

**Abstracts of Papers Presented At The 154<sup>th</sup> Research Meeting of The Medical Research Centre of Bombay Hospital Trust on Monday 8<sup>th</sup> October 2007, Convener Dr. HL Dhar**

1. Molecular Studies of The Spinal Muscular Atrophy Gene Locus

*MA Rao, RS Dastur, PS Gaitonde, JJ Nadkarni*

Spinal Muscular Atrophy (SMA) is an autosomal recessive neuromuscular disorder caused by the loss of anterior horn cells of spinal cord leading to progressive muscle weakness and atrophy. This motor neuron degenerative disease is the leading genetic cause of infant mortality. The three types of SMA present with diverse symptoms and differ in age of onset, mode of inheritance, distribution of muscle weakness and progression of symptoms.

SMA of all types is associated with homozygous mutation in SMN1 (Survival motor neuron1) gene located at the telomeric end of chromosome region 5q13. Along with SMN1 other genes present on this locus are NAIP gene (Neuronal apoptosis inhibitory protein) and p441 gene. A large inverted repeat at the centromeric region of 5q13 consists of homologous genes SMN2, YNAIP and p44c. The majority of full-length functional protein is produced by SMN1. Hence the absence of SMN1 results in SMA.

The role of other genes of the SMA locus in disease severity is not yet clearly understood. Hence it is interesting to study the correlation between the deletion pattern of these genes and the disease severity.

2. Unusual Presentation of Intramedullary Astrocytoma

*Rajiv Kumar, CE Deopujari, Rajan Shah, Nimit Gupta*

A 26 yr old businessman presented with complains of difficulty in swallowing for 10 days, vomiting off and on for 5 days, fever off and on for 5 days. He consulted Physician, ENT Surgeon. Laryngoscopy was normal. Barium swallow was normal, except there was residual barium in oral cavity. CT Scan abdomen was normal. Later patient consulted Neurophysician who advised MRI brain.

On examination patient was conscious, oriented. Speech had nasal twang. Palatal movement was present, but pharyngeal movement was depressed. Gag reflex was depressed. Motor system was normal. He had decreased pinprick, temperature sensation over C3-7 in cape like distribution. JPS was impaired in both lower limbs. Reflexes were normal. Plantar were flexor.

MRI brain and spine showed medium sized, ovoid shaped, strongly enhancing intramedullary lesion at D5-7 associated with syringobulbia and extensive syringohydromyelia from cervicomedullary junction to D11.

He underwent D5-8 laminectomy with near total excision of the lesion. He had immediate deterioration in motor power but gradually he improved. His swallowing also improved within a week.

His histopathology comes out to be intramedullary astrocytoma.

3. Comparative Analysis Between 1p and 19q co Deletion and Extent of Surgical Resection as Markers of Survival in Patients with Oligodendroglial Tumours of the Brain

*Sandeep Inchanalkar, A Dank, S Joshi, N Sharma, KE Turel*

The present study is a retrospective and prospective analysis of 32 cases of pathologically proven oligodendroglial tumours with a comparison between co-deletion of 1p and 19q and extent of surgical resection as markers of survival in the patients with this particular pathology. Despite unfavourable results of 1p and 19q codeletion studies, the survival rates are comparable suggesting that factors like extent of resection of the tumour, adjuvant radiotherapy, tumour biology and tumour location play an important role in the prognosis of these patients.

The main goal of the study was to investigate the efficacy of extent of surgical excision and to compare it with 1p and 19q codeletion as a prognostic marker, it can be concluded that there are still many other unknown factors which play an important role and these factors need to be searched for by further investigations.

## **Abstracts of Papers Presented At The 155<sup>th</sup> Research Meeting of The Medical Research Centre of Bombay Hospital Trust on Monday 10<sup>th</sup> December 2007, Convener Dr. HL Dhar**

### **1. Hypercalcaemic Crisis : A Case Study of Three Cases of Vitamin D and Calcium Intoxication**

***Bhakti Desai, Anand Gokani***

Severe hypercalcaemia is potentially life-threatening complication of several diseases. Most commonly it is caused by cancers that enhance bone resorption. Though rare, Vitamin D intoxication is an important cause of hypercalcaemia. Massive doses of ergocalciferol continue to be prescribed inappropriately or without adequate supervision and the dangers to this therapy are still not appreciated. Impaired renal calcium excretion resulting from a combination of volume contraction and calcium-induced renal injury (nephrocalcinosis) plays a critical role in the genesis and aggravation of hypercalcaemia. Hypervitaminosis-D is a known cause for morbidity and mortality. The potential hazards of Vitamin D intoxication are also well known.

