ICU and Cardiology ward and also Advanced Nurse Practitioners, Atrial epicardial pacing wires are commonly available in post-operative cardiac patients in ICU. However not all rotational doctors are aware of atrial ECG and some of them have no experience of doing atrial ECG. All doctors showed interest in learning the skill to perform atrial ECG. The Flow chart illustrating steps of doing atrial ECG were constructed and made available in ICU and Cradiology ward. Recommendation put forward to conduct teaching sessions on atrial ECG in induction programme for new rotational trainees. Flow charts were included in updated trust guidelines.

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A case series of thyroid hormone resistance

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Thyroid hormone resistance is a rare condition where there is an impaired sensitivity of target tissues to thyroid hormone. This leads to a situation where both the thyroid hormone levels and the thyroid stimulating hormone (TSH) are raised, as TSH is not suppressed as would normally be expected. Incidence of this condition is around 1 in 40,000 live births. TR-beta gene mutation is the most common cause of thyroid hormone resistance. Clinical manifestations are dependent on the type of mutant thyroid hormone receptor and each target tissue's predominant thyroid hormone receptor expression. Affected patients can present with a range of hyperthyroid or hypothyroid signs and symptoms. Majority of the cases described so far have an autosomal dominant inheritance. In this case series, we describe two young children and their father who have thyroid hormone resistance. They all possess a TR beta- gene defect secondary to a heterozygous mutation. Due to the widely variable signs and symptoms and the non-typical trend of laboratory markers in thyroid hormone resistance, it can be difficult for the clinician to diagnose if one is not familiar with it. Thus, this entity should be taken into consideration when one encounters a patient with elevated serum FT4, unsuppressed TSH and decreased serum T4/T3 ratio.

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Cardiac function in neonatal sepsis

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Objective: Neonatal sepsis is associated with the presence of the Systemic Inflammatory Response Syndrome (SIRS) in response to a culture-proven infection. It is known as one of the most frequent causes of mortality in the neonatal intensive care units. The study has aimed to investigate the effects of neonatal sepsis on cardiac function of the infants.

Methods: The study is based on prospective cohort research. It consists of two groups; control group and focus group. The focus group comprised of 30 full-term neonates with neonatal sepsis admitted to NICU; whereas, healthy neonates were included in the control group. Neonatal sepsis was diagnosed among the infants with the presence of at least two clinical signs of sepsis including feeding intolerance, temperature instability, apnea, poor reflexes, poor capillary refill>2 seconds. The clinical examination of neonates including CBC, CRP, blood culture and sensitivity was also conducted. Moreover, echocardiography was performed on participants of both groups.

Results: The results revealed that 50% of the patients from both the groups were male. The mean weight of the infants ranged from 2.2 to 3.5kg with a mean of 2.9±0.3kg. Results showed that 63.3% patients had low platelet count and 16.7% patients suffered from leukocytosis. 11 patients (36.7%), suffering from sepsis, were diagnosed with significant shift in their neutrophil count. There were significant changes in the echocardiogram of the patients suffering neonatal sepsis; whereas, dramatic improvement in cardiac function was observed by comparing the parameters before and after resolution of sepsis.

Conclusion: The septic neonates experienced significant cardiovascular changes that are revealed through the technique known as echocardiography.

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Hemodynamic effects of dobutamine versus dopamine in preterm infants: An update meta-analysis

