

# MINUTES FOR THE 4<sup>th</sup> PAN-ASIAN CONSORTIUM FOR TREATMENT AND RESEARCH IN ALS (PACTALS)

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The 4<sup>th</sup> PACTALS meeting was held on 7 December 2016 at Room Liffey 2a, Dublin Convention Centre, Ireland in conjunction with the 27<sup>th</sup> International Symposium on ALS/MND.

The meeting began at 1pm and was presided over by Matthew Kiernan and Nortina Shahrizaila.

1. Introduction – Matthew Kiernan (Australia) and Nortina Shahrizaila (Malaysia)
  - a) Welcome remarks – Matthew Kiernan
    - i. Discussed the strength of an Asia-Pacific collaborative network
    - ii. Current meeting provides a platform for researchers in the region to put forward collaborative research proposals to the group that can address some of the uncertainties in ALS
  - b) Update on the progress of the PACTALS Registry – Nortina Shahrizaila
    - i. Discussed the proposal presented at the last meeting in Kuala Lumpur by Paul Talman, current chair of the Australian Registry
    - ii. Web-based data entry: an overarching Ethics approval allows de-identified patient data from member countries to be hosted on one platform
    - iii. Individual member countries will require local Ethics approval prior to including patient information into Registry
    - iv. PACTALS Registry will be governed by the PACTALS Steering Committee comprising of representatives from the different contributing Member countries
  - c) Next PACTALS meeting to be held in conjunction with the forthcoming World Congress of Neurology in Kyoto, Japan (September 16-21, 2017)
    - i. Local organising Faculty: Gen Sobue, Ryuji Kaji and Satoshi Kuwabara
    - ii. Tentative plans are for a half-day Scientific Meeting
    - iii. Further updates will be disseminated to Members via email and PACTALS website
2. Alliance Support Grants - Carol Birks, Chair of International Alliance of ALS/MND Associations
  - a) Explained the role of International Alliance of ALS/MND Associations
  - b) Discussed the success of the program in supporting member countries that have limited resources to partake in International meetings as well as offers seed money to initiate the development of Patient Organisations with the support of the Alliance
3. The natural history of ALS among different populations - Dongsheng Fan (China)
  - a) Background: Current research largely arise from Western populations but Pan-Asia represents >50% of world population with diverse ethnic, social and cultural background
  - b) Aims and objectives:
    - i. To describe the natural history of ALS in Pan-Asian countries
    - ii. Identify prognostic markers
    - iii. Comparative studies between countries and between regions

- iv. Elucidate environmental, ethnic, social and cultural factors that are unique to the region
    - v. To accelerate the establishment of the multi-centre database for PACTALS
  - c) Advantages of natural history study in PACTALS include the large and growing number of patients, multi-ethnic populations, developed and developing countries representing diverse existing resources, social and environmental diversity
  - d) Consensus on standardising the tools for data acquisition at the outset, in particular cognitive studies in the region.
- 2. Identification of De Novo Variants by Trio-based Whole Exome Sequencing and Functional Analysis of Candidate Genes in Asia-Pacific Patients with Sporadic ALS - Seung Hyun Kim (South Korea)
  - a) Described the existing genetic profile of Korean ALS patients – SOD1 remains the most common and c9orf72 mutation is not seen
  - b) Proposed ALS TRIO study to investigate potential *de novo* mutations in patients from the PACTALS cohort
    - i. Include young-onset ALS patients and their parents – trio-base whole exome sequencing to identify potential *de novo* mutations
    - ii. Depending on results, functional analysis based on skin fibroblasts to elucidate potentials pathogenic role in motor neurone cell death
- 3. Japanese Consortium for ALS (JaCALS) - Naoki Atsuta (Japan)
  - a) Introduced the structure and role of JaCALS registry
    - i. To investigate the longitudinal courses of Japanese patients with ALS
    - ii. To create a genomic gene repository linked to the longitudinal clinical information
    - iii. To search for clinical factors that affect the progression and prognosis of ALS
    - iv. To search for genetic factors that affect the pathogenesis of ALS
  - b) Discussed the wealth of data gleaned from JaCALS registry
    - i. The number of ALS patients are increasing
    - ii. Annual prevalence in Japan: 9.9/100000 per year; incidence 2.2/100000
    - iii. Age of onset is significant in survival and decline of patients
    - iv. Projecting and validating the long-term outcome of trial drugs (eg ederavone)
    - v. Comprehensive analysis of ALS-related genes and discovering genes associated with ALS
- 4. Closing remarks – Matthew Kiernan
  - Significant contributions from each Speaker and an important start to establishing much-needed collaborations in the region