

Rapid Genomic Sequencing in NICU Patients-Past, Present and Future

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Background: The effectiveness of targeted gene panels to improve the diagnosis and care of critically ill infants remains unknown. **Methods:** After developing RapSeq, a panel targeting 4,503 disease-causing genes, we selected candidate patients for testing in our neonatal and pediatric/cardiac intensive care units (NICU and PICU/CICU) based on specific criteria. Fifty infants and their parents were sequenced from October 2018 to March 2021. We assessed diagnostic yield, turnaround times, and clinical consequences; and we performed cost analysis of one case. **Results:** a firm diagnosis was made in 25/50 neonates (50%); thirteen had de novo variants, eleven were either compound heterozygote or homozygous, and one had a maternally inherited GNAS variant. Preliminary reports were generated by 9.6 days (mean); final reports after Sanger sequencing at 16.3 days (mean), with no discrepancies. In all positive infants, the diagnosis changed management. In two cases with congenital myasthenia, diagnosis and treatment occurred at 17 days with RapSeq vs. 7 months, and hospitalization costs were at least \$71,393 higher with conventional testing. **Conclusion:** This study shows that a gene panel that includes the majority of known disease-causing genes can rapidly identify a diagnosis in a large number of tested infants. Due to simpler deployment, easier variant interpretation, and lower costs, this approach might represent a valid alternative to exome and genome sequencing in the care of critically ill newborns.

Biography

Dr. Mao is the medical director of Molecular Genetics and Genomics in ARUP Laboratories and a professor of pathology at the University of Utah School of Medicine. She received her MD from Capital University of Medicine in Beijing and her MS in molecular pathology from Beijing Union Medical College. She is board certified by the American Board of Medical Genetics, with a subspecialty in clinical molecular genetics, and certified with the New York State Department of Health, with a subspecialty in genetic testing. Her research interests include the genotype-phenotype correlations in inborn errors of metabolism and genetic diseases in the RAS/MAPK pathway; she is also involved with implementing next-generation sequencing techniques into molecular diagnostics.

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Three kinds of caesarean sections: the foetal/neonatal perspective**Michel Odent***Primal Health Research Centre, UK*

From the "point of view" of the foetus/neonate, we provide reasons to contrast "birth without labour" (that is birth by pre-labour caesarean section) and all the other vaginal and abdominal modes of birth. From a great diversity of theoretical reasons, one can anticipate that babies born by pre-labour caesarean sections are different from the others. We also provide reasons to popularize the concepts of "in labour non-emergency caesarean sections" and "planned in-labour caesarean sections".

Biography

Michel Odent, MD studied medicine at Paris University. He has been in charge of the surgical unit and the maternity unit at the Pithiviers (France) state hospital (1962-1985). He is the author of the first article in the medical literature about the use of birthing pools (Lancet 1983), of the first article about the initiation of lactation during the hour following birth (1977), and of the first article applying the 'Gate Control Theory of Pain' to obstetrics (1975). He coined the term "hormone of love" when mentioning oxytocin. He created the Primal Health Research database (www.primalhealthresearch.com).

A 27-Year-Old Lebanese Man with Stomach Perforation and Regurgitation of a Beef Tapeworm (*Taenia saginata*): A Case Report and Review of the Literature.

Samer dbouk

Lebanese University, Lebanon

The global burden of *Taenia saginata* (T. Saginata), the beef tapeworm, includes economic loss, and its pathogenicity is considered mild. T. Saginata can infect the human definitive host when people ingest larval cysts from raw or undercooked beef, as cattle are the intermediate host. This report is of a case of gastric perforation and pneumoperitoneum with regurgitation of T. Saginata in a 27-year-old Lebanese man, and includes a review of previous cases of gastrointestinal perforation due to T. Saginata.

Case Report: We report a rare case of stomach perforation caused by T. Saginata, in which the tapeworm was subsequently expelled orally. A computerized tomography (CT) scan was done, revealing pneumoperitoneum and abdominal fluid, which was consistent with evidence of a perforated hollow viscus. Three days after exploratory laparoscopy, the patient vomited a 3-meter tapeworm and the diagnosis was subsequently made. On the fourth day, a CT scan of the abdomen with oral contrast was performed and showed no leakage. A clear fluid diet was started on the fifth day. The patient was discharged home on the seventh postoperative day in good condition. One week after the discharge, the patient was examined; he was in a good condition and symptoms were completely relieved 1 week after worm expulsion.

