eral nerves, mainly around myelinated fibers, with demyelination and axonal loss, consistent with CIDP. The spinal cord revealed severe neuronal loss in the anterior horn, axonal loss in the corticospinal tract, and large numbers of phagocytes in the anterior and lateral tracts, indicative of ALS. Whether demyelinating polyneuropathy was coincident with ALS or was a cause or consequence of motor neuron degeneration in these patients remains to be elucidated. This unusual combination may provide an important clue in elucidating the pathogenesis of ALS in some patients.

一项对 3 例肌萎缩侧索硬化症及类似 CIDP 的多发性神经病变患者的研究

本文报道 3 例符合慢性炎症性脱髓鞘性多发性神经根神经病变（CIDP）临床及实验室诊断标准的患者，患者使用免疫抑制剂治疗无效，并逐渐进展为肌萎缩侧索硬化症(ALS)。患者死亡前的平均病程为 23 个月(13～38)，2 例患者有 ALS 家族史，但无 SOD1 基因突变，1 例患者的尸检结果显示，腰部神经根、远端及近端周围神经的单核细胞神经内膜浸润，表现为有髓鞘神经纤维的脱髓鞘及轴突缺失，与 CIDP 的表现一致。脊髓表现为前角神经元缺失、皮质脊髓束轴突缺失以及前束与侧束的大量吞噬细胞，提示为 ALS。在上述患者中，脱髓鞘性多发性神经病变与 ALS 同时发生还是与运动神经元变性互为因果尚有待于阐明，上述两种疾病同时发生异常少见，可能为解释部分患者 ALS 的发病机制提供重要线索。

0596. Novel LAMP－2 gene mutation and successful treatment with heart transplantation in a large family with Danon disease

Echaniz-Laguna A. / Mohr M. / Epailly E. et al. [Dr. A. Echaniz-Laguna, Department de neurologie, Hôpital Civil, 1 Place de l'Hôpital, 67091 Strasbourg, France] －MUSCLE NERVE 2006, 33/3 (393－397)

Lysosome-associated membrane protein－2 deficiency (LAMP2 deficiency), or Danon disease, is a rare X-linked lysosomal disease characterized by cardiomyopathy, vacuolar myopathy, and mental retardation. Less than 20 families with mutations of the Lamp－2 gene have been reported. We describe a family from Sardinia with eight affected patients (4 females and 4 males) and a novel mutation in exon 2 of the Lamp－2 gene (c. 102_103delAG). Females developed isolated cardiomyopathy in adulthood, whereas males presented with cardiomyopathy, myopathy, and mental retardation before the age of 20 years. Cardiomyopathy was lethal in three females in their 40s and in three males before the age 20 years. One patient was successfully treated by heart transplantation with more than 5－year follow-up. This study demonstrates that Danon disease is a frequently fatal condition that is potentially treatable with heart transplantation.

在一个 Danon 病大家族中的新 LAMP－2 基因突变与成功的心脏移植治疗

溶酶体相关膜蛋白 2(LAMP2) 缺乏或 Danon 病是一种罕见的 X 连锁溶酶体疾病，以心肌病、空泡肌病及精神发育迟缓为特征。已对不到 20 个具有 Lamp－2 基因突变的家族作过报道，本文对来自撒丁岛的 1 个家族进行描述，其中 8 例家族成员(4 例女性和 4 例男性)发病，并发现有 Lamp－2 基因 2 号外显子 (c. 102_103delAG) 的新突变。女性患者在成年后进展为心肌病，而男性患者则在20岁之前出现心肌病、肌病和精神发育迟缓。3 例女性患者在其 40 多岁时因心肌病而死亡，3 例男性患者则在其 20岁前因心肌病而死亡，1 例患者经心脏移植后成功治愈，术后随访超过 5 年。本研究证实 Danon 病是一种常见的致死性疾病，可通过心脏移植而治愈。

0597. Pure neural leprosy: Diagnostic value of the polymerase chain reaction

Da Cunha F. M. B. / Werneck M. C. M. / Scola R. H. / Werneck L. C. [Dr. L. C. Werneck, Servic. o de Doenc. as Neuromuscu-lares/Neurologia, Hospital de Clnicas, UniversidadeFederal do Paran′a, Rua Gal. Carneiro, 181, 80060－900 Curitiba, Brazil] －MUSCLE NERVE 2006, 33/3 (409－414)

Pure neural leprosy (PNL) is often difficult to diagnose when acid-fast bacilli (AFB) cannot be detected. We undertook the present study to evaluate use of the polymerase chain reaction (PCR) in diagnosing PNL. Fifty-eight patients (41 men and 17 women) suspected of pure neural leprosy (PNL) were examined. Patients were classified as borderline tuberculoid (BT, 40 cases) and polar tuberculoid (TT, 18 cases) types. Nerve biopsy was performed and was positive for AFB in 20 patients (all BT patients), i. e., 34. 5% of total cases. DNA was extracted from the nerve biopsy samples and amplified using PCR for a specific repeated sequence of DNA from Mycobacterium leprae. PCR analysis was positive in the nerve samples from 29 patients (50%), 27 of the BT type, and 2 of the TT type patients. Further, PCR analysis was positive in 14 of 38 cases that were negative for AFB by