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Hemodynamic effects of dobutamine versus dopamine in preterm infants: an update meta-analysis: It is a meta-analysis to compare the effects and safety of dobutamine versus dopamine in preterm infants with abnormal hemodynamic status. Study sources were up to 2017 for RCTs in which dobutamine and dopamine treatment was adopted. Included studies were conducted on preterm infants with abnormal hemodynamic status that reported mortality <28 days, treatment failure and organ effects. 7 articles were included with a total 286 patients. 5 studies reported mortality (180 patients), 4 studies reported P/IVL (145 patients), 4 studies reported P/IVH (160 patients), 2 studies reported severe P/IVH (105 patients), 3 studies reported NEC (140 patients), 2 studies reported BPD (55 patients) and 6 studies reported treatment failure (266 patients). Meta-analysis showed an increased probability in treatment failure using dobutamine treatment (RR, 1.67; 95% CI, 1.14-2.45; P = 0.008), whereas there was no significant difference in mortality <28 days (RR, 1.16; 95% CI, 0.70– 1.91; P = 0.57), P/IVL (RR, 2.90; 95% CI, 0.93-9.11; P = 0.07), P/IVH (RR, 1.23; 95% CI, 0.73-2.08; P = 0.44), severe P/IVH (RR, 0.58; 95% CI, 0.21-1.62; P = 0.30), NEC (RR, 2.21; 95% CI, 0.60-8.09; P = 0.23) and BPD (RR, 1.04; 95% CI, 0.38-2.82; P = 0.94) between two groups. Dopamine was more effective in treatment success in therapy of preterm infants with abnormal systemic hemodynamic status. No difference was found existed in mortality <28 days and incidence of adverse organ effects in two groups.

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Label-free proteomics of the fetal pancreas identifies deficits in the peroxisome in rats with intrauterine growth restriction

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Aim: The objective of the present study was to identify differentially expressed proteins (DEPs) in the pancreas of a fetus with intrauterine growth restriction (IUGR) and to investigate the molecular mechanisms leading to adulthood diabetes in IUGR.

Methods: The IUGR rat model was induced by maternal protein malnutrition. The fetal pancreas was collected at embryonic day 20 (E20). Protein was extracted, pooled and subjected to label-free quantitative proteomics analysis. Bioinformatics analysis (GO and IPA) was performed to define the pathways and networks associated with DEPs. LC-MS results were confirmed by western blotting and/or quantitative PCR (q-PCR).The principal parameters of oxidative stress-superoxide dismutase (SOD) were determined in blood samples of fetal rats.

Results: A total of 57 DEPs (27 upregulated, 30 downregulated) were identified with a 1.5-fold change threshold and a p-value≤0.05 between the IUGR and control pancreas. Bioinformatics analysis revealed that these proteins play important roles in peroxisome biogenesis and fission, fatty acid beta oxidation (FAO), mitotic cell cycle and histone modification. The peroxin Pex14 was downregulated in the IUGR pancreas as confirmed by western blotting and q-PCR.Pmp70, a peroxisomal membrane protein involved in the transport of fatty acids, was upregulated. Hsd17b4 and Acox1/2, which catalyze different steps of peroxisomal FAO, were dysregulated. SOD plasma concentrations in the IUGR fetus were higher than those in the control, suggesting partial compensation for oxidative stress.

Conclusion: The present study identified DEPs in the fetal pancreas of IUGR rats by proteomics analysis. Down regulation of pancreas peroxins and dysregulation of enzymes involved in peroxisomal FAO may impair the biogenesis and function of the peroxisome and may underlie the development of T2 diabetes mellitus in adult IUGR rats. The present data provide new insight into the role of the peroxisome in the development of the pancreas and may be valuable in furthering our understanding of the pathogenesis of IUGR-induced diabetes.

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The effects of helmet therapy, relative to the size of the anterior fontanelle in patients with nonsynostotic plagiocephaly: A retrospective study

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Background: Many studies have reported on the importance of treating patients with nonsynostotic plagiocephaly using helmets, a non-surgical treatment. We retrospectively examined the effects of helmet therapy according to the size of the anterior fontanelle.

Material and Methods: We enrolled 200 patients with nonsynostotic plagiocephaly who received helmet therapy between January 1, 2016 and December 31, 2018. We collected data pertaining to age at treatment initiation and treatment duration. We measured anterior fontanelle size using X-ray imaging and divided patients into one of three groups according to the structure's size. Group A was 0~25%, Group B was 25~75% and Group C was 75~100% of anterior fontanelle size. Helmet treatment feasibility was evaluated using Cranial Vault Asymmetry (CVA), Cranial Vault Asymmetry Index (CVAI), Anterior Symmetry Ratio (ASR), Posterior Symmetry Ratio (PSR) and Overall Symmetry Ratio (OSR) at baseline and at the end of the treatment.

