JAWAHARLAL INSTITUTE OF POST GRADUATE MEDICAL EDUCATION AND RESEARCH (JIPMER)
PUDUCHERRY- 605 006

(An Institution of National Importance under the Ministry of Health and Family Welfare, Government of India)

TELEMEDICINE SERVICES

National Medical College Network (NMCN)
Regional Resource Centre (RRC) JIPMER

No. JIP/TELE-CGR/VC/2019/12/06 Date: 07/12/2019

To
The HOD
Department of Medicine
JIPMER

Sub: Telemedicine – Video Conferencing CGR Session between JIPMER and SGPGIMS, Lucknow on 7th December 2019 at 8:00 AM to 9:00 AM - Reg.

This is to inform you that there will be Video Conferencing Session will be held.

Date/Time : 07/12/2019 (Saturday) at 8:00 AM to 9:00 AM

Topic: 'Von Hippel-Lindau (VHL) disease as a prototype of familial cancer'

Summary: Familial cancer syndromes are cancers which are found to run in families. Most such disorders are due to mutations which are inherited from generation to generation in an autosomal dominant fashion, and precisely should be designated as "Hereditary cancer". Here, we describe the clinical and molecular data of 69 patients with suspected VHL or having VHL associated tumours. Sanger sequencing of coding sequences and conserved splice sites of VHL gene was done in all patients. Multiplex Ligation-dependent Probe Amplification (MLPA) of VHL gene to detect large deletions/duplications was performed for 18 patients with no pathogenic sequence variations.
Among tumor types at presentation pheochromocytoma was seen in 49 % (34/69), hemangioblastoma in 30 % (21/69), renal cell carcinoma in 7 % (5/69). Rest had other tumors like paraganglioma, endolymphatic sac papillary tumors,cerebellar astrocytoma and pancreatic cyst) . Seven patients had more than one tumor at the time of diagnosis. Pathogenic variations in VHL gene were identified in 31probands by Sanger sequencing; 18 were missense, 2 nonsense and 2 small indels. Family history was present in 7 cases in all of whom mutation was detected. A heterozygous deletion of exon 3 was detected by MLPA in one patient among 18 patients for whom MLPA was done. Overall, the molecular etiology could be confirmed in 46% cases (32/69). In addition, 11 families opted for pre-symptomatic mutation testing.

Between : SGPGIMS, Lucknow – KEM, Mumbai – AIIMS, Delhi – NEIGRIHMS, Shillong – IMS, BHU, Varanasi – TMC, Trivandrum - IGMC Shimla, – JIPMER, Puducherry.

Venue : Telemedicine Center, Department of Plastic Surgery, 4th Floor, SSB, JIPMER.

Contact : Mr. Mohamed Ishaq (M-9751590265) / Ms Kavitha (M-9789114987)

Kindly inform Faculty and Residents to attend.

Thanking you,

Yours Sincerely,

Prof Dr Ravi Kumar Chittoria
Head of IT Wing & Telemedicine