We studied three unusual cases of hypercalcaemia. An 80 yr old male, 67 yr old female and 74 yr old female, who were admitted to the hospital with simple complaints of nausea, vomiting and generalized weakness. On investigating them thoroughly, they were found to have severe hypercalcaemia with normal parathormone levels. Detailed history revealed that the first patient received 18 injections of Vitamin D, second case received 3 injections of Vitamin D while the third case had an excessive intake of oral calcium supplements as a part of treatment of Osteoporosis.

Life-threatening hypercalcaemia was treated with isotonic saline infusion followed by diuretics. Vitamin D and calcium supplements were discontinued and diet containing low calcium was advised. Bone resorption was reduced by using Bisphosphonates. Calcitonin which has more rapid hypocalcaemic effect than bisphosphonates was administered. Rapid control over hypercalcaemia was achieved. Acute renal failure which was due hypercalcaemia depressing the glomerular filtration rate, rapidly disappeared with the control of serum calcium. On a series of follow up the patients calcium levels were found to be normal. This case report is to sensitize about overcorrection of osteoporosis with calcium and vitamin D supplementation leading to hypercalcaemia. A vigorous emphasis on the use of Vitamin D and calcium as of other vitamins, implies several risk and must be prescribed only when needed and under strict medical supervision.

## 2. Thyroid Storm : An Atypical Presentation

*Bhakti Desai, Alpna Shukla*

Thyroid storm, an extreme exacerbation of the hyperthyroid state, is a rare but life-threatening emergency. Because of the potentially high mortality rate, early diagnosis and treatment of thyroid storm is of the utmost importance. It typically occurs in patients with untreated or partially treated thyrotoxicosis who experience a precipitating event such as surgery, infection, or trauma. Thyroid storm must be recognized and treated on clinical grounds alone, as laboratory confirmation often cannot be obtained in a timely manner. Patients typically appear markedly hypermetabolic with high fevers, tachycardia, nausea and vomiting, tremulousness, agitation, and psychosis. Late in the progression of disease patients may become stuporous or comatose with hypotension. Thyroid storm is a rare disorder. Approximately 1-2% of patients with hyperthyroidism progress to thyroid storm.

A 63 yr old female, non-diabetic, non-hypertensive was admitted to intensive care unit with complaints of loss of appetite, loose motions and intermittent fever. Within few hours patient became drowsy, tachycardic and tachypnoeic hence put on ventilator. On investigating atypical pneumonia profile was positive for legionella and parainfluenza, she also had severe hypoalbuminaemia, low TSH with normal T3 and T4 levels. In view of low albumin, free hormone levels were done which revealed high free thyroxine, triiodothyronine, and low thyroid stimulating hormone. Antithyroglobulin antibody, antimicrosome antibody were also high. With the presumptive diagnosis of thyroid crisis induced by pneumonia, collosol iodine, neomercazole were started. Steroids and antibiotics were continued for aggressive management of infection. Clinical improvement of patient within 48-72 hrs of treatment.

Eventually patient was extubated. Thyroid hormones were normalized. Patient was discharged on neomercazole. On subsequent follow up patient had controlled thyroid functions. The intention of this case report is to point to the yet possible occurrence of thyroid crisis, which is now-a-days extremely rare owing to appropriate management of hyperthyroidism. Nevertheless may the disease failed to recognized on time and therapy is introduced too late, along with other unfavourable factors, such as acute infection, the disease can still occur sporadically. Although the mortality rate has been drastically lowered it is still high i.e. about 7% therefore these patients should be treated in intensive care unit.

## 3. Assessment of Case Sheets of Patients Admitted in ICCU

*Sonali Pandloskar, HL Dhar*

**Aim :** To find out Major Disorders and their outcome including cause of death.

**Methods :** Retrospective review of medical records of patients from 3<sup>rd</sup> floor ICCU, BH with various diagnoses from 1<sup>st</sup> Jan to 30<sup>th</sup> April 2006. A total of 162 medical records were reviewed. Their Clinical history, Age, Sex, Hospital stay, symptoms at the time of admission were collected. The entire patient's past and present surgical intervention and their biochemical markers were reviewed.

**Result :** Patients were divided into groups on the basis of their diagnosis as : coronary artery disease (CAD) (n=105), Rheumatic heart disease (n=4) and group 5 (n=30) and Deaths (n=23). The patients in Group 5 were those who were admitted in ICCU with various diseases. Male preponderance was seen in all the groups. Patients with CAD were older (mean age 60.85). The most common symptoms were chest pain, orthopnoea, DOE and breathlessness. The most common risk factors were hypertension (41.18%) and diabetes (35.29%) in CAD patients. The mean cholesterol (178.28), Serum Creatinine (1.42), and Fasting sugar (214). PTCA with stent to LAD were done in 50% of patients with CAD.

Deaths occurred in 14.19% patients. The main cause of death was acute renal failure followed by cardiac arrest.

