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OBITUARY

In memoriam of Shozo Ohdo MD, PhD: one of the pioneers of clinical geneticist in Japan

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Shozo Ohdo was born on 19 December 1937 and died at the age of 76 years on 16 August 2012.

In the early days of clinical genetics in Japan, he provided several remarkable contributions in the field of clinical dysmorphology as one of the pioneers.

In 1986, he originally described a syndrome with mental retardation, congenital heart disease, blepharophimosis, blepharoptosis and hypoplastic teeth in a brother and sister and in a daughter of their paternal aunt. Subsequently, the condition has been eponymically known as 'Ohdo syndrome' (MIM no. 249620) and nearly 25 cases have been published worldwide. Recently, it has been proposed that Ohdo and Young–Simpson syndromes might be the same condition. In 2011, Clayton-Smith *et al.* described Young–Simpson syndrome as a variant of Ohdo syndrome (MIM no. 603736), and identified heterozygous mutations in the *KAT6B* gene. Responding to their report, we analyzed the *KAT6B* gene in sisters from Ohdo's original paper in 2012. However, we could not identify any pathogenic KAT6B mutations, suggesting that these two syndromes might be different or caused by a different gene involved in underlying signaling cascade.

Second significant contribution of Dr Ohdo is the discovery of EEM (ectodermal dysplasia, ectrodactyly, macular dystrophy) syndrome (MIM no. 225280). He found complex families with this syndrome in the course of field study carried out in a remote island, Koshiki-jima, Kagoshima Prefecture in Japan. He summed up EEM syndrome as a thesis of Graduate School of Medicine, Kagoshima University in 1968, and reported in the *Journal of Medical Genetics* in 1983. In 2005, Kjaer *et al.* identified homozygosity for mutations in *CDH3* gene as a cause of EEM syndrome in Danish and Brazilian families. To my regret, DNA samples of the original family members were not available anymore.

Other than those above, he described several other papers reporting patients with a rare malformation syndrome; for example, tetraamelia with ectodermal dysplasia and lacrimal duct abnormalities (MIM no. 273390), Puretic syndrome (MIM no. 228600), camptodactyly–symphalangism, microcephaly–short palpebral fissure–micrognathia and so on. In addition, he and his co-workers described large numbers of papers corresponding to chromosomal aberrations.

Almost all of these works were done at Miyazaki Medical College, where he was at a position of the Associate Professor of the Department of Pediatrics. After leaving the College in 1993, he served as a pediatrician at Nichinan National Sanatorium, and an educator at Kyushu University of Health and Welfare.

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