

Frontier in Neurology Research Beyond Neurodegenerative Diseases

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As a guest editor for this special issue of Neurology Diseases in the *Chinese Medical Journal* (English Edition), I am pleased to announce a completion of 19 articles (13 research articles, 4 review papers and 2 case reports) which cover a spectrum of research frontier in neurology diseases for publication in this issue. These articles provide an update on the state of clinical and basic science research in movement disorders (Huntington disease, Parkinson's disease, spinocerebellar ataxias (SCAs), and Wilson's disease), amyotrophic lateral sclerosis, cerebral vascular disease, epilepsy, infection/immunology diseases, and rare genetic disorders (mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes [MELAS] and facioscapulohumeral muscular dystrophy [FSHD]). While substantial progress in our understanding of these disorders has been achieved, development of accurate diagnosis procedures and precise treatments remains an unrealized goal. Among them, 5 papers are related to Parkinson's disease, which show the promise for translational research in identifying genetic, biochemical, electrophysiological and imaging biomarkers, aimed to help better diagnosis and management of this disease.

Beyond these common neurological diseases and neurodegenerative disorders, there are three interesting papers discussing the recent progress in diagnosing rare genetic diseases MELAS,^[1] FSHD^[2] and SCAs.^[3] The first article^[1] is a review to present recent progress in diagnosing MELAS that is a progressive, multisystem affected mitochondrial disease associated with a number of candidate genes. This disease has unpredictable presentations and clinical course, and it can be commonly misdiagnosed as encephalitis, cerebral infarction, or brain

neoplasms, which can result in a delay or mistreatment of disease. Thus, the early diagnosis is important for the management of MELAS. Recent new findings in clinical features, blood biochemistry, neuroimaging, muscle biopsy and genetics would lead to early and accurate diagnosis of the disease.

The second interesting article^[2] is a research report of clinical and genetic analysis of FSHD in 178 individuals from 136 unrelated families in Chinese population. FSHD is a common autosomal dominant muscular disorder with remarkable intra- and inter-familial clinical heterogeneity. This disorder is caused by contraction of D4Z4 array on chromosome 4q35. Until now, the specific biological mechanism involved in the clinical variability has yet to be resolved, and the complicated genotype-phenotype relationship among different ethnic population remains a controversial subject. Using the p13E-11, 4qA and 4qB probes after pulsed field gel electrophoresis separation and southern blotting, and a Modified Medical Research Council scale score and a 10-grade FSHD clinical severity scale (CSS) core, the authors observed a roughly inverted correlation between age-corrected CSS and short fragment size in 159 symptomatic patients. Compared to male patients, a significant higher proportion of females in both asymptomatic carriers and severe patients showed larger variation in the size of short fragment. High incidence of asymptomatic carriers (minimally affected individuals) in family members with the contracted allele was in our study. Their results suggest that there are multi-factors synergistically modulating the clinical phenotype.

The third article^[3] is also a research paper reporting 27 SCA patients from 10 Chinese families that have been identified with CAG nucleotide repeat mutations in SCA1, SCA3/MJD, SCA7 and SCA8 genes. Although several similar studies

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have been reported previously, this paper provided a detailed analysis not only on clinical and genetic characteristics, but also the radiological features in those patients with SCA. The authors found that SCA3/MJD was the most common subtype in Han population in China, and the ratio of the pontine tegmentum and the posterior fossa area is negatively correlated with the number of CAG repeats. The authors also documented that the disease duration was positively correlated with International Cooperative Ataxia Rating Scale score, and the number of CAG repeats was negatively correlated with the age of onset.

From those publications, we can learn that the diagnosis of these diseases requires a complex synthesis of clinical, biochemical, histological, radiology and genetic investigations. Despite their phenotypic diversity, the improvement in understanding of the disease mechanisms and the recent development of new laboratory techniques have created rapid changes to adopt the precise medical practice in the near future.

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