Results: The average starting age was 20w (12-40w). The mean treatment period was 13w (10-24w). Group A had 53 cases, Group B had 102 and Group C had 45. The CVA differences between groups were 6.8mm (A & B), 9.5mm (A & C) and 2.7mm (B & C). The CVAI differences between the groups were 7.28% (A&B; p=0.001), 8.9% (A&C; p=0.003) and 1.62% (B&C; p=0.381). There were no interval changes for ASR in Group A; Group B changed 1% from 0.9 to 0.91, Group C changed 2% from 0.89 to 0.91. For PSR, Group A changed 6%, from 0.82 to 0.88; Group B changed 10%, from 0.79 to 0.89 and Group C changed 17%, from 0.73 to 0.9. In Group A, OSR changed 5%, from 0.85 to 0.90 in Group B, it changed 9%, from 0.81 to 0.90 and in Group C it changed 13%, from 0.78 to 0.91. There was no difference in the effects of helmet therapy according to the sex of each group.

Conclusion: The mean changes of CVA, CVAI and PSR were significantly greater for Group C, whose members presented with greater anterior fontanelle size. Helmet therapy is more useful for occipital, rather than frontal, asymmetry correction. Patients with greater fontanel sizes appeared to demonstrate relatively free cranial bone movement. Therefore, anterior fontanelle size could act as a prognostic factor for estimating the outcomes.

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Quality indicators of a pediatric emergency department of a public sector hospital, Pakistan

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Child Life Foundation, Pakistan

Introduction: Evaluation of quality indicator in any emergency department is a step towards better outcomes. Despite the high burden of pediatric mortality from preventable conditions in low and middle income countries and the existence of multiple tools to prioritize critically for early identification. While in Pakistan due to poor socioeconomic condition, there is high flow of patients in public sector hospital. In order to identify areas of improvement regular assessment of outcome is essential. Different parameters are part of quality indicators of any emergency department. For example, survival rate and number revisits.

Objective: The aim of this study is to evaluate quality indicators of pediatric emergency department of one of the biggest public sector pediatric hospital of Sindh, Pakistan.

Method: This is a retrospective cross sectional study over a period of 6 months (From January 2019-June 2019). All pediatric patients who were triaged as P1 were evaluated for survival rate. While number of revisit within 72 hours were evaluated irrespective of triage category.

Result: 76,297 number of patient present to emergency department during study period. 5,585(%) pediatric patients were triaged as P1. Among them 844(%) patient expired. Overall survival rate among pediatric age group is 84%. While during study period 3,537(%) number of patients revisit emergency department irrespective of triage category. Out of them 3,150(%) were planned while 387(%) revisits were unplanned.

Conclusion: Quality indicators indicate quality of care being provided at any emergency department. Survival rate and number of revisit enlightens areas of improvement. Regular assessment with implementation of hospital based protocols is crucial to improve quality indicators.

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Telemedicine based assessment of CPR performance in paediatric emergency departments of Pakistan

Muhammad Waqar Ibrahim, Sharmeen Nasir, Huba Atiq and **Shah Ali Ahmed**
Child Life Foundation, Pakistan

Introduction: Basic Life Support (BLS) and Pediatric Advance Life Support (PALS) are necessary for physicians working in pediatric emergencies. In Pakistan, similar to other developing countries, number of Pediatric Emergency Departments (PEDs) is low and available PEDs does not have credentialed and certified health professionals to provide life sustaining measures in a critically ill patient. We, Child Life foundation, started working in 2011, have developed 7 PEDs throughout Sind, Pakistan. We have American Heart Association's certified instructors who train our physicians and nursing staff for Basic Life Support (BLS) skills. These skills help us saving lives and increase the level of confidence of these physicians while managing a critically ill child. Child Life foundation has a sophisticated Telemedicine (TM) Program, which provides 24/7 real time, video based consultations as well as assessment of CPR skills of the bedside physicians.

Objective: This study aims to assess the CPR performance of bedside physicians via Telemedicine.

Method: Cardio-Pulmonary Resuscitation (CPR) being performed in the resuscitation rooms of either of the 07 Pediatric Emergency Departments (PEDs) of Child Life foundation, are frequently assessed by the Telemedicine Physicians on a predesigned checklist. It is a 21 points checklist, designed as per AHA guidelines for BLS. Assessment is done via High Definition Cameras and live monitoring to provide feedback to the physician. All data is kept strictly confidential in a password protected data sheet and feedback of outcome of CPR is given individually and maintain in their monthly performance sheet. We conducted a retrospective data review of CPR assessments done from January 2019 till June 2019.

