Eisenmenger syndrome is a triad of pulmonary vascular resistance and right-to-left shunting of blood with a systemic-to-pulmonary circulation connection and cyanosis. Although rare, patients with congenital heart disease can present in their adulthood. Primary care doctors need to recognize this syndrome and execute initial work-up since prognosis and quality of life is very poor when it is left untreated.

CASE

40 year old female with Down syndrome presented with progressive shortness of breath, fatigue and altered mental status.

**Physical exam:** BP 90/50, HR102, O2 sat 80%
- Lungs: scattered, bilateral rales.
- Heart: 3/6 harsh holosystolic murmur heard best at the apex radiating to the axilla.
- Extremities: clubbing, 2+ pitting edema up to knees bilaterally.

**Studies:** Hematocrit of 58.
- Transthoracic echocardiogram (TTE): severe right ventricle hypertrophy, 4+ mitral regurgitation, EF 55-70%.
- Echocardiogram w/ bubble study: a right-to-left shunting.
- Transesophageal echocardiogram (TEE): 2.6 cm ventricular septic defect with bidirectional flow. A ventricular septal aneurysm and small secundum atrial septal defect is noted.

**Hospital course:**
- Trial of inhaled nitric oxide (iNO) decreased the patient oxygen requirement.
- Cardiac Cath (Graph 1) shows a persistent high pulmonary resistance despite increasing iNO. Patient is not a candidate for VSD closure and mitral valve repair.
- She was treated with Sildenafil and was placed on a waiting list for possible heart and lung transplant.

**Graph 1: Testing for Reversibility of Pulmonary Vascular Resistance with Right Heart Cath**

<table>
<thead>
<tr>
<th>Mean Pulmonary Arterial Pressure (mm Hg)</th>
<th>Favorable candidate for cardiac repair (&gt;10 mm Hg drop and final PAP &lt; 40 mm Hg with iNO)</th>
<th>Our patient</th>
</tr>
</thead>
<tbody>
<tr>
<td>80</td>
<td>F02 10%</td>
<td></td>
</tr>
<tr>
<td>70</td>
<td>F02 100%</td>
<td></td>
</tr>
<tr>
<td>60</td>
<td>F02 10%</td>
<td></td>
</tr>
<tr>
<td>50</td>
<td>F02 100%</td>
<td></td>
</tr>
</tbody>
</table>

**DISCUSSION**

Recognizing both cardiac and pulmonary symptoms in patients with risk factors for cardiac anomalies is very important. Patients with genetic disorders listed in Table 2 may have cardiovascular malformations that may not present until adulthood.

- Symptoms include: Central cyanosis, clubbing and exercise intolerance.
- Complications: Bleeding, thrombotic diathesis, stroke and bacterial endocarditis.

Initial evaluation should include:
- Chest radiography, ECG, arterial oxygen saturations, exercise tolerance and echocardiography.
- TTE is often the initial test, but it is associated with low sensitivity for smaller defects.
- If suspicion for cardiac defect is high, TEE should be performed even with a negative TTE.
- Other diagnostic tools include: Echocardiogram with bubble study, cardiac catheterization, cardiovascular magnetic resonance imaging or computed tomography.

Although prognosis for Eisenmenger syndrome is poor, emerging medications such as pulmonary vasodilating agents and advances in transplant technology may bring hope for these patients.

**REFERENCES:** Additional reference is provided in a separate sheet.