Kim's Story: 4 Generations

Kim is a 5’11”, 52-year-old Registered Nurse who is descended from a line of tall people with hyperflexible joints and especially elastic skin. Her 27-year-old daughter, also tall and hyperflexible, underwent a workup for complaints of joint pain and fatigue. The daughter was diagnosed with probable Ehlers-Danlos syndrome, a connective tissue disorder known to be genetically transmitted and associated with aortic disease. The daughter had no current aortic manifestations but will now be on a periodic surveillance schedule.

The daughter’s physician recommended immediate aortic imaging for Kim. Kim’s comment was, “At that point, the only thing I knew about thoracic aortic dissection was once watching a man die of it on the table in the O.R. I thought this couldn’t be me, there’s some mistake.” There was no mistake. Kim’s imaging study revealed a 5.7 cm thoracic aortic aneurysm. The cardiovascular surgeon recommended surgery without delay. She underwent open chest surgery for complex graft repair including several defects where the aortic wall was dangerously thinning. She recovered from the surgery over the subsequent months, danced at her 27-year-old daughter’s wedding, and is now restored to a normal life expectancy. Her sister, other daughter and others in the family are undergoing testing.

Kim gained a new understanding about the sudden deaths of her grandfather, uncle and father at the respective ages of 66, 67 and 68, presumed to be due to myocardial infarction. They most likely all died of undiagnosed familial thoracic aortic aneurysm and dissection (TAAD) syndrome. Beginning with Kim’s generation, her family will transition from a 100% death rate to a 100% rate of detection, surveillance and timely treatment.
Anatomy of Aortic Dissection

An aortic dissection is defined as disruption of the medial layer of the aorta with bleeding within and along the aortic wall. Medial degeneration and atherosclerosis are often observed. Dissection has been described as similar to a tear in the lining of a coat sleeve that enlarges as the hand of the wearer delves into the space behind the lining instead of the proper “lumen” of the sleeve. Blood enters the space behind the medial layer and exerts a hematoma effect, putting pressure on the aortic wall. Hypertension adds to the pressure.

- Aneurysm can and often does occur without dissection.
- Dissection can and often does occur without aneurysm.

Dissection Versus Rupture. Aortic dissection is life threatening. The condition can be managed with surgery if it is done before the aorta ruptures. If rupture occurs out of hospital, mortality is virtually 100%. If rupture occurs in hospital, only 10-25% survive to be discharged.

Poor Odds of Diagnosing. Acute aortic dissections are rare, difficult to diagnose and treat, and they are associated with poor clinical outcomes. They may be mistaken for more common conditions such as stroke, heart failure, coronary ischemia, pleurisy, and acute abdominal illness (Braverman, 2011). In a study of 49 aortic dissection patients, initial misdiagnosis was identified by reviewers in 31% (15) of patients (Asouhidou and Asteri, 2009). AM Rogers and others (Circulation, 2011) cite numerous studies showing that acute aortic dissection is suspected at the initial evaluation less than half the time for patients who were ultimately diagnosed with it. This monograph is aimed at improving the diagnosis rate.

Risk factors for aortic dissection include (Braverman, 2011; Buchholz, 2011):

- Hypertension, old age, smoking and atherosclerosis
- Genetic connective tissue disorders (Marfan, Loeys-Dietz, Ehlers-Danlos and Turner syndromes), hereditary thoracic aortic aneurysm or dissection, vascular syndrome, coarctation of the aorta, bicuspid aortic valve, familial TAAD syndrome and tetralogy of Fallot iatrogenic trauma (History of aortic, cardiac, or vascular surgery, catheter or stent)
- Blunt trauma (In 1997, the unbelted Princess Diana died of a torn aorta in a high speed motor vehicle accident.)
- Cocaine use
- Inflammatory diseases
- Pregnancy
Location of Aneurysm Versus Location of Dissection

75% of aortic aneurysms occur in the abdominal aorta and about 25% in the thoracic aorta. However, only about 5% of dissections occur in the abdominal aorta with the rest occurring in the thoracic. Thoracic aorta dissections account for twice as many known deaths as AAA.

