Intracranial Calcification in a Patient with HDR Syndrome and a GATA3 Mutation

Yuichi Hayashi¹, Tetsuya Suwa² and Takashi Inuzuka¹

Key words: HDR syndrome, GATA3, intracranial calcification

(DOI: 10.2169/internalmedicine.52.8911)

Hypoparathyroidism, sensorineural deafness and renal dysplasia (HDR) syndrome is an autosomal dominant disorder (1). We herein demonstrate the brain computed tomography (CT) findings of a 31-year-old woman with HDR syndrome caused by a GATA3 heterozygous mutation (2). Her 1-year-old son had renal cystic dysplasia. Brain CT revealed bilateral and symmetrical calcifications in the basal ganglia, subcortical white matter, thalamus and cerebellum and slight widening of the skull bones (Picture A). Abdominal CT revealed secondary right renal hypertrophy caused by left renal hypoplasia with calcification (Picture B). The cause of calcification was hypoparathyroidism which resulted from the GATA3 mutation.

Brain calcification in patients with HDR syndrome was previously considered to be basal ganglia calcification (3). However, our patient showed extensive brain calcification, similar to that observed in idiopathic hypoparathyroidism (4), at the gray-white junction. The calcification mecha-

¹Department of Neurology and Geriatrics, Gifu University Graduate School of Medicine, Japan and ²Department of Diabetes and Endocrinology, Gifu University Graduate School of Medicine, Japan

Received for publication September 4, 2012; Accepted for publication September 25, 2012
Correspondence to Dr. Yuichi Hayashi, hayashiy@gifu-u.ac.jp
nism present in patients with hypoparathyroidism is unclear. The extent of calcification varies according to the stage of the disease, the duration of the metabolic abnormalities and the volume of calcium deposits (4).

This is a demonstrable case of brain calcification caused by HDR syndrome with a \textit{GATA3} mutation presenting as a familial brain calcification disease.

The authors state that they have no Conflict of Interest (COI).

\textbf{References}