# Pentalogy of Fallot in a patient with Down syndrome

*Down sendromlu bir olguda Fallot pentalojisi*

We present a case referred to our clinic with a lower respiratory tract infection and later diagnosed as Down syndrome with concurrent Fallot pentalogy.

A 39-week 3000 gr girl baby born from the second pregnancy of a 29-year-old mother as the second living child was brought to our hospital on the postnatal 21st day because of coughing and stridor. There was nothing of note in the prenatal and natal history and no consanguinity between the parents. The baby had a Down syndrome phenotype on physical examination with mongoloid eye structure, epicanthal fold, flattened nasal bridge, short and blunt fingers and bilateral Simian lines. The baby`s weight was 3500 gr, the height - 51 cm, the head circumference - 35.8 cm and the vital findings were stable. There was no tachypnea or tachycardia but the respiratory sounds were coarse and there was a pansystolic murmur of 3/6 severity over the mitral focus. Other system findings were normal. There was no pathological finding in routine laboratory tests while a chest X-ray showed right paracardiac infiltration and the 'wooden shoe' sign. The transthoracic echocardiography was consistent with the Fallot pentalogy with pulmonary stenosis, ventricular septal defect, dextraposition of the aorta with septal override, right ventricular hypertrophy and atrial septal defect and the chromosome analysis revealed Trisomy 21.

Congenital heart disease (CHD) is present in 40-50% of individuals with Down syndrome (1). The lesions within the heart can be single or multiple. The type of CHD in patients with Down syndrome is known to vary according to geographical location. Atrioventricular canal defect is the most common single congenital cardiac malformation in most countries and especially in the United States of America and Europe (2). The most common lesion is a single ventricle defect in Asia and an atrial septal defect in Latin America (2). Multiple defects are much rare. Fallot tetralogy is one of the multiple defects in Down syndrome patients and the frequency may be 0-15.5% in different series (1-5). The Fallot tetralogy has been reported to exist together with an atrioventricular canal defect in Down syndrome (1). However, it seems that a case with Fallot pentalogy has not been reported.

Our patient had the Down syndrome phenotype and had not been evaluated from the cardiac point of view previously. She underwent this evaluation when admitted to our clinic for a lung infection and the echocardiographic examination was consistent with Fallot pentalogy. An echocardiogram should be considered in patients with Down syndrome even if there is nothing indicative in the physical examination as there may be concurrent cardiac malformations (1).

In conclusion, this case has been presented because of the previously unreported Fallot Pentalogy and Down syndrome association. Our aim was to emphasize the importance of cardiac investigations in patients with the Down syndrome phenotype.

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