Figure 1.

Symptoms And Diagnosis of Abdominal Aortic Dissections

The majority of abdominal aortic dissection patients present with acute, severe pain in the abdomen or back. Patients may also present with early symptoms of peripheral ischemia, distal embolization, and a pulsatile abdominal mass. In a previous review of 47 patients with abdominal aortic dissections, approximately 33% presented with lower limb ischemia, 30% with abdominal pain, and 20% with back or flank pain (Borioni et al., 2005).

The differential diagnoses may include bowel obstruction, gastritis, intestinal ischemia, musculoskeletal pain and mild pyelonephritis, or pancreatitis in hemodynamically stable patients. In unstable patients, pulmonary embolism, myocardial infarction, or perforated viscus should be ruled out. A high index of suspicion should be maintained in patients with the risk factors for developing abdominal aortic aneurysms.
Diagnosis of abdominal aortic dissection is typically made based on ultrasound, CT, and/or MRI results. In a study using data from the International Registry of Acute Aortic Dissection (IRAD), abdominal aortic dissection patients treated with surgical or endovascular procedures had decreased mortality rates relative to medically managed patients (Trimarchi et al., 2007).

**Symptoms and Diagnosis of Thoracic Aortic Dissections**

Recall that 95% of dissections occur in the thoracic aorta. Of those, approximately 65% involve the ascending aorta, 20% occur in the descending aorta, and 10% in the aortic arch (PIAA, 2010). The remaining 5% category “Other” may include thoracoabdominal and unknown. In patients with acute dissection of the ascending aorta, emergency cardiac surgery is recommended, thus these patients should be identified as quickly as possible (Braverman, 2011).

**Figure 2. Thoracic Aortic Dissection Locations**

![Thoracic Aortic Dissection Locations](image)

The most common presenting symptom is sudden acute pain (implying dissection or impending rupture), or chronic pain, from compression or distension. The location of pain may indicate the area of aortic involvement, but not always. Physical examination findings may vary widely (Braverman, 2011).
Ascending aortic aneurysms tend to cause anterior chest pain.

Descending thoracic aneurysms can cause back pain localized between the scapulae.

If the trachea is compressed, there may be wheezing, stridor, or nagging cough.

If the aneurysm involves the aortic root, a murmur may be heard.

In some patients with acute dissection of the ascending aorta, the predominant symptom may be acute heart failure. Syncope and vascular insufficiency may also occur in these patients.

Neck pain. A neurosurgeon in Baton Rouge, Louisiana recounts this experience about a longtime friend and colleague, who was a cardiothoracic surgeon and a smoker. “This was maybe 25 years ago. In the doctor’s lounge, he complained to me about neck pain. I told him his symptoms sounded to me like cervical disc degeneration and said come see me in my office. Two weeks later, he died in the Emergency Department of an out of town hospital of thoracic aortic dissection. It came as a shock to the whole medical community. He was in his 50s.”

Over 90% of patients with thoracic aortic dissections present with acute onset of severe pain in the back, chest, or both. This pain has been described as sharp, tearing, ripping, and stabbing. Aortic dissection-related pain differs from that of myocardial infarction or angina, which tends to increase in severity over time. A confounding factor is that the pain from thoracic aortic dissection can suddenly move, lessen or even disappear.

Revisit Family History of Sudden Presumed Cardiac Deaths

Note that as occurred in Kim’s family, many thoracic dissections have gone undetected and uncounted due to the similarity of symptoms with myocardial infarction and other conditions. If the family history reveals a family member who died suddenly, especially more than one family member, asking for the details can be key to making the diagnosis. How sudden was it? Was it witnessed? Was it presumed to be a heart attack? Was there an autopsy?

“One must consider the potential underlying diagnosis when a patient reports a family history of ‘sudden death’ or ‘heart attack.’... If the patient’s father... had sudden onset of chest pain and then died moments later, there is a chance that the death may have been from an acute AoD rather than an acute MI.” (Hiratzka et al., 2010 ACCF/AHA study).

