ORIGINAL ARTICLE

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Comparison of brain perfusion SPECT and MRI findings in children with neuronal ceroid-lipofuscinosis and in their families

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Purpose: Neuronal ceroid-lipofuscinoses (NCL) are among the progressive encephalopathies of childhood that are inherited in an autosomal recessive manner. In this study we specifically aimed to investigate any white-matter changes in the carriers (parents) and the healthy siblings of individuals with neuronal ceroid lipofuscinosis disease and whether we may be able to predict the occurrence of any neurological symptoms in healthy children in the future thus enabling early management. ***Materials and Methods:*** Since the NCLs are genetically determined diseases, we investigated fifteen individuals in three families that had diseased children of the juvenile type, with brain perfusion SPECT and MRI. Brain perfusion SPECT was performed after administering 222–555 MBq (6–15 mCi) Tc-99m HMPAO intravenously in a dimmed and quiet room. Imaging was performed at least one hour after injection, with a three headed gamma camera equipped with high resolution collimators. A Metz filter (FWHM: 11 mm) was used for processing. Cranial MRI was performed with an imager operating at 1.5 Tesla. Spin-echo T1- and T2-weighted and FLAIR slices were obtained for each individual. ***Results:*** In all of the five diseased children we observed pathologic findings both on MRI and Tc-99m HMPAO SPECT. The findings on MRI were mainly features of cerebral and cerebellar atrophy and the observations on Tc-99m HMPAO SPECT were regional perfusion abnormalities. We observed some structural abnormalities on MRI in four of the parents and two of the four healthy siblings. We also noted perfusion abnormalities on Tc-99m HMPAO SPECT in two of the parents and two of the healthy siblings. ***Conclusion:*** Because the disease is inherited in an autosomal recessive manner, the parents and the healthy siblings were not supposed to exhibit any demonstrable brain lesions, but the brain perfusion SPECT and MRI examinations clearly revealed multiple lesions in some of the parents and healthy siblings. Detailed neurological examinations of these individuals were normal except for one apparently healthy sibling (EY). Follow-up imaging of these families is being undertaken and further studies are essential in understanding the pathogenesis and genetics of neuronal ceroid-lipofuscinoses.

Key words: neuronal ceroid-lipofuscinosis, brain perfusion SPECT, MRI