Conclusions: This report shows that in countries or societies where eating raw beef is common, a diagnosis of infestation with T. Saginata should be considered in patients who present with gastrointestinal symptoms.

Biography

Samer dbouk is a Faculty of Medicine and head of general surgery in Lebanese University, Lebanon

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Childhood tmj ankylosis: a surgical challenge**Naghm bazzi***Lebanese University, Lebanon*

Temporomandibular joint (TMJ) ankylosis is a rare and a debilitating condition. The major etiological factor is trauma in young age. This is due to greater potential of growth when compared with adults. The management of TMJ ankylosis is challenging due to the high incidence of recurrence and to the absence of a published consensus. We presented a long-term follow-up for a rare case of recurrent TMJ ankylosis post childhood trauma. Patient underwent multiple surgeries with autologous reconstruction, interposition arthroplasty, and coronoidectomy. Finally and after the development of skull base ankylosis, patient was treated with alloplastic reconstruction at adulthood. She was then followed up for 10 years post-op with excellent mouth opening. Thus, the treatment of recurrent TMJ ankylosis in children is still controversial with a debate about the applicability of TMJ prosthesis to prevent multiple procedures.

Biography

Naghm Bazzi has completed her medical Diploma at the age of 25 years from The Lebanese University, Faculty of Medical Sciences. She holds also a baccalaureate degree in the piano from the national conservatory of Lebanon. She has published more than 10 papers in peer-reviewed journals and has been serving as a reviewer in the journal of Cosmetic Dermatology.

Role of GNE gene specific p.V727M ethnic founder mutation in HIBM patients using various bioinformatics approaches

Vikas Sharma

Chaudhary Devi Lal University, India

Statement of the Problem: GNE gene-specific c.2179G>A(p.V727M) is a key alteration reported in Hereditary inclusion body myopathy(HIBM) patients and represents an ethnic founder mutation in the Indian/ Asian cohort. However, the underlying role of this mutation in pathogenesis remains largely unknown.

Methodology & Theoretical Orientation: In the present study, we aimed to access possible mechanisms of V727M mutation that could be leading to myopathy disease via various in-silico tools. Briefly, MutPred and PredictSNP tools were used to identify V727M induced functional and phenotypic changes. The vibrational entropy and enthalpic changes of flexible conformations of the mutation was studied using DynaMut that integrates normal mode approaches with graph-based distance matrix in the mutating residue environment. We also used simulation studies for studying V727M mutation on the structure of the GNE protein. Tools to study protein-protein interaction were also explored. Further, the available ChIP-seq data was analyzed to predict the possible interactions.

Findings: Our results propose that V727M mutation could induce deleterious effects or pathogenicity and affect the stability of GNE protein. Analysis of differential genes reported in the V727 mutant case suggests it can affect GNE protein interaction with various transcription factors.

Conclusion & Significance: We conclude that V727M mutation could alter the interaction of GNE with various transcription factor including MYC, thereby altering transcription of sialyltransferase(STs) and neuromuscular genes. Thus understanding these effects could pave the way for developing effective therapies against HIBM.

Biography

Dr. Vikas Sharma is working as a scientist in AIIMS-New Delhi, India. He has worked extensively in the field on Neuro-oncology. Most of his research work is focused on Genetic and epigenetic modification in GBM tumors of the brain. He has also reported epigenetic modification profiles in glioma using ChIP-sequencing approaches. Presently, he is exploring mutational effect in HIBM myopathy by studying ethnic founder mutations that are prevalent in HIBM patients in Indian Cohort.