Results: A total of 377 Cardio-Pulmonary Resuscitations (CPRs) were observed. 122 Number of physician observed. 99% of physicians checked response of patient and 95% called for help. 86% physicians checked for pulse while 81% looked for breathing. 84% of physicians performed appropriate rate of compressions and 69% let the chest recoil completely. In 81% of the encounters, appropriate airway positioning was maintained with head tilt chin lift and in 80% of patients correct E-C clamp was maintained. Ambu bag was attached to oxygen device in 98% but chest rise was being observed by only 51% of physicians. 78% physicians performed appropriate number of CPR cycles. Monitor was attached in 85% of CPRs observed, but rhythm check after every 2 minutes was done by only 45% physicians. Epinephrine was given in correct dose and appropriate dilution in 90% and 89% encounters, respectively. 45% avoided interruptions during CPR. Overall CPR performance was 76%.

Conclusion: Adequate Cardio-Pulmonary Resuscitation (CPR) performance is the key factor in saving lives in Pediatric Emergency Departments (PEDs) and regular, frequent assessments are necessary to keep a check and balance on performance to provide quality of care and to improve teaching and training.

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The first association of HB Knossos: (HBB: c.82G> T) with (HBB: c.118C> T) mutation causes thalassemia homozygous in Algerian children

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Beta-thalassemia is the most common disease among hemoglobinopathies in Algeria. Mutations found in Algerian beta-thalassemia patients constitute a heterogeneous group, consisting mostly of point mutations. Only in very rare cases did deletions or insertions cause affected or carrier phenotypes. Hb Knossos (HBB: c.82G> T) is a rare variant. In this study, we aimed to investigate the effect of compound heterozygosis for Hb Knossos (HBB: c.82G> T) and (HBB: c.118C> T). To our knowledge, this is the first report of such a combination related with beta-thalassemia major phenotype in a Algerian family, we used the minisequencing assay as a rapid screening procedure to identify most common HBB genetic variants and direct DNA sequencing to detect the rare mutations of HBB gene. Heterozygous inheritance of the mutation results in severe beta-thalassemia phenotype. The proband was a 13-year-old boy when first studied. He was referred because of severe anemia. Hematological analysis of the reveals Hb 7.2 g/dl with microcytosis of 71.1fl, hypochromia 25pg and the number of red blood cells is 2.9, 106/mm^3. In addition, a significantly secondary thrombocytosis and leukocytosis were reported in patient. Electrophoresis of hemoglobin in an alkaline medium shows Hb A2 = 4% HbF = 65% and blood smear confirms microcytosis hypochromia and showing the presence of many dacryocyte with hyper eosinophilia. The combination of these mutations Hb Knossos (HBB: c.82G> T) and (HBB: c.118C> T) causes the beta-thalassemia major phenotype and this is important for genetic counseling.

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Childhood infections and risk of type 1 diabetes in Tlemcen, Northwest Algeria

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Background: Type 1 diabetes is an autoimmune multifactorial disease caused by a complex interaction of genetic and environmental factors. Several studies have shown that viral infections cause the onset of type 1 diabetes by inducing immune responses that can damage β- cells.

Aim: This study aims to examine the interaction between type 1 diabetes and childhood viral infections in children of Tlemcen in northwest Algeria.

Patients and Methods: This is a case-control study of a population of 338 children under the age of 15 years, including 137 diabetic children and 201controls children, living in Tlemcen in Western Algeria. The data were collected using questionnaires submitted by a physician to the parents of the cases and controls, from February to May 2018. The data were analyzed by a logistic regression processed by Minitab.16 software.

Results: The total frequency of exposure to childhood infections (varicella, measles, rubella, mumps or tonsillitis) in early childhood is higher in diabetic children (81.75%) than in controls (66.66%), p=0.003. The risk of type 1 diabetes for children exposed to a single infection was only statistically significantly for rubella (p=0.016), odds ratio: confidence interval (CI) (OR: 3.73, 95% CI, 1.28-10.88). However, if two or more infections was contracted during the years before the onset of diabetes, the risk increases significantly (p=0.000), (OR: 3.33, 95% CI, 1.92-5.78).

