Clinical Genetics

AB118. Association between clinical phenotypes and neurodevelopmental outcomes in pediatric patients with neurofibromatosis type 1 at Siriraj hospital

Chatchai Thamrongajariyakul[1], Achara Sathienkijkanchai[2]

[1]Department of Pediatrics, [2]Division of Medical Genetics, Department of Pediatrics, Faculty of Medicine Siriraj Hospital, Mahidol University, Bangkok, Thailand

Background: Neurofibromatosis type 1 (NF1) is characterized by multiple café-au-lait spots, skinfold freckling, cutaneous neurofibromas, and Lisch nodules. Additional signs and symptoms of NF1 include macrocephaly, skeletal abnormalities, hypertension, and learning disabilities. This study has objective to analyze the association between clinical phenotypes and neurodevelopmental outcomes in pediatric patients diagnosed with NF1 from 2005-2015 at Siriraj Hospital. The neurodevelopmental outcomes included intellectual disability (ID), developmental delay, epilepsy, attention deficit hyperactive disorder (ADHD), and learning disabilities (LD).

Methods: A retrospective chart review of all children clinically diagnosed with NF1 who were seen at Siriraj Hospital from 2005–2015 was performed. All children were 7–18 years old and underwent neurodevelopmental assessment by pediatricians or pediatric psychiatrists.

Results: Out of 256 patients with NF1, 70 (30%) patients were included in this study, including 38 males and 32 females. There were 6 (8.6%) patients with ID, 14 (20%) with ADHD, 14 (20%) with LD, and 4 (5.7%) with epilepsy. The associations were analyzed and found that patients with Lisch nodules had less common ID (5.1% *vs.* 27.3%; OR 0.143; 95% CI 0.024–0.833), patients with neurofibromas had less common ADHD (11.6% *vs.* 33.3%; OR 0.263; 95% CI 0.077–0.899), and patients with macrocephaly had more common LD (42.9% *vs.* 10.2%; OR 6.600; 95% CI 1.861–23.405).

Conclusions: In this hospital-based study of 70 patients with NF1, we found an association between macrocephaly and LD in patients with NF1. However, in patients with Lisch nodules and neurofibromas ID and ADHD were less commonly found, respectively. Given, this study is a hospital-based study and with limited number of patients, further study on a national scale are warranted to better elucidate the association of clinical phenotypes and neurodevelopmental outcomes in patients with NF1.

Keywords: Neurofibromatosis type1; mental retardation; attention deficit hyperactive disorder (ADHD); learning disabilities (LD); epilepsy

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