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Meeting Summary

62nd Meeting of the French Society of Neuropathology

Meeting Abstracts

December 4th, 2020

The French Society of Neuropathology was created in 1989, succeeding the French Club of Neuropathology set up in 1965. The Society organizes two scientific meetings per year.

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Plu I, Evrard B, Duchesne M, Regnault B, Pérot P, Chrétien D, Eloit M, Seilhean D (2020) Etiological diagnosis of a plasma cell encephalitis by next generation sequencing. Free Neuropathol 1, 34: 2

Meeting Abstract
[Short communications]

Etiological diagnosis of a plasma cell encephalitis by next generation sequencing

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A 59-year-old man, working as a forester, developed a fever and temporal headache associated with cranial nerves deficits. Etiological investigations including multiplex PCR and the search for autoantibodies remained negative. His condition quickly worsened leading to coma and death after 2 months.

Neuropathology revealed lymphoplasmacytic infiltrates in the leptomeninges, cranial nerve roots and around the vessels in cerebral gray and white matter. Perivascular cuffs were associated with astrocytic gliosis and microglial activation. Immunochemistry against known viruses was negative. The New Generation DNA Sequencing (NGS) without *a priori* has led to the identification of a type 1 Lyssavirus of the European bat (EBLV1), the reservoir of which is an insectivorous bat. Only two cases of transmission to human have been reported to date. This case of zoonosis illustrates the interspecies transmission of an unusual virus leading to a potential emerging disease. NGS coupled with neuropathology provide a valuable tool for identifying new causes of encephalitis.

Teyssou E, Muratet F, del Mar Amador M, Gyorgy B, Guegan J, Marie Y, Meininger V, Salachas F, Millecamps S, Seilhean D (2020) A novel mutation in SOD1 causing unusual neuropathological findings. Free Neuropathol 1, 34: 3

Meeting Abstract
[Short communications]

A novel mutation in SOD1 causing unusual neuropathological findings

Elisa Teyssou[1*], François Muratet[1*], Maria del Mar Amador[1,2], Beata Gyorgy[1], Justine Guegan[1], Yannick Marie[1], Vincent Meininger[2,3], François Salachas[1,2], Stéphanie Millecamps[1], Danielle Seilhean[1,4]

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A 63-year-old man with no personal or family history developed amyotrophic lateral sclerosis (ALS) at age 63, fatal in 17 months. In the anterior horns of the spinal cord, neuronal loss was moderate. Cystatin C immunohistochemistry (IHC) showed numerous Bunina bodies. IHC of ubiquitin, p62 or TDP43, did not show skein-like inclusions. Swollen neurons in clusters, were labelled with anti- ubiquitin, SOD-1 and phosphorylated neurofilaments. They were different from inclusions usually observed in SOD-1 mutations.

Whole exome sequencing analysis identified a novel SOD1 mutation c.164C>T, p.Thr55Ileu, confirmed by Sanger sequencing. No other rare variant was identified in any other ALS-related genes. Association of neuropathology and whole exome sequencing can provide a useful tool for the identification of unknown forms of the disease, better understanding of the physiopathology and lead to new therapeutic targets.

Siegert E, Dittmayer C, Schneider U, Preuße C, Goebel HH, Stenzel W (2020) Myositis in scleroderma – Capillary pathology is fundamental. Free Neuropathol 1, 34: 4

Meeting Abstract
[Short communications]

Myositis in scleroderma – Capillary pathology is fundamental

Elise Siegert, Carsten Dittmayer, Udo Schneider, Corinna Preuße, Hans-Hilmar Goebel, Werner Stenzel

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Systemic sclerosis is a chronic disease of connective tissues characterized by fibosis, vasculopathy and autoimmunity. Affected patients show signs of the skin, internal organs and sometimes overlap myositis. The vasculopathy is considered obliterative but the pathogenic basis is not known to date.

We are presenting light-microscopic and ultrastructural as well as clinical data of 18 patients suffering form scleroderma and myositis. We have applied a new electron microscopical technique which we call large scale electron microscopy allowing a 'pan and zoom' approach similar to 'google earth' viewing.

This analysis allows to study >1000 capillaries of patients and controls, highlighting reduplications of basement.

We show that this type of ultrastructural changes is membranes, endothelial activation and proliferation of pericytes specific for a subtype of scleromyositis and discus possible pathogenic mechanisms.

Boluda S, Wallon D, Rovelet-Lecrux A, Campion D, Nicolas G, Duyckaerts C (2020) Neuropathological variability of four tauopathy cases with *MAPT* microduplication. Free Neuropathol 1, 34: 5

Meeting Abstract
[Short communications]

Neuropathological variability of four tauopathy cases with *MAPT* microduplication

Susana Boluda[1,2], David Wallon[3,4,5], Anne Rovelet-Lecrux[3,4,6], Dominique Campion[3,4,6], Gaël Nicolas[3,4,6], Charles Duyckaerts[1,2]

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We report the neuropathology of four *MAPT* duplication carriers, a rare chromosomal rearrangement involving the 17q21.31 chromosomal region that causes an early onset dementia (EOD) clinically mimicking Alzheimer disease or an atypical extrapyramidal syndrome, as recently described. They were three males and one female with ages ranging between 37 and 57 years. They were all tauopathies with a variability in the morphology and distribution of the aggregates. The cases either mimicked Pick disease with neuronal globular aggregates similar to Pick bodies and pathology involving predominantly the cortical and limbic regions and subcortical nuclei or they resembled progressive supranuclear palsy with tufted astrocytes with major involvement of the brain stem and subcortical nuclei. The tau isoform expression also varied from either only 3R, only 4R or a mixed 3R/4R expression.

