Title: A large Chinese kindred with familial ALS without SOD1 mutation

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**NUS-09 A prevalence study of epilepsy in HKSAR, China**

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**Introduction:** Epilepsy is a common disorder. Epidemiological data is crucial for physicians and health care administrators for taking care patients with epilepsy. In this communication, we report the the epidemiology data of the Hong Kong West (HKW) region of Hong Kong Special Administrative Region (HKSAR).

**Methods:** With the implementation of clustering system in our clinic since 1996, the epilepsy clinic of Queen Mary Hospital is managing a vast majority of adult patients (> 15 years old) with chronic seizure disorders resided in the HKW region where hosted an adult population of 475,900. Seven hundred and thirty-six patients [female 42.9%, male 57.1%, mean 40.8, SD 13.6] with epilepsy were recruited. All patients underwent EEG examination and each subject was independently assessed by two epileptologists for diagnosis and classified according to ILAE recommendations.

**Results:** The prevalence rate of active epilepsy at or above 15 years of age was 1.54 in 1,000 at the prevalent date (January 1, 2002). 285 (38.8%) had idiopathic epilepsy syndromes, 100 (13.6%) had cryptogenic, and 285 (38.8%) had a remote symptomatic etiology. Seizure type was partial in 408 patients (55.4%) and generalized in 285 (38.8%). Thirty-one patients (4.2%) had positive family history. Interestingly, common idiopathic generalized epilepsy syndromes like juvenile myoclonic epilepsy (0.68%) and childhood absence epilepsy (0.95%) were uncommonly encountered.

**Conclusions:** In summary, this clinic based epidemiological study provides crucial data for epilepsy service development and research in HKSAR. Further hospital based or, preferably, door-to-door population based epidemiological study is indicated to ascertain the population based epidemiologic data for epilepsy with patients resided in HKW of HKSAR.

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**NUS-10 A large Chinese kindred with familial ALS without SOD1 mutation**

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**Introduction:** Amyotrophic lateral sclerosis (ALS) is a lethal neurodegenerative disorder with progressive muscle weakness and wasting. About 10% of familial ALS (FALS) cases have SOD1 mutations.

**Methods:** We ascertained a large Chinese kindred with autosomal dominant FALS. All consented family members underwent detailed clinical, electrophysiological and, if indicated, pathological examination.

**Result:** A total of 16 members (12 living) were classified as affected. Eighteen living (eight affected and ten unaffected) members were available for study. Historical review of the clinical features, and clinical, electrophysiological and pathological assessments showed a phenotypic spectrum in this family, from typical ALS (N=8), who rapidly deteriorated with progressive muscle wasting, weakness and respiratory failure to a group (N=8) with very slowly progressive predominantly lower motor neuron lesion. The clinical features of a member from each group appear to lie between the two ends of this spectrum. Mutation screening for SOD1 mutation were all negative in all 5 exons.

**Conclusion:** The interesting phenotypic spectrum observed in this family is distinctive although similar FALS families with heterogenous phenotypes were reported within the same pedigree. Further studies to identify the causative gene/s in this ALS kindred are indicated.

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