Jaslok Hospital & Research Centre, Mumbai

Research eBulletin

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From the Chairman's Desk

It is July 2016, and this year Jaslok completes forty-three years. On Founder's day, I am happy to report that Jaslok has made great progress. Research has played an important part in this process of moving forward. Many of our doctors are conducting research along with their practice. Their papers appear in prestigious medical journals and are then adopted and integrated in various medical procedures. In addition, every year we get more than hundred DNB students who join our institute and do research, because our research facility, they say, is the best.

Research is the effort not of a single person but of a group of people pursuing ideas with curiosity, inquisitiveness, openness and discipline. Many scientists rely on information and techniques borrowed from other scientists. However, one should also rely on intuition and think 'out of the box', in an unconventional way or from a different perspective, for new ideas. Research leads to innovation and innovation leads to newer and better treatment for patients.

Finally, patients reap the benefits of completed research and that is our goal and what really matters. I am happy to be associated with this Research eBulletin and wish it the very best.

Mr. M.K. Chanrai
Chairman, Jaslok Hospital and Research Centre

Editorial

Listening to Feedback

For a research journal to be meaningfully engaged in research, it should be open to receiving and acting upon feedback from its readers. Over the past year, we have received some helpful feedback from our colleagues at Jaslok and occasionally outside it as well. This feedback has related to matters such as inclusion or exclusion of articles, design, circulation and accessibility. The suggestions have helped us and we hope have improved the journal. We have incorporated most ideas including some we were initially skeptical of such as print copies. One would think that a printed eBulletin would be a contradiction in terms and maybe it is; but some among us (I confess my guilt) are more comfortable in the print world than in the virtual one and we navigate daily between these two realities. Thank you readers and please keep up the feedback.

In continuation with our Legends of Jaslok series, we are pleased to feature Dr. R.D. Lele who has been pivotally associated with our institution since its inception. I personally owe him an enormous debt because he inducted me into Jaslok, 27 years ago. Among his other roles, Dr. R.D. Lele has been one of the most effective Medical Directors the hospital has ever had. One of the key features of his administration was that all matters were usually cleared within 24 hours with a simple "yes" or "no". He hated the words "maybe" and "I'll see". Jaslok is what it is today because of legends like Dr. R.D. Lele.

Rajesh M. Parikh, M.D., D.P.M., D.N.B.
Director, Medical Research

Research News

Dr. Fali Poncha and Dr. Azad Irani were awarded the first prize in the case presentation "Post viral self-limited flaccid quadripareisis: 3 similar case reports" at the Bombay Neurosciences Association meeting held on 18th October 2015.

Abstracts

Current Clinical Practice Scenario of Osteoporosis Management in India
Jhaveri S, Upashani T, Bhadauria J, Biswas S, Patel K
Journal of Clinical Diagnosis and Research 2015;9:RC04-8.

BACKGROUND: Various osteoporosis guidelines are available for practice.
AIM: To understand the current clinical practice scenario from the perspective of Indian orthopaedicians, especially about the epidemiology, clinical manifestations, approach to diagnosis and management and patient compliance patterns to long term treatment.
MATERIALS AND METHODS: A pre-validated structured questionnaire containing questions (mostly objective, some open-ended) catering to various objectives of the study was circulated amongst orthopaedic surgeons across India by means of post/courier, after giving a brief overview of the study telephonically. Data was extracted from the completed questionnaires, and analysed using Microsoft Excel software.
RESULTS: The questionnaire was filled by a total of 84 orthopaedicians throughout India. The prevalence of osteoporosis in India according to the orthopaedic surgeons was 38.4% and there was a female preponderance. Most of the respondents felt out of every 100 osteoporosis patients in India, less than 20 patients are actually diagnosed and treated for osteoporosis. The most common initial presenting feature of established osteoporosis cases was general symptoms. Most respondents preferred Dual-energy X-ray absorptiometry (DEXA) as the initial investigation for the diagnosis of osteoporosis in a patient presenting with typical features. While most respondents preferred once-a-month oral over intravenous (IV) bisphosphonates, they agreed that IV administration had advantages such as lower gastrointestinal side effects and improved compliance. The average duration of therapy of oral bisphosphonates was the longest (27.04 months) among the other anti- osteoporosis therapies that they used. On an average, the patient compliance rate in osteoporosis management was around 64%. IV Zoledronic acid (ZA) and intranasal calcitonin were infrequently used than other anti- osteoporosis therapies. While concerns about cost and availability deterred more frequent usage, there was an agreement that if used regularly these two agents may improve compliance rates among patients.
CONCLUSION: Current clinical practice scenario of osteoporosis management in India largely adheres to various clinical practice guidelines for osteoporosis. Side effects and lengthy duration of therapy with bisphosphonates seem to be the main factors leading to a low patient compliance. Widespread popularization of once-yearly Zoledronic acid and intranasal calcitonin spray may improve patient compliance and reduce side effect incidence.