**Conclusion** : ICCU in BH manages critically ill patients with fewer patients dying.

## **Abstracts of Paper Presented at The 156<sup>th</sup> Research Meeting of the Medical Research Centre of Bombay Hospital Trust on Monday 14<sup>th</sup> January 2008, Convener Dr. HL Dhar**

### 1. Management of Corneal Epithelial Ingrowth – A Rare Complication of Lasik

*Sandeep Kataria, Jitendra Pandey*

**Purpose** : To report the results of our surgical technique in 4 eyes of corneal epithelial in growth following lasik.

**Method** : Four eyes of 2 patients were diagnosed to have clinically significant epithelial in growth under the lasik flap. All four eyes underwent surgical debridement with flap suturing. Refractive outcome and recurrence incidence was recorded as outcome measures. Alternative treatment options have been discussed.

**Results** : All cases had a better visual outcome as compared to pre-operative refractive error and visual status. Recurrence was reported as lower than as reported in the literature.

**Conclusion** : The incidence of clinically significant epithelial ingrowth as a complication of lasik is reported to be as 1.71%. Progressive Epithelial ingrowth is associated with permanent visual loss if not treated in time. The surgical treatment involves relieving the flap and debridement. The recurrence of epithelial ingrowth in literature is reported to be as high as 44% to 60%. Many adjunctive methods like tissue glue bandage contact lens or debridement alone have been suggested to reduce the recurrence. We report lower recurrence rate with our technique of flap suturing.

### 2. Endoscopic Transnasal Optic Nerve Decompression

*Neeraj Sharma, Nishit Shah*

We present a study of optic nerve injury managed by endoscopic transnasal decompression of the nerve. This route is associated with much less morbidity than the intracranial route and offers the advantages of decompressing the optic nerve without having to retract the brain as in intracranial decompression. There is no external scar, hospital stay is much less and if there is any CSF leak it can be repaired. All the decisions in the management are taken with close consultation with ophthalmologist, neurosurgeons and the patient's relatives. For decompression to be effective patients are to be chosen very carefully with particular attention to the duration of injury and the onset of diminished vision. The imaging should also show obvious fracture of the optic canal on CT scan, swelling or haematoma or compression of the optic nerve on MR. The degree of residual vision after injury is also important for determining the prognosis. All the cases also receive high dose steroids pre and post operatively.

### 3. A Rare Case of Post Traumatic Cholesteatoma

*Neeraj Sharma, Anand Shah*

Post-traumatic cholesteatoma is a rare condition that may present years after the original injury. Cholesteatoma can develop as a late complication of fracture of the temporal bone. The otologist must be wary of it since the growth of the cholesteatoma resulting from a temporal bone fracture can be undetected for years allowing for invasive and extensive growth. The aetiology may be implantation of epithelium into the middle ear or mastoid cavity through the fracture line or ear drum perforation. A history of head injury, delayed onset and large size are the characteristic of post-traumatic cholesteatoma and it usually develops in well-pneumatized temporal bone. The potentially devastating acute issues of temporal bone fracture including conductive and sensorineural hearing, facial paralysis, cerebrospinal fluid leakage and vestibular injury are well known. Contrasting with these acute complications is the delayed presentation of an implantation cholesteatoma in the middle ear and mastoid.

We report one case of post-traumatic extensive middle ear cholesteatoma, extending into the middle cranial fossa, at the site of a previous temporal bone fracture presenting with a long interval between the injury and appearance of clinical symptoms.

### 4. Small Fenestra Stapedotomy Using Teflon Piston with Vein Graft Interposition

*Seemab Khan, Anand Shah*

Osteosclerosis is a hereditary disease of the embryonic bone derived from the otic capsule that causes fixation of the ossicles with resultant conductive or mixed hearing loss. It can be associated with significant disability due to hearing impairment. Advances in the treatment of otosclerosis in recent years have centred on technical modifications of the surgical procedure itself.

The surgical technique of small fenestra stapedotomy is well established. The following study reports our experiences with small fenestra stapedotomy using Teflon Piston with vein graft interposition performed in last few years. The results especially immediate and long-term hearing outcomes are evaluated and complications encountered using this technique have been analysed. We hope that identifying the problem area using this technique would form a guideline for future training.

### 5. Re-Do Surgery in ROP

*Jitendra Pandey, Karobi Lahiri*

This is a retrospective study of 6 eyes reoperated for ROP of which 4 eyes were operated for stage V ROP and 2 eyes for stage IV-buckling.

The criteria for suggesting resurgery was that the funnel should have opened at the primary surgery with reattachment where no break were formed. In stage IV to reattach the detached retina surgical outcome showed good results (66.66%). 2 eyes went physical (44.44%). Surgery primary and resurgery will be demonstrated.