Registrars' Prize in Clinical Neuroscience

Director: Dr. Karen O'Connell

Minerva Room, RDS, Dublin

November 11th, 2022

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08.30 – 08.50 *Registration, tea and coffee*

08.50 – 09.00 **Welcome and Introduction**
Dr. Karen O'Connell.

INTERESTING CASE REPORTS
10 minutes: 8 minutes + 2 minutes questions

Session I

09.00 Adult Onset Alexander Disease – phenotype, genotype and radiological findings.
Bradley M, Ryan AM.

09.11 Cystic Medial Degeneration – a rare intracranial vasculopathy
Durcan R, Fullam S, Farrell M, McGuigan C.

09.22 Targeted antiepileptic treatment in genetic early infantile epileptic encephalopathy
Fisher A, Webb D, Byrne S, O'Regan M.

09.33 The diagnostic importance of respiratory chain analysis on muscle: a case series.
Fullam S, Alexander M, Farrell M, Beausang A, Cryan J, Murphy SM.

09.44 The obfuscated F wave: fasciculations and pain in a 68-year-old man.
Gilligan M, Widdess Walsh P, McQuillan R, Connolly S.

09.55 Cerebellar ataxia in a 35 year old female
Hamilton T, O'Connor G.

10.06 'Brothers'
Joyce E, Elamin M.

10.17 Duelling etiologies: Longitudinally extensive spinal cord lesion mimicking spinal cord infarct with simultaneous positive Lyme serology and amphiphysin antibody.
Kalaszi M, Donlon E, Wan Ahmad M, Sheikh Mohamed A, Boers P.

10.28 Typical Congenital Nemaline Myopathy diagnosed in the seventh decade.
Killian O, McGrath E.

10.40 – 11.10 Tea and Coffee Break.
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Session II

INTERESTING CASE REPORTS
10 minutes: 8 minutes + 2 minutes questions

11.10 A case of refractory focal epilepsy evaluated with Stereo EEG and treated with a targeted frontal lobe resection.
Klaus SP, Widdess-Walsh P, Sweeney K, O'Brien D, Doherty CP.

11.21 Do not underestimate neurological symptoms in postoperative neurosurgical patients.
Koustais S, Delanty N, Ewins K, Crockett M, McAnena L, Javadpour M.

11.32 A de novo mutation in IRF2BPL gene leading to adult onset neurodevelopmental disorder with regression, abnormal movement, loss of speech and seizures (NEDAMSS).
McElligott L, Lockhart A, Mulrooney M, McGovern E, Costelloe L.

11.43 POL3A-related late-onset ataxia: clinical clues from reverse phenotyping.
McKenna MC, Lockhart A, Doherty CP, Costigan D, Brett FM, Langan Y, Bede P, Hutchinson S.

11.54 Be "SMART" when taking a history.
Mulkerrin G, Friel O, Faul C, Farrell T, Looby S, Delanty N, McGovern E

12.05 "Confounding Contractures" A Non-Lethal Presentation of a Lethal Disorder
Mullane G, Smyth S.

12.16 Behavioural variant dementia with Lewy Bodies - an overlooked syndrome?
O'Connor A, Lyons S, Finnegan M, Walsh J, O'Dowd ST.

12.45 – 13.45 Lunch

Session III

INTERESTING CASE REPORTS
10 minutes: 8 minutes + 2 minutes questions

13.45 A cautionary tale.
Olszewska DA, Bhowmick SS, Lang AE.

13.56 Concurrent diagnoses of Tuberous Sclerosis and Multiple Sclerosis
Quigley S, Doherty CP, Cronin S, Kearney H.

14.07 A Forgotten Point: A Case of CHANTER syndrome with Excellent Neurological Recovery.
Reynolds A, Sheehan J, O'Donnell M, O'Donohoe R, Tubridy N.

14.18 An unusual case of epilepsy and anarthria in a patient living with HIV in Zambia.
Smyth M, Asukile M, Saylor D.

14.29 "In Hindsight: A Rare Cause for Cognitive Decline Diagnosed Post Mortem".
Tacheva A, Guilfoyle P, McGovern E, Costelloe L, Chalissery A.

14.40 Return of the Germ.
Tallon E, McGuigan C, Devadass A, O'Riordan S.

14.51 Imitating Sweetly: An Epilepsy Diagnosis Reconsidered
Troy E, Kgosidialwa O, Costello DJ.

15.10 – 15.25 Afternoon Tea and Coffee Break

Session IV

RESEARCH PRESENTATIONS
12 minutes: 10 minutes + 2 minutes questions

15.25 Urbach Wiethe Disease: Uncovering the role of an ECM1 mutation in BBB dysfunction.
Brennan D, Greene C, Togher Z, Hutchinson S, Doherty CP, Campbell M.

15.38 Preclinical interrogation of novel Immunotherapy treatment strategies in Glioblastoma (GBM) using a novel clinically relevant disease model
Clerkin J, Connora K, White K, Sweeney K, Shiels L, Van Brussel T, Arijs I, Lambrechts D, Maher SG, Marignole L, Prehna J, O'Brien D, Byrne AT.

15.51 "Somatic variants as a cause of drug-resistant epilepsy including mesial temporal lobe epilepsy with hippocampal sclerosis"
Doyle MG, Benson K, Carton R, Kearney H, Sweeney K, O'Brien D, Henshall D, Cavalleri GL, Delanty M.

16.04 Social cognition in cervical dystonia
Rafee S, Herings R, Hutchinson M, Reilly R.

16.17 The contribution of thalamic pathology to the clinical manifestations of frontotemporal dementia phenotypes.
McKenna MC, Hutchinson S, Bede P.

16.30 An Audit of Impact and Cost of Genetic Testing in Beaumont Hospital.
Tallon E, Moloney P, Delanty N.

16.45 Judges Deliberation

17.00 Announcement of Awards

17.15 Close of Meeting