Autism and sleep disorders
Devnani PA, Hegde AU
Journal of Pediatric Neurosciences 2015;10:304-7.

"Autism Spectrum Disorders" (ASDs) are neurodevelopment disorders and are characterized by persistent impairments in reciprocal social interaction and communication. Sleep problems in ASD, are a prominent feature that have an impact on social interaction, day to day life, academic achievement, and have been correlated with increased maternal stress and parental sleep disruption. Polysomnography studies of ASD children showed most of their abnormalities related to rapid eye movement (REM) sleep which included decreased quantity, increased undifferentiated sleep, immature organization of eye movements into discrete bursts, decreased time in bed, total sleep time, REM sleep latency, and increased proportion of stage 1 sleep. Implementation of nonpharmacotherapeutic measures such as bedtime routines and sleep-wise approach is the mainstay of behavioral management. Treatment strategies along with limited regulated pharmacotherapy can help improve the quality of life in ASD children and have a beneficial impact on the family. PubMed search was performed for English language articles from January 1995 to January 2015. Following key words: Autism spectrum disorder, sleep disorders and autism, REM sleep and autism, cognitive behavioral therapy, sleep-wise approach, melatonin and ASD were used. Only articles reporting primary data relevant to the above questions were included.

Pure Cerebellar Ataxia with Homozygous Mutations in the PNPLA6 Gene
Wiethoff S, Bettencourt C, Paudel R, Madon P, Liu YT, Hersheson J, Wadia N, Desai J, Houlden H
Cerebellum. 2016 DOI 10.1007/s12311-016-0769-x.

Autosomal-recessive cerebellar ataxias (ARCA) are clinically and genetically heterogeneous conditions primarily affecting the cerebellum. Mutations in the PNPLA6 gene have been identified as the cause of hereditary spastic paraplegia and complex forms of ataxia associated with retinal and endocrine manifestations in a field where the genotype-phenotype correlations are rapidly expanding. We identified two cousins from a consanguineous family belonging to a large Zoroastrian (Parsi) family residing in Mumbai, India, who presented with pure cerebellar ataxia without chorioretinal dystrophy or hypogonadotropic hypogonadism. We used a combined approach of clinical characterisation, homozygosity mapping, whole-exome and Sanger sequencing to identify the genetic defect in this family. The phenotype in the family was pure cerebellar ataxia. Homozygosity mapping revealed one large region of shared homozygosity at chromosome 19p13 between affected individuals. Within this region, whole-exome sequencing of the index case identified two novel homozygous missense variants in the PNPLA6 gene at c.3847G>A (p.V1283M) and c.3929A>T (p.D1310V) in exon 32. Both segregated perfectly with the disease in this large family, with only the two affected cousins being homozygous. We identified for the first time PNPLA6 mutations associated with pure cerebellar ataxia in a large autosomal-recessive Parsi kindred. Previous mutations in this gene have been associated with a more complex phenotype but the results here suggest an extension of the associated disease spectrum.

Wegener's Granulomatosis: A report of two Cases with different clinical and laboratory features

Ram H Malkani , Rahul Dixit , Maninder Singh Setia

Indian Journal of Basic and Applied Medical Research 2015; 4:109-12.

