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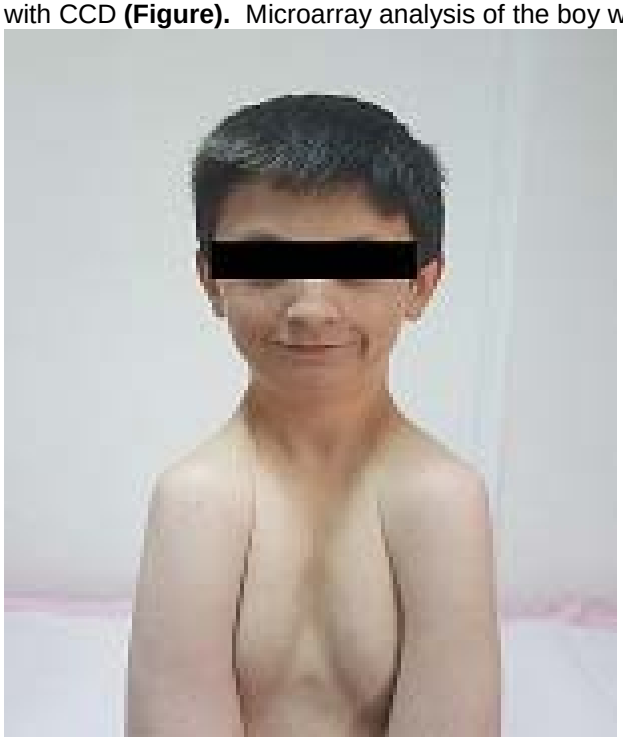
Two Siblings With Primary Pulmonary Hypertension And Cleidocranial Dysostosis:Report Of A New Association

Aypar E., Ertugrul I., Alehan D., Utine E., Kumbasar U., Demircin M.
Hacettepe University Hospital, Ankara, Turkey

Introduction: Cleidocranial dysostozis (CCD) (MIM #119600) is an autosomal dominant skeletal dysplasia characterized by abnormal clavicles, open fontanelles, short stature, dental anomalies and skeletal changes. We report two siblings with CCD and primary pulmonary hypertension (PH). In literature, PH or other cardiovascular system abnormalities have not been reported previously in association with CCD. Two siblings, a 17 year-old girl and a 14 year-old boy were admitted with dyspnea. In both patients, physical examination showed narrow sloping shoulders that can be apposed at midline due to bilateral absence of clavicles, and variety of skeletal changes associated with CCD **(Figure).** Microarray analysis of the boy was normal.



In both patients, echocardiography showed severe PH, right heart failure, massive pericardial effusion (PE). Computed tomographic examination of the lungs were normal. Catheterization confirmed severe PH and exluded congenital heart disease. In the first patient, systolic pulmonary artery pressure (PAP):90mmHg, diastolic PAP:28mmHg, mean PAP:51mmHg, PCWP:11mmHg, Qp/Qs:1, Rp/Rs:66%. In the second patient systolic PAP:76mmHg, diastolic PAP:37mmHg, mean PAP:52mmHg, PCWP:11mmHg, Qp/Qs:1, Rp/Rs:57%.Diagnostic examinations ruled out other causes of PH. Despite specific PH therapy (bosentan 2x 125 mg, PO, inhaled iloprost) and thoracoscopic pericardial window surgery due to chronic massive PE, the first patient was lost during the first year.The second patient was started on bosentan 2x125 mg PO and sildenafil 3x20 mg, PO and had pericardial window surgery. On 4th year of follow-up, last echocardiography showed systolic PAP:85 mmHg, mean PAP:50 mmHg. 6-min walk test: 275 m, resting oxgen saturation:95%.Potts shunt surgery was planned. **Conclusions:** In literature, PH or other cardiovascular system abnormalities have not been reported previously in association with CCD. This is the first report of two siblings with primary PH and CCD.