Conclusion: The high prevalence of infectious diseases among young children in Tlemcen's population may explain the development of type 1 diabetes in children in this region.

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Exploration of the FLT3-ITD mutation in acute myeloid leukemia patients in the western Algerian population

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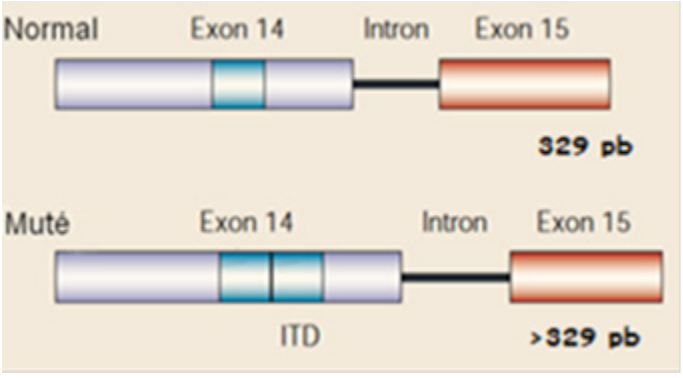
Statement of the Problem: Leukemia affects 9,000 people worldwide each year; with 3/700 have acute myeloid leukemia (AML). They arise from mutations that affect the genes influencing hematopoiesis. FMS-related tyrosine kinase 3 (FLT3) is a tyrosine kinase receptor usually expressed in hematopoietic progenitors, is the most common genetic lesion in AML with mutations detected in 25% to 40% of cases. There are two main types of mutations: tandem internal duplication (ITD), which is the most common (~ 25% of cases) and a point mutation D835 (TKD) (~ 5%). The detection of FLT3-ITD is important for the prognosis especially in those who have a normal karyotype.

Aim: The aim of study is evaluate the FLT3-ITD mutation frequency in the western Algeria population.

Material and Methods: We analyzed eighty-one patients with cytogenetic and molecular biology department at the University Hospital of Oran (EHU) and those from March 2014 to March 2018. We explored the FLT3- ITD mutation using the polymerase chain reaction (PCR).

Results: Statistical analysis showed that out of eighty-one AML patients, only eleven cases had the FLT3-ITD mutation with the heterozygous state. Which corresponds to a frequency of 12%? These results are in perfect agreement with the Chinese population estimated at 11%. However, our results are in disagreement with those reported in European population (50%) and the Egyptian population of 34.6%.

Significances: In this study, we highlighted the frequency of the FLT3-ITD mutation in the western Algerian population. It would be very interesting to consider undertaking a study on the impact of the size of the FLT3-ITD fragment on the prognosis since a study has shown that duplications of 48 to 60 base pairs are associated with very poor prognosis.



FLT3-ITD normal/mutation version

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Highly sensitive C-reactive protein–an independent determinant of risk of formation of essentsial arterial hypertension at teenage children

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Existence of signs of chronic inflammation at which increase in pro-inflammatory cytokine and as a result-growth of concentration of the C-reactive protein (CRP) is of the characteristic of the arterial hypertension (AH). CRP and arterial blood pressure are independent determinants of cardiovascular risk. 78 school students at the age of 14-16 years are investigated (middle age 14.7±0.2). From them 20 children with normal arterial blood pressure (NBP-control group), with high normal (HNBP) – 20 children (group of comparison), with the labile arterial hypertension (LAH) – 20 and with the stable arterial hypertension (SAH) – 18 school students (the main groups). Values 0-3.0mg/L are taken for the reference CRP. The highest CRP level at children from stable AH (1.38±0.4mg/L) with a reliable difference in comparison with control is established (0.39±0.1mg/L, $p < 0.05$). Increased values of level are noted also at children with LAH (0.9±0.2mg/L, $p < 0.05$) and HNBP (0.78±0.11mg/L, $p < 0.05$) in relation to parameters of children with NBP. For group of children with HNAP the CRP level> 1.19mg/L had moderated (% Se=78.6) sensitivity and high specificity (% Sp=86.2). At children with AH this diagnostic test had higher sensitivity (%Se=87.5), and the specificity was identical (%Sp=86.2). Increase in indicators AH is associated with increase in the CRP level. Determination of the CRP level at children with AH has high diagnostic efficiency.