We present two cases of Wegener's granulomatosis (WG) with systemic involvement. Our first case, a 50 year old female, presented with painful vesicles on the left side of neck and chest. She was diagnosed as post-primary tuberculosis due to persistent cough and a nodular opacity in the left lower lobe of the lung. A wedge biopsy of the lung tissue did not show any mycobacteria, and an open biopsy showed granulomatous inflammation with necrosis. The serology for cytoplasmic- antineutrophil cytoplasmic antibodies (c-ANCA) and perinuclear – antineutrophil cytoplasmic antibodies (p-ANCA) was negative and renal parameters were normal. Our second case, a 50 year old male, presented with fever, cough with expectoration, and blood stained nasal discharge for the past 15 days with no cutaneous lesions. The renal biopsy showed cresentic glomerulonephritis and serum tested positive for c – ANCA. Both the cases were diagnosed as WG and managed using corticosteroids and/or cytotoxic medications. These cases represent the different clinical and laboratory features of limited and classical forms of WG respectively, and though the second case did not require extensive investigations, the first case posed a diagnostic challenge since one of the common differential diagnosis of nodular lung involvement with persistent cough in developing countries is tuberculosis.

Case-based discussion: an unusual manifestation of diaphragmatic hernia mimicking pneumothorax in an adult male

Vyas PK, Godbole C, Bindroo SK, Mathur RS, Akula B, Doctor N

International Journal of Emergency Medicine 2016;9:11.

Diaphragmatic hernia is an important cause of emergency hospital admission associated with significant morbidity. It usually results from congenital defect or rupture in the diaphragm due to trauma. Prompt and appropriate diagnosis is necessary in patients with this condition, as surgical intervention by either abdominal or thoracic approach may be necessary. Here, we report a case of left-sided diaphragmatic hernia presenting with sudden onset of breathlessness, respiratory distress and left-sided chest pain radiating to the abdomen, mimicking pneumothorax, treated successfully with surgical intervention.

Hypokalemic paralysis secondary to tenofovir induced Fanconi syndrome

Ramteke VV, Deshpande RV, Srivastava O, Wagh A

Indian Journal of Sexually Transmitted Diseases 2015;36:198-200.

Tenofovir induced fanconi syndrome (FS) presenting as hypokalemic paralysis is an extremely rare complication in patients on anti-retroviral therapy. We report a 50-year-old male with acquired immunodeficiency syndrome on tenofovir-based anti-retroviral therapy who presented with acute onset quadriparesis. On evaluation, he was found to have hypokalemia with hypophosphatemia, glucosuria and proteinuria suggesting FS. He regained normal power in limbs over next 12 h following correction of hypokalemia. Ours would be the second reported case in India.

The Indian consensus guidance on stroke prevention in atrial fibrillation: An emphasis on practical use of nonvitamin K oral anticoagulants

Dalal J, Bhave A, Oomman A, Vora A, Saxena A, Kahali D, Poncha F, Gambhir DS, Chaudhuri JR, Sinha N, Ray S, Iyengar SS, Banerjee S, Kaul U

Indian Heart Journal 2015;67 Suppl 2:S13-34.

The last ten years have seen rapid strides in the evolution of nonvitamin K oral anticoagulants (NOACs) for stroke prevention in patients with atrial fibrillation (AF). For the preparation of this consensus, a comprehensive literature search was performed and data on available trials, subpopulation analyses, and case reports were analyzed. This Indian consensus document intends to provide guidance on selecting the right NOAC for the right patients by formulating expert opinions based on the available trials and Asian/Indian subpopulation analyses of these trials. A section has been dedicated to the current evidence of NOACs in the Asian population. Practical suggestions have been formulated in the following clinical situations: (i) Dose recommendations of the NOACs in different clinical scenarios; (ii) NOACs in patients with rheumatic heart disease (RHD); (iii) Monitoring anticoagulant effect of the NOACs; (iv) Overdose of NOACs; (v) Antidotes to NOACs; (vi) Treatment of hypertrophic cardiomyopathy (HCM) with AF using NOACs; (vii) NOACs dose in elderly, (viii) Switching between NOACs and vitamin K antagonists (VKA); (ix) Cardioversion or ablation in NOAC-treated patients; (x) Planned/emergency surgical interventions in patients currently on NOACs; (xi) Management of bleeding complications of NOACs; (xii) Management of acute coronary syndrome (ACS) in AF with NOACs;(xiii) Management of acute ischemic stroke while on NOACs.