In conclusion, *MAPT* duplication may lead to a tauopathy spectrum with a range of 3R and 4R expression.

Seilhean D, Mokhtari K, Plu I, Boluda-Casas S, Mathon B, Cao A, Hervé D, Mégarbane B, Bielle F, Levavasseur E, Malet I, Marot S, El Hachimi H, Marty S, Prigent A, Duyckaerts C, Potier MC, Haïk S, Delatour B, Marcelin AG (2020) Multiple cerebral angiopathy in SARS-COV-2 infection. Free Neuropathol 1, 34: 6

Meeting Abstract
[Short communications]

Multiple cerebral angiopathy in SARS-COV-2 infection

Danielle Seilhean[1,2], Karima Mokhtari[1], Isabelle Plu[1], Susana Boluda-Casas[1,2], Bertrand Mathon[3], Albert Cao[4], Dominique Hervé[5], Bruno Mégarbane[6], Franck Bielle[1,2], Etienne Levavasseur[2], Isabelle Malet[7], Stéphane Marot[7], Hamid El Hachimi[2], Serge Marty[2], Annick Prigent[2], Charles Duyckaerts[1,2], Marie-Claude Potier[2], Stéphane Haïk[1,2], Benoît Delatour[2], Anne-Geneviève Marcelin[7]

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SARS-CoV-2 is the cause of a pandemic characterized by its severity in elderly subjects or those presenting metabolic or vascular risk factors. Brain damage, although relatively rare, is often fatal. From four cases (one autopsy and three brain biopsies) we analyzed the involvement of small cerebral arteries associated with white matter lesions, without signs of vasculitis or encephalitis. Viral RNA has not been detected in the brain. SARS-CoV2 spike protein (S) has been detected in the Golgi apparatus of endothelial cells, colocalized with a host protease. Our observations suggest the possibility of hematogenous neuroinvasion. The interaction of a small amount of protein S with endogenous proteases is thought to be able to disrupt the permeability of brain endothelial cells causing vascular damage. This result could provide therapeutic avenues to prevent or cure severe brain forms in patients at risk.

Bourhis A, Peyre M, Bielle F (2020) A rare tumor of the peripheral nerve mimicking a schwannoma. Free Neuropathol 1, 34: 7

Meeting Abstract
[Short communications]

A rare tumor of the peripheral nerve mimicking a schwannoma

Amélie Bourhis[1], Matthieu Peyre[2,3], Franck Bielle[2,4]

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A 38 year-old man presented with pain of the left forearm for five years. He had a history of old HIV infection and several past opportunistic infections. The pain was paroxysmal, favored by pronation and was increasing for two years. MRI evidenced a tumor nodule on the path of the posterior interosseous nerve mimicking a schwannoma. Intracapsular tumor resection stopped the pain. Microscopic examination showed a tumor proliferation of fascicled eosinophilic fusiform cells. Immunostaining showed the expression of the muscular markers smooth muscle actin and desmin, and the absence of expression of SOX10. In situ hybridization for EBER RNA of EBV was positive in tumor cells. We diagnosed an EBV-associated leiomyoma of the immunocompromised, which was located in the peripheral nerve, a localization not previously reported. We discuss the tumorigenesis of this rare neoplasm.

Eloit M (2020) Virus spillover from animal reservoirs and vectors to human detection using agnostic tools. Free Neuropathol 1, 34: 8

Meeting Abstract
[Conferences]

Virus spillover from animal reservoirs and vectors to human detection using agnostic tools

Marc Eloit

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OIE Collaborating Centre for Detection and Identification in Humans of Emerging Animal Pathogens, Pasteur Institute, Paris, France

Professor of Virology, Veterinary School of Maisons Alfort, France

Viruses from wildlife including arboviruses vectored by mosquitoes have caused dramatic outbreaks over the last 25 years. On the one hand, new sequencing capacities have deeply affected the means by which new viruses are discovered, leading to the identification of ever more viruses in animal reservoirs and arthropods. On the other hand, it is worth noting that the continually growing knowledge regarding these viruses does not per se serve to identify potential human or animal threats. Our laboratory aims to fill this gap by identifying, in selected ecosystems, whether as yet unknown, unexpected or neglected mosquito or wildlife viruses are responsible for frequent but sub-clinical or mild infection in humans highly exposed to wild life/arthropods, and thus represent good candidates for global spreading. It combines high throughput screening of viruses in animal/arthropods linked to antibody screening in healthy exposed human populations and virus search in patients presenting with severe diseases of unknown etiology. I will show our methodology and some results

Ludes B (2020) Migration of Siberian populations of the past: contribution of a multi-genetic markers approach. Free Neuropathol 1, 34: 9

Meeting Abstract
[Conferences]

Migration of Siberian populations of the past: contribution of a multi-genetic markers approach

Bertrand Ludes

University of Paris-CNRS, FRE 2029

Head of the Forensic Institute of Paris (Institut Médico-Légal de Paris)

Molecular markers of nuclear DNA (autosomes and Y chromosome) and mitochondrial DNA from human samples help clarifying the migrations of the first peoples of the Eurasian steppes. We analyzed samples dating from the middle of the 2nd millennium BC to 4th century AD and originating from the Krasnoyarsk region (South Siberia). We confirmed that, during the Bronze and Iron Ages, southern Siberia was a region dominated by European peoples, suggesting an eastward migration of the Kurgan peoples across the Russo-Kazak steppe. The results further showed that at that time, the inhabitants of southern Siberia must have had fair eyes, skin and hair.