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Study of certain genetic polymorphisms of folate and homocysteine metabolism and neural tube defects

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Neural tube defects (NTDs) represent a major cause of infant mortality among congenital malformations whose pathogenesis remains poorly understood. In this study realized on an Algerian population, we investigated the frequency of many polymorphisms involved in the of folate/homocysteine metabolism, their possible contribution in the etiology of NTDs, as well as their influence on folate and homocysteine concentrations.

The study involved children with NTD, mothers who had a child with NTD and a control population. The mutations were determined by the PCR/RFLP method while the assay of the biochemical parameters was performed by chemiluminescence.

The analysis of genetic polymorphisms has shown that C677T polymorphism in MTHFR gene affects homocysteine metabolism in mothers of NTD cases leading to homocysteine concentration values higher in mothers with TT genotype of the C677T ($p < 0.05$).

For the polymorphism A2756G of the MTR gene, although, its association with NTD risk appears to be negative, It was found to decrease the RBC folate level strengthen the contribution of A2756G mutation in NTDs occurrence ($p <0.05$).

The major challenge remains translating study observations into a clinical setting such as gene screening in women before pregnancy, to identify predisposition and seek targeted prevention. Measurement of homocysteine concentration may also be a useful indicator of the need for pre-conception intervention. There is optimism that food is an element on which intervention possibilities exist.

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Assessing pediatric index of mortality 2 score and factors affecting outcome of patients admitted to pediatric intensive care unit in St. Paul's Hospital Millennium Medical College, Addis Ababa, Ethiopia, 2016 to 2019

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Background: Pediatric Index of Mortality2 (PIM2) is one of mortality scoring methods which can be used to predict mortality, to assess quality of care and to determine severity of illness in pediatric intensive care units (PICU). Despite PIM2 is regarded to be a better tool, its use has not been well validated in Ethiopia. The objective of this study was to evaluate the usefulness of PIM2 score in predicting mortality and to assess associated factors affecting mortality.

Method: A cross sectional study was conducted on 282 children admitted to PICU of St. Paul's Hospital. An adopted checklist was used to collect data from patient records from July 1-30, 2019. Data was entered and analyzed using SPSS version23. The predictive ability of PIM2 score for patient mortality was analyzed using Receiver Operative Characteristics curve and the corresponding sensitivity and specificity of the PIM2 model was also calculated. Multiple binary logistic regression was conducted and P-value <0.05 was considered statistically significant.

Results: A total of 282 patients were included in this study. The predicted mortality by PIM2 score was 11.8% (n=34) while the observed mortality was 39.7% (n=112).

Neurological diseases contributed to 33.0% mortality followed by cardiac 14.3% and renal diseases 13.4%. PIM2 score discriminated well between death and survival with area under receiver operating characteristic curve 0.86 (95%CI 0.82 to 0.91), with sensitivity of 75% and specificity 83.5%. Significant statistical association with mortality was seen in children who required mechanical ventilation Adjusted Odd's Ratio (AOR) 9.2, children with high risk diagnosis (AOR) 8.4 and in those with dilated pupils at admission (AOR) 9.4.

Conclusion: PIM2 model discriminated well between survivors and death at PICU and therefore is recommended for routine use in clinical practice. Mortality was more likely in children who required mechanical ventilation, have a high risk diagnosis and dilated pupils at admission.