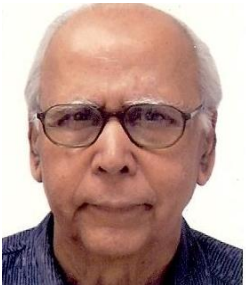
Challenges in Type 1 diabetes management in South East Asia: Descriptive situational assessment

Kesavadev J, Sadikot SM, Saboo B, Shrestha D, Jawad F, Azad K, Wijesuriya MA, Latt TS, Kalra S

Indian Journal of Endocrinology and Metabolism 2014;18:600-7.

Treatment of type 1 diabetes is a challenging issue in South East Asia. Unlike in the developed countries, patients have to procure insulin, glucometer strips and other treatment facilities from their own pockets. Coupled with poor resources are the difficulties with diagnosis, insulin initiation, insulin storage, marital and emotional challenges. Being a disease affecting only a minority of people, it is largely ignored by the governments and policy makers. Comprehensive diagnostic, treatment and team based educational facilities are available only in the speciality diabetes centers in the private sector whereas majority of the subjects with type 1 diabetes are from a poor socio-economic background. Unlike in the Western world, being known as a diabetes patient is a social sigma and poses huge emotional burden living with the disease and getting married. Even with best of the resources, long-term treatment of type 1diabetes still remains a huge challenge across the globe. In this review, authors from India, Pakistan, Nepal, Sri Lanka, Myanmar and Bangladesh detail the country-specific challenges and discuss the possible solutions.

Legends of Jaslok Hospital



Dr. Ramchandra Dattatraya Lele is called the father of Nuclear Medicine in India and one of its leading international academicians. Born on 16th January 1928, he finished school at the age of 14 and MBBS at 21. Inspired by Mahatma Gandhi's exhortation to serve in the villages, Dr. Lele did voluntary rural medical service for 5 years before proceeding to study at the London School of Hygiene and Tropical Medicine in 1955. He was awarded the MRCP (Edinburgh) within six months and the MRCP (London) next year.

Dr. R.D. Lele got exposure to a cyclotron and at the Hammersmith Hospital in 1966, he obtained a Fellowship in Nuclear Medicine in Canada. On his return, he was appointed Professor of Medicine at the G.M.C and J. J. Hospitals (1968-1972) and Dean (1972-73).

He was invited to join as Chief Physician and Director of Nuclear Medicine at Jaslok at its inception in July 1973, where he created the first full-fledged hospital based Nuclear Medicine Department and RIA in the country. His son, Dr. Vikram Lele, joined the department in 1988. In 1996, the Indo American society felicitated them as father and son achieving eminence in the same field.

Dr. Lele has authored 10 books and over 100 publications in indexed journals. He has given over 150 invited orations in 120 cities in India. He has been awarded the Padma Bhushan, the Dhanvantari Award and the Homi Bhabha Life Time Achievement Award, amongst others. He is actively involved in research such as the validation of Ayurvedic drugs using mechanism based screening and radiolabeling with C-14 and Tritium in small animals and the efficacy of 10 Medhya Rasayan in a transgenic mouse model of Alzheimer's disease. His most exciting project is the creation of aptamers for the in vivo imaging of Tuberculosis and various aspects of its treatment.

Dr. R.D. Lele has brought honour to India and to the Jaslok Hospital & Research Centre making him one of our living legends.

Research Event

Research Workshop: 'Protocol Writing' by Dr. Maninder Setia, Consultant Epidemiologist on 19/04/2016 for DNB students.

Editorial Board