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Comparing the Diagnostic Accuracy of Procalcitonin and C-Reactive Protein in Neonatal Sepsis: A Systematic Review**NagaSpurthy Reddy Anugu***California Institute of Behavioral Neurosciences & Psychology, USA*

Neonatal sepsis remains a significant diagnostic challenge in newborn care. It has the potential to be disastrous, but precise diagnosis is difficult. No biomarker has yet demonstrated sufficient diagnostic accuracy to rule out sepsis when clinical suspicion exists. As a result, neonates with suspected sepsis are treated with empiric antibiotics. These unnecessary antibiotics promote bacterial antibiotic resistance, raise economic costs, and alter the composition of the gut microbiota. This study aimed to determine the diagnostic accuracy of procalcitonin in the prompt diagnosis of neonatal sepsis. Articles were systematically screened in PubMed/MEDLINE, PubMed Central (PMC), and ScienceDirect, using keywords and Medical Subject Heading (MeSH) terms to identify the relevant articles. Additionally, one article from the Indian Journal of Applied Research was also used. Inclusion/exclusion criteria were applied post article screening via title and abstracts. Quality appraisal check was done using the Scale for the Assessment of Narrative Review Articles (SANRA) checklist, A Measurement Tool to Assess Systematic Reviews (AMSTAR) checklist, and Newcastle-Ottawa checklist. Six related articles were strictly reviewed. Procalcitonin is a useful biomarker in the early diagnosis of neonatal sepsis. Because procalcitonin has a better correlation with proven sepsis and is an early biomarker in diagnosing neonatal sepsis, it should be included in the overall sepsis evaluation. Future clinical trials on optimal cut-off levels of procalcitonin with shifting neonatal ages and its use in the post-op setting are needed.

Biography

NagaSpurthy is a Researcher at California Institute of Behavioral Neurosciences & Psychology, Fairfield, USA. She has done her under graduation from Medciti Institute of Medical Sciences, Telangana, India and was awarded with a gold medal for her merit. She is interested to further her research in neonatology, pediatric cardiology and adolescent medicine.

Challenges of Irradiation on Extrathoracic and Non-thoracic Organs in Portable Neonatal Chest Radiography: Do We Need Mandatory Protective Rules?

Sadeghi Majid

Gonabad University of Medical Sciences, Iran

Background: Chest X-ray (CXR) is known as the most common radiography used for adult and pediatric patients worldwide. Improper X-ray field collimation can result in excessive radiation dose on non-thoracic organs in chest radiographs.

Objectives: This study was to investigate X-ray field collimation quality in neonatal chest radiography.

Methods: A total of 213 chest radiographs of neonates from three hospitals were analyzed for collimation quality assessment in a retrospective study. Accordingly, ideal imaging field (IIF) and current imaging field (CIF) were initially defined. The margins of the IIF included acromioclavicular (AC) level to lower costal margin (i.e. top to bottom) and one centimeter beyond the broadest area of the chest on each side (that is, right to left). The CIF size was also defined as the square borders of collimators.

Results: The findings revealed that the area of the CIF was 1.65 _ 0.39 times to the ideal imaging field (IIF) for three hospitals, suggesting that collimation quality in neonatal chest radiographs was not accurate and it had defects. According to the results, acceptable collimation percentage (36.6%) in Hospital A was more than that in two other centers, and the given center also provided the lowest radiation due to the exposure of non-thoracic structures to primary beams.

Conclusions: It was concluded that training radiographers and using patient immobilization devices and stabilizers were important points that could reduce radiation exposure to non-thoracic organs in pediatric CXR.

Biography

I am Dr. Majid Sadeghi, a member of the faculty of Gonabad University of Medical Sciences. My research interests are in the fields of medical imaging dosimetry and radiation protection and imaging techniques.

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Clinical pharmacist's intervention and its acceptance rate in the Pediatric ward of Jimma University Medical Center, South West Ethiopia**Desalegn Feyissa Mechessa***Mizan-Tepi University, Ethiopia*

Background: Drug therapy problem is associated with morbidity, mortality, increased hospital stay and reduced quality of life. Moreover, pediatric patients are quite susceptible to drug therapy problems. Clinical pharmacist services had a positive impact on pediatric patient care.

Objective: To assess the prevalence of drug therapy problem, the activities of clinical pharmacists and its acceptance rate at pediatric ward of Jimma university medical center.

Method: A hospital-based cross-sectional study was conducted in the pediatric ward of Jimma University Medical Center from April 01 to June 30, 2018. The collected data were classified according to Pharmaceutical care network Europe version 5.0. Data were entered into Epi data version 4.0.2 and then exported to statistical software package version 21.0 for analysis. To identify predictors of drug therapy problems occurrence, multiple stepwise backward logistic regression analysis were done.