Contrast-enhanced CT is the most commonly used for diagnosing acute aortic dissections. MRI is also highly sensitive, but may not be practical in emergency situations. Transesophageal echocardiography (TEE) is not as sensitive as CT or MRI but may be more expeditious for an unstable patient.
Recent Development of Tools for Reducing Mortality: The TAD Guidelines and the Add Risk Score

In 2010, the American College of Cardiology Foundation, American Heart Association and numerous other related professional organizations published the Guidelines for the Diagnosis and Management of Patients with Thoracic Aortic Disease (Hiratzka et al., 2010), referred to here as “the TAD Guidelines.” They present a simple bedside screening tool for TAD to be used for patients presenting with any of the following: chest pain, back pain, abdominal pain, syncope or signs of perfusion deficit. The tool lists high risk pain features, predisposing conditions and examination findings. The associated algorithm recommends what to do next.

A portion of the algorithm for aortic dissection risk assessment from the TAD Guidelines is shown below. Physicians are encouraged to consult the full guidelines and algorithm.

<table>
<thead>
<tr>
<th>High Risk Pain Features</th>
<th>High Risk Predisposing Conditions</th>
<th>High Risk Examination Features</th>
</tr>
</thead>
<tbody>
<tr>
<td>Abrupt onset of pain</td>
<td>Known thoracic aortic aneurysm</td>
<td>Evidence of perfusion deficit:</td>
</tr>
<tr>
<td>Severe intensity of pain</td>
<td>Known aortic valve disease</td>
<td>- Pulse deficit</td>
</tr>
<tr>
<td>Pain described as ripping or tearing</td>
<td>Family history of aortic disease</td>
<td>- SBP differential between extremities</td>
</tr>
<tr>
<td></td>
<td>Marfan syndrome</td>
<td>- Focal neurologic deficit with pain</td>
</tr>
<tr>
<td></td>
<td>Recent aortic manipulation</td>
<td>New murmur of aortic insufficiency with pain</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Hypotension or shock</td>
</tr>
</tbody>
</table>

In 2011, investigators of the International Registry of Acute Aortic Dissection (IRAD) database used the risk algorithm from the TAD guidelines to create an Aortic Disease Detection (ADD) Risk Scoring system, demonstrating it to be highly sensitive in identifying high-risk patients (Rogers AM et al., 2011).
To calculate an ADD Risk Score, the clinician adds 1 point for each of the 3 categories above in which any single criterion is met, creating a Risk Score of 0, 1, or 2-3. Note that a single criterion from any two of the above columns suffices for an ADD score of 2-3, “Immediate Risk.” The algorithm continues with recommendations for diagnostic evaluation for each Risk Score. In the case of ADD Risk Score 2-3 “Immediate Risk,” the algorithm calls for immediate surgical consultation and expedited aortic imaging.

**Aortic Dissection Evaluation Pathway**

Adapted from Rogers AM et al., 2011. [http://circ.ahajournals.org/content/123/20/2213](http://circ.ahajournals.org/content/123/20/2213).

**Exercise:** Review the following case, assign an ADD Risk Score and state “next steps.”
Case Study: Failure to Diagnose Thoracic Aortic Dissection

Case adapted from TSG Newsletter, Summer 2012.

Patient: A 55-year-old white male at home experienced an acute onset of substernal chest pain radiating into the back. His wife called 911. The paramedics' note indicated that the patient rated his pain a 10 on a scale of 10, with no prior history of this. He had also begun vomiting and had several episodes of diarrhea that day. During transport to the emergency department (ED), the patient indicated that his pain had decreased to 8 on a scale of 10.

Course: On arrival to the ED, the patient gave a medical history of hypertension and a pericardial window for the treatment of chronic pericardial effusion in the past. The ED staff performed an ECG. There was normal sinus rhythm and no ST or T wave changes. The ECG was of poor quality, but no abnormalities were noted. The physical assessment report showed that the patient was anxious, but the examination was otherwise unremarkable.

