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研究成果の刊行物・別冊

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Original Article

Endocrinological Characteristics of 25 Japanese Patients with CHARGE Syndrome

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Abstract. CHARGE syndrome is a congenital disorder caused by mutation of the chromodomain helicase DNA binding protein 7 (*CHD7*) gene and is characterized by multiple anomalies including ocular coloboma, heart defects, choanal atresia, retarded growth and development, genital and/or urological abnormalities, ear anomalies, and hearing loss. In the present study, 76% of subjects had some type of endocrine disorder: short stature (72%), hypogonadotropic hypogonadism (60%), hypothyroidism (16%), and combined hypopituitarism (8%). A mutation in *CHD7* was found in 80% of subjects. Here, we report the phenotypic spectrum of 25 Japanese patients with CHARGE syndrome, including their endocrinological features.

Key words: CHARGE syndrome, *CHD7*, endocrinological features

Introduction

CHARGE syndrome is a disorder with multiple anomalies. The occurrence of the syndrome was estimated to be approximately 1:8,500 births by Issekutz *et al.* in 2005 (1). The syndrome was first reported in 1979 by Hall and Hinter, and Pagon *et al.* proposed its main features in 1981 (2). The syndrome was named for the acronym of coloboma of the eyes, heart defects, atresia choanae, retarded growth and development, genital and/or urological abnormalities, and ear anomalies and hearing loss. The criteria for CHARGE syndrome were defined by Blake *et al.* in 1998 (3) and updated by Verloes in 2005 (4). In 2004, chromodomain helicase DNA binding protein 7 (*CHD7*), located on chromosome 8q12.1, was identified as the

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