© Kamla-Raj 2009 PRINT: ISSN 0972-3757 ONLINE: 2456-6360 Int J Hum Genet, 9(3-4): 255-261 (2009) DOI: 10.31901/24566330.2009/09.03-4.07

A Case with the Combination of Bilateral Microphthalmia, Unilateral Pulmonary Agenesis, Diaphragmatic Eventration and Atrial Septal Defect: PDAC Syndrome

Metin Demirkaya¹, Betül Sevinir¹, Yakup Canitez², Özlem Bostan³ and Meral Yildiz⁴

Department of Pediatric Oncology¹, Pediatric Allergy², Pediatric Cardiology³, Ophthalmology⁴, Faculty of Medicine, Uludag University, Bursa, Turkey

KEYWORDS Microphthalmia. Pulmonary Agenesis. Congenital Heart Disease

ABSTRACT The combination of pulmonary agenesis and anophtalmia or microphthalmia has been described previously. This condition is known as Matthew-Wood syndrome and PDAC syndrome (pulmonary hypoplasia/agenesis, diaphragmatic hernia/eventration, anophthalmia/microphthalmia, and cardiac defect). We report a sporadic case of female infant with the combination of bilateral microphthalmia, unilateral right pulmonary agenesis and diaphragmatic eventration in addition to atrial septal defect (ASD) suggesting PDAC syndrome.