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Factors influencing intention to obtain the HPV vaccine among a population of university women in Morocco

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Cervical cancer is a public health problem in the world with approximately 527 624 new cases and 265 672 deaths annually of which more than 86% occur within developing countries. In Morocco, cervical cancer is the second women's cancer after the breast cancer, with an estimated of 2258 new cases and 1076 deaths annually. The last decade has been marked by the introduction of a vaccine against this cancer. It is clear that the possibility of preventing cervical cancer through vaccination is an invaluable opportunity but also it is a difficult challenge. Young girls between the ages of 9 and 26 represent the target population for the HPV vaccine. As a result, the decision to vaccinate depends largely on two populations; young girls and their parents. The adoption of this vaccination by the population is dependent on several factors, in particular socio-economic. Thus, in this work we are interested in evaluating the knowledge and attitudes of university girls towards this vaccine. To this end, we conducted a multicenter survey with 1087 young woman in different regions of Morocco. The results show that 70.4% of participants are sensitized towards cervical cancer, 85% of whom said they have never heard of HPV. The rate of sensitization to HPV vaccination is about 7.8%. The rate of HPV vaccination coverage in our study population does not exceed 0.09%. Only one participant reported being vaccinated. The Analysis of the results showed, that the main barriers to the practice of this vaccine among participants sensitized towards HPV vaccine are as follows; the need for more information about the vaccine (40.5%), the fear of side effects (19%) and the need for a medical prescription (10.7%).

After sensitizing all participants towards HPV and its means of preventing, we recorded an acceptability rate of anti-HPV vaccination of about 68%. The main barriers reported by participants who expressed their refusal to practice HPV vaccination are: price (48.1%), fear of side effects (14.8%). In addition, age, family income, branch of study, educational level of parents and awareness of HPV vaccine and cervical cancer were found as a factors influencing the acceptability of this vaccine. Our study revealed a low level of awareness and knowledge about HPV and HPV vaccination. Similarly, we have focused on reticence to this vaccine in our study population. This situation will not help to improve the prevention of this cancer in our country, which requires the establishment of awareness actions towards the disease and its means of primary and secondary prevention. The cost of this vaccine is a universal barrier. As a result, the introduction of HPV vaccination into the national immunization program, or its reimbursement by social security, will encourage a large population to be vaccinated. In addition, efforts must be made to sensitize physicians on their axial role in raising awareness and prescribing this vaccine.

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Latissimus dorsi muscle flap in treating recurrent eventration of diaphragm

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Background: Diaphragmatic eventration-Permanent elevation of a hemi-diaphragm without defects in continuity. It is a rare anomaly, occurring in about 0.001-0.003% of live births (Urschel et al.,) It is associated with recurrent pneumonia, severe respiratory distress, ventilator dependency.

Case report: 11 month old child with recurrent respiratory tract infections requiring hospitalization. Hostory of full term normal vaginal delivery. Respiratory distress at birth, and was found to have left sided eventration of diaphragm. The child underwent laparotomy and plication repair on 8th day of life and was discharged home on 22nd day of life. However, the child developed recurrent episodes of cough and fever from one month post op.

Investigations: X Ray- elevated left dome of diaphragm Barium meal follow through- recurrence with stomach and splenic flexure occupying the eventration.

Operative procedure: Left dorsolateral incision, Left latissimus dorsi identified, dissected and safeguarded, Thoracotomy done in 5th ICS, Eventration of left lobe of diaphragm, Thin diaphragm/sac on the anteromedial aspect left dome of diaphragm, Diaphragm repaired by plication and fixation to the anterior chest wall, Diaphragm repair reinforced with left latissimus dorsi flap Incision closed in layers, wound dressed, Discharged on POD 22, with no complications.

Discussion: Neonates with large defects require prosthetic patch closure because of the paucity of native diaphragmatic tissue. As the child grows, patch separation can occur necessitating reoperation. Because of reliable blood supply and the innate strength, LD flap can support the diaphragm well and can prevent its eventration.

Conclusion: Diaphragmatic plication is a standard, well-described technique to treat diaphragmatic eventration. LD flap has been used in treating CDH, while its use in eventration is not well established. However, in our patient we performed a latissimus dorsi muscle flap to treat recurrent eventration of the left hemi diaphragm successfully.

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Limb salvage in an extensive and complicated vascular lesion in an infant

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Background: Extensive vascular lesions can endanger the life of a child by their virtue of consumptive coagulopathy or cardiac failure. A conservative surgical approach is difficult and can be life-threatening due to uncontrolled bleeding. We report successful limb salvage in an infant despite am extensive and infected arterio-venous malformation of upper limb, complicated by severe consumptive coagulopathy

Case: A term male, 3.2kgs, presented with a swelling over left arm detected antenatally. Antenatal scan- soft tissue swelling 10.8 x 6.8 x 5.4cm from left shoulder to elbow with heterogeneous cystic spaces and minimal vascularity. O/E- non compressible, no bruit heard.