Results: From a total of 304 patients included in the study, nearly three fourth of them had at least on drug therapy problem per patient. A total of 356 pharmaceutical interventions were performed in the study setting. Dosing problem 112(31.46%) and drug choice problem 101(28.37%) were the predominantly identified drug therapy problem. The most important interventions provided was adherence and counseling (22.75%) followed by change of medication (13.76%). The acceptance rate of clinical pharmacist intervention was (79.49%). Type of admission (transferred) [AOR=1.85, 95% CI= (1.05-3.28)] and Number disease conditions [two diseases AOR=2.60, 95% CI= (1.25-5.39)], (three diseases AOR=4.26, 95% CI=(1.95-9.27)

Conclusions: Drug therapy problems were common in the pediatric ward of the study area. Pharmaceutical interventions performed were significant and contributed to therapeutic optimization and prevent actual drug therapy problems in pediatric inpatients.

Biography

Desalegn Feyissa Mechessa is Director/Prof/Assistant or Associate prof/Researcher/ Scientist /PhD Student in Mizan -Tepi University

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A frequent sign, an infrequent pathology. Kikuchi-Fujimoto Disease in a clinical case**Mariana Casullo***Castro Rendon Regional Hospital of Neuquén, Argentina*

Introduction: Kikuchi-Fujimoto disease was first described in Japan in 1973 by Kikuchi and Fujimoto. It is most often a benign and self-limited pathology, which is characterized by fever and adenopathies. It affects mostly young women and it is only very rarely observed in children. There has been only one case of a pediatric patient reported in Argentina.

Objectives: to observe, analyze and understand a rare, benign pathology, which generates anxiety and anguish due to its manifestation.

Case Description: Healthy twelve-year old female patient, with a medical history of cervical adenopathy at 3 years of age. This was assumed to be an inflammatory adenitis with focal necrosis of probable viral origin (diagnosed through lymph node biopsy). Currently, she seeks primary care due to mobile, non-tender lateral cervical adenomegalies, with a twenty-day evolution, which are all associated with leukopenia without neutropenia. She refers pain to the touch (intermittent fever, myalgia and livedo reticularis in lower limbs). Not responsive to antibiotics treatment. In subsequent controls, plateletpenia and a slight anemia are detected. Neck-ultrasound shows hypervascular adenomegalies, with a tendency to confluence and a loss of hilum. VIH, VHB, VHC, TOXO, CMV and E. BARR serologies come back negative, as do rheumatological antibodies and PPD. A smear and a bone marrow aspiration eliminate the possibility of an oncological disease. A biopsy is performed, which determines the existence of histiocytic necrotizing lymphadenitis. A histological comparison is performed with the previous sample, and found to match. Kikuchi-Fujimoto is diagnosed.

Discussion and conclusion: Prevalence of ECKF has not been correlated with gender in infancy. It can affect patients of all ages, including very young children. An infectious etiology has not been proven, and it is considered to be a hyper-immune reaction. Evolution is often benign, with a limited course, which resolves within two to six months. Relapses, such as the one observed in this patient, are infrequent and have been observed in only 4% of the cases.

The adenopathy must be extracted in its entirety for diagnostic purposes, since partial biopsies or punctures can render unreliable results

Biography

Casullo Mariana is studied at Castro Rendon Regional Hospital of Neuquén, Argentina

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Importance of breast feeding**Asma tauqeer***Shifa international hospital, Pakistan*

Breastmilk gives the baby a healthy start in life. It is the only food that baby needs for about the first 6 months. Breastmilk is always fresh, clean and the right temperature. Babies who are breastfed are also sick less often than babies who are not breastfed. Breast milk contains everything baby needs for the first 6 months of life, in all the right proportions. Its composition even changes according to the baby's changing needs especially during the first month of life. During the first days after birth, breasts produce a thick and yellowish fluid called colostrum. Colostrum provides high amount of immunoglobulin A as well as several other antibodies. Breast milk is loaded with antibodies that help the baby fight off viruses and bacteria, Breast feeding reduce risk for baby are following

- Middle ear infections
- Respiratory tract infections
- Gut infections
- Child hood leukemia
- Intestinal tissue damage

Breast feeding reduce risk for mother are following

- Its help the uterus in contraction
- Prevent from postpartum depression
- Help in birth spacing
- Weight losing
- Time consuming

This information is provided as general information only. Best efforts have been used to develop this information.

Biography

Asma tauqeer is a Neonatal nurse in Shifa international hospital, Pakistan