Physician’s Actions: The emergency physician noted that the laboratory results were related to upper abdominal pain. The white count was mildly elevated, and the glucose was elevated at 165. The physician noted that his initial diagnosis was biliary colic related to the abdominal pain with radiation to the back. He ordered a gallbladder ultrasound, which was negative for biliary disease. The physician next ordered a contrast-enhanced CT of the abdomen. The patient was not transported to the CT until over 4 hours after his arrival to the ED.

Although the CT order was for the abdomen and pelvis, the radiologist identified abdominal and thoracic dissection. The CT was then extended into the chest, and the radiologist noted a 5.8-cm aneurysmal dilatation of the ascending aorta. The radiologist relayed his findings to the emergency physician and recommended a follow-up chest CT.

The emergency physician did not describe the CT results to the attending physician or contact a cardiologist or cardiovascular surgeon; he instead ordered the recommended follow-up CT. The patient was transported back to the CT, where he became agonal. Advanced life support was implemented, but the physician was unable to resuscitate the patient.

Legal Allegations: The surviving spouse and family filed a suit against the emergency physician, alleging:

- Failure to obtain an initial CT scan of the chest based on the paramedics’ information.
- Failure to pursue the diagnosis of aortic dissection in the clinical context of a patient with history of hypertension and previous pericardial effusion surgery.
- Failure to obtain an immediate consultation for emergency surgical intervention base on the radiologist’s findings.
- Failure to communicate with the treating healthcare providers to report the unexpected CT results.
Use of the TAD Guideline algorithm and the ADD Risk Score might have aided in identifying the diagnosis in this case. Step 1 of the guideline states that all patients with chest, back, abdominal pain, or syncope should be evaluated for aortic dissection.

This patient had at least 1 item under High Risk Pain Features (10/10 pain) and at least 1 item under High Risk Predisposing Conditions (previous pericardial window). So he qualified as “Immediate Risk 2-3” score even without considering examination features. For a patient with this ADD Risk Score, the algorithm would have recommended immediate surgical consult and expedited aortic imaging.

Of course, the immediate availability of an experienced cardiothoracic surgery team would also have been necessary to have changed the outcome for this patient.

**Limitations of the Add Risk Score**

While the ADD Risk score has been found to be highly sensitive, it has been criticized for having a lesser degree of specificity. As noted in this article in the *Cleveland Clinic Journal of Medicine* (Braverman, 2011):

> Important to recognize is that almost two-thirds of patients who suffered dissection in this large database did not have one of the “high-risk conditions” associated with dissection. Additionally, the specificity of the ACC/ AHA algorithm is unknown, and further testing is necessary.

[http://www.ccjm.org/content/78/10/685.full](http://www.ccjm.org/content/78/10/685.full)

An emergency medicine physician commented in an article titled “Screening tool for TAD falls short” (*Journal Watch*, 2011):

> ...until we learn more about use of this scoring system prospectively, particularly its specificity, no change is indicated from our current practice of obtaining imaging (computed tomography or echocardiography) for patients with suspected acute aortic dissection.

[http://emergency-medicine.jwatch.org/cgi/content/full/2011/701/5](http://emergency-medicine.jwatch.org/cgi/content/full/2011/701/5)
Summary and Risk Management Recommendations

— Thoracic aortic dissection is a rare and catastrophic occurrence, easily mistaken for other conditions, and difficult to diagnose and treat in time. Those who reach the operating room in time are often restored to a near normal life expectancy.

— Consider that a family history of sudden death due to presumed heart attack may actually have been due to unrecognized thoracic aortic dissection. Detecting a patient with familial thoracic aortic disease can save lives in the generations to come.

— Emergency medicine physicians, cardiologists and adult primary care physicians are encouraged to become familiar with the risk factors for thoracic aortic disease captured in the TAD Guidelines of the ACCF/AHA, and to understand the strengths and limitations of such tools as the ADD Risk Scoring system.
Bibliography: Aortic Disease II: Diagnosing Dissection

Note: Web links can change over time. If link no longer works, web-search the article by title.

References


