ASSOCIATION BETWEEN POLYCYSTIC KIDNEY DISEASE AND AORTIC ANEURYSM, PRESENTATION OF CASE REPORT AND LITERATURE REVIEW

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Disclosure

Speaker name:

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I have the following potential conflicts of interest to report:

- Consulting
- Employment in industry
- Stockholder of a healthcare company
- Owner of a healthcare company
- Other(s)

- I do not have any potential conflict of interest
CASE REPORT

✓ Male patient 65 yrs
✓ Hypertension
✓ Ex Smoker
✓ Four brothers with aneurysmal aorta disease
✓ Polycystic kidney disease
✓ 2006: huge symptomatic AAA with diameter of 120 mm treated with open repair
2006-2012

SUPRARENAL AND ANASTOMOTIC ANEURYSM OF 68 mm
2012: FEVAR: scallop CT, covered stents in SMA, RRA and LRA
2016: saccular thoracic aorta aneurysm 61 mm and aneurysm of right iliac artery 41 mm
TEVAR
Iliac stentgraft
Decreased renal function – preparing for peritoneal dialysis
# Genetic aneurysma syndromes

<table>
<thead>
<tr>
<th>Syndrome</th>
<th>Gene mutation</th>
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<tbody>
<tr>
<td>Marfan syndrome</td>
<td>FBN1</td>
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<tr>
<td>Ehlers-Danlos syndrome</td>
<td>COL5A1, COL5A2 and COL3A1</td>
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<tr>
<td>Loeys-Dietz syndrome</td>
<td>TGFBR11 AND TGFBR2</td>
</tr>
<tr>
<td>Familial aortic aneurysm and/or dissection syndrome</td>
<td>TGFBR2, MYH11, and ACTA 2</td>
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<tr>
<td>Bicuspid aortic valve</td>
<td>Unknown</td>
</tr>
<tr>
<td>Autosomal dominant polycystic kidney disease</td>
<td>PKD1 and PKD2</td>
</tr>
<tr>
<td>Turners</td>
<td>45X</td>
</tr>
<tr>
<td>Neurofibromatosis</td>
<td>NF1</td>
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Cury et al, 2012
Autosomal dominant polycystic kidney disease (ADPKD)

- Most common genetic disorder with prevalence of 1/1000 (Bauer et al, 2002)
- Caused by mutation PKD1 (85%) and PKD2 (15%)
- Noncystic abnormalities
  - Intracranial aneurysm and dolichoectasias (9.3%-23.3%)
  - Dilatation of the aortic root
  - Dissection of the thoracic aorta
  - Dissection of the cervicocephalic arteries
  - Coronary artery aneurysm
  - Atrial septal aneurysm
  - Mitral valve prolapse
  - Abdominal wall hernias
  - Hepatic and pancreatic cysts
- Aortic medial cystic myxoid degeneration – primary collagen defect in the pathogenesis of aortic dissection
- PKD1 and PKD2 are the genes who code the proteins polycystin-1 and polycystin 2 in the SMC
Correlation between ADPKD and AAA?

✓ Case reports
✓ Patients with polycystic disease

Chapman et al: 9.7%
Roodvoets et al: 5%
Kato et al: 7.1%
CONCLUSION

Patients with ADPK have a higher incidence and prevalence of

- Intracranial aneurysm and dolichoectasias (9.3%-23.3%)
- Dilatation of the aortic root
- Dissection of the thoracic aorta
- Dissection of the cervicocephalic arteries
- Coronary artery aneurysm
- Atrial septal aneurysm
- Mitral valve prolapse

It seems that patients with ADPK have also a higher incidence of aortic aneurysms

Screening of the aortic size using Duplex and/or computed tomography is recommended

Intensive evaluation and follow up monitoring of the whole aorta seems reasonable
Screening of IA using high-resolution CTA or MRA
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