Cerebrospinal Fluid Research

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Oral Presentation

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Neuroradiological criteria of human L1CAM syndrome – report of 24 human L1CAM mutations including 17 noble mutations and clinical evaluation

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from 48th Annual Meeting of the Society for Research into Hydrocephalus and Spina Bifida
Dublin, Ireland, 23–26 June 2004
Published: 23 December 2004

Cerebrospinal Fluid Research 2004, **1**(Suppl 1):S35 doi:10.1186/1743-8454-1-S1-S35

This article is available from: http://www.cerebrospinalfluidresearch.com/content/1/S1/S35

X-linked hydrocephalus (XLH), MASA syndrome and certain forms of X-linked spastic paraplegia (SPG1) and X-linked agenesis of corpus callosum (ACC) are now known to be due to mutations in the gene for the neural cell adhesion molecule L1CAM and reclassified as human L1 syndrome. The purpose of this study was to perform the nation-wide mutation research of L1 gene in Japan and evaluate the clinical futures. We defined the criteria for L1 CAM gene mutation analysis in two categories. The first was the patients with hydrocephalus with X-linked inheritance, and the second included the sporadic cases with clinical and neuro-radiological characteristics of XLH or MASA syndrome.

Ninety one samples from 55 families are selected for L1 CAM mutation research. We identified 23 types of LICAM gene mutations in 24 families with X-linked hydrocephalus (XLH) and MASA syndrome. Seventeen noble mutations were included in 23 mutations. Of 24 mutations, there were 15 in coding region (class 1–1, class 2–5, class 3–9) and there were 9 mutations in non-coding region. Adducted thumbs, hypoplasia of corpus callosum are seen in all patients. The patients with L1ED mutations showed severe retardation with severe hydrocephalus, patient with L1CD mutations showed moderate retardation without ventricular dilatation. Rippled ventricular wall after shunting, localized hypoplasia of the anterior vermis or total hypoplasia, enlarged quadrigeminal plate and large masa intermedia are characteristic neuroradiological signs in human L1ED (extracellular) mutations. This study further confirms the importance of the L1CD functional regions for axon tract development and the L1ED functional regions for ventricular dilatation in humans.