Investigations: Doppler – hypoechoic lesion+ heterogeneous echotexture, numerous cystic spaces, weak color Doppler signal- S/O- Hemangioma MRI- lobulated soft tissue mass 17 x 16 x 9.5cm with an intensive enhancement, necrotic and hemorrhagic areas, involving flexor and extensor muscles of the arm. Supplied by branches of axillary and brachial arteries. Diagnosed as- Hemangioma, hence was started on propranolol 0.5 mg/kg/day. Presented at 2 months of age with severe anemia and consumptive coagulopathy refractory to medical therapy. At 4 months of age surgery became an imperative lifesaving mode of treatment.

Operative procedure: Planned for amputation/disarticulation, Axillary vessels identified and controlled, Tumor excised in toto, Flexor and extensor muscles preserved as much as possible, Radial nerve through the tumor was divided and re-anastomosed, Disarticulation was hence avoided, HPE suggestive of AVM, Arm and hand movements preserved at 10mth follow-up.

Discussion: Vascular malformations are usually present at birth; they expand secondary to trauma, sepsis or hormonal modulation. They are less likely to cause consumptive coagulopathy but can destroy, distort or cause hypertrophy of the adjacent bone. They may shunt the blood leading to cardiac failure. Most of the vascular tumors and a few of vascular malformations can be managed conservatively. The indication for surgery is functional or aesthetic. When medical management fails, surgery is unavoidable.

Conclusion: A conservative limb salvage surgery is possible even in case of an extensive vascular anomaly. A good vascular control is a must to prevent life threatening hemorrhage in a conservative surgery.

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Torsion of a splenule in a case of splenogonadal fusion mimicking strangulated inguinal hernia

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Background: Splenogonadal fusion (SGF) is a rare congenital anomaly which occurs due to an abnormal fusion of splenic tissue with the derivatives of the perimesonephros. It usually presents as an inguinal hernia, testicular mass or undescended testis. To the best of our knowledge, this is the first time, SGF presented as a strangulated inguinal hernia.

Case: A six-year-old boy with an acute painful left inguinal swelling since that morning. No fever, abdominal distension or vomiting. General examination- tachycardia. Groin exam- A tense, tender, irreducible left inguinal swelling with normal bilateral descended testes. RS, CVS, CNS- Normal.

Decision making: USG- A soft tissue swelling as the content of the hernia with reduced vascularity. DDx- ? Torsion of supernumerary testis

Intra operative findings: Left indirect inguinal hernia, No omentum/ bowel, but a purplish mass connected distally to upper pole of the left testis and proximally entering the peritoneum through a fibrous cord, Fibrous cords–360° torsion → early gangrenous changes, Fibrous cords excised from upper pole of testis distally and as high as possible proximally. Herniotomy done.

Histopathology report: Histopathology revealed a fibroelastic capsule covering a tissue comprised of red pulp, white pulp, dilated sinusoids; consistent with an ectopic spleen. There was no testicular tissue noted in the excised specimen, clinching the diagnosis of SGF, the continuous type.

Discussion: SGF is a rare entity - less than 200 cases reported till date.The etiology-unknown, but few theories-(a). Post-inflammatory adhesions between gonadal ridge and spleen (b). An abnormal retroperitoneal pathway allowing the communication between the gonad and spleen 3 (c). The gonadal tunica enveloping the developing spleen. Treatment of choice-Complete excision of the splenic tissue with preservation of gonad. Unfortunately, 37% of the reported cases underwent unnecessary orchidectomy as many surgeons were unfamiliar with SGF.

Conclusion: Splenogonadal fusion, although rare, can mimic a strangulated inguinal hernia, uncomplicated inguinal hernia, undescended testis, torsion testis or supernumerary testis. Every surgeon should be aware of this condition to avoid the dreaded morbidity of orchidectomy in this beginning condition.