ASSOCIATION BETWEEN AUTOSOMAL DOMINANT POLYCYSTIC KIDNEY DISEASE AND A SEVERE DILATED CARDIOMYOPATHY: A CASE REPORT

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Autosomal dominant polycystic kidney disease (ADPKD) is caused by mutations in PKD1 and PKD2 genes, encoding polycystin 1 and 2.
We report the case of a 51-year-old woman with arterial hypertension and stage V chronic kidney disease from polycystic kidney disease, without a significant family health history.
She was admitted to our Cardiology Ward for worsening dyspnea, dependent oedemas and a 3/6 systolic heart murmur detected.
Blood tests showed mild anemia and hyperkalemia, severe renal failure (creatinine 6 mg/dl) and normal both liver and thyroid function.
The ECG showed sinus rhythm with normal atrioventricular conduction, low voltage in precordial leads and non-specific repolarization disturbances.
Hemodialysis and intravenous decongestion therapy were started.
A transthoracic two-dimensional echocardiography was performed and a dilated left ventricle with severe global systolic dysfunction (EF 25%) was documented, with severe secondary mitral regurgitation (MR) with a central jet (EROA 0,5 cm2). A possible bicuspid aortic valve was suggested with moderate aortic regurgitation.
CT scans showed small pleural and pericardial effusions, polycystosis affecting both kidneys and liver.
Coronary CT showed a subcritical interventricular artery stenosis and confirmed a bicuspid aortic valve.
Magnetic resonance angiography (MRA) was negative for intracranial aneurysms.
Finally, genetic testing revealed a heterozygous pathogenic mutation of PKD1 gene (c.9622del G p.Ala3208Hisfs*108 exone 28).
Maximum tolerated heart failure therapy was prescribed after discharge, resulting in symptoms reduction but only mild echocardiographic improvement (EF 30% with severe MR). An edge-to-edge transcatheter mitral valve repair was proposed.
In literature we learn that PKD1 mutations are linked to various cardiomyopathies: hypertrophic, dilated, left ventricular non compaction. We report the association of ADPKD with a complex dilated cardiomyopathy with severe secondary MR, an aortic congenital defect and an associated subcritical coronary artery stenosis.
Further investigations are needed to study the genetic aspect of the ADPKD when associated with different cardiac anomalies.