**Down Syndrome and Heart Disease**

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If a 45-year-old man presents to the emergency room with chest pain, what is the big concern? Coronary ischemia (low blood flow to the heart muscle) and possibly myocardial infarction (heart attack). What if that 45-year-old man has Down syndrome? The likelihood that his chest pain is related to a heart attack or low blood flow to the heart muscle is significantly reduced.

The incidence of coronary artery disease (narrowing of the arteries that supply the heart muscle) seems to be very low in people with Down syndrome. The reason for this is not known. It may be that there is a protein coded for on chromosome 21 that reduces atherosclerotic plaque build-up in the arteries. With an extra 21st chromosome in people with DS, there may be more of the protein. It may be that there is a gene on chromosome 21 that turns off a process that contributes to plaque build-up.

The answer is unknown. It is certainly an interesting question for researchers. It will be interesting in the future to see if a prevention or treatment for coronary artery disease (in people without DS) is devised from this information.

In the meantime, the information can help guide an evaluation of a person with Down syndrome who has chest pain. The incidence of heart attacks is quite low in people with DS but not zero. Therefore, it should be considered as a possibility when a person with DS presents with chest pain.

However, there are other conditions that can cause chest pain that are more common in people with Down syndrome (and here are a few examples):

- Heartburn from gastroesophageal reflux
- Chest pain associated with gallstones
- Motility problems of the esophagus (the tube between the mouth and the stomach sometimes doesn’t propel food in its normal rhythmic fashion and that can cause pain)

Therefore, the approach to a person with DS with chest pain will likely have a different focus than someone without DS.

[I should clarify that “holes in the heart” (like ventricular septal defect) and valvular heart disease are more common in infants with Down syndrome. However, we are discussing atherosclerotic heart disease in this article. It is the most common cause of death in the U.S. but is quite uncommon in people with DS.]

**What should we do to prevent coronary artery disease?**

In people without Down syndrome, there is a great deal of attention (that is appropriate) given to reducing risk factors for heart disease. Some of them are not treatable and these will be addressed first.
Risk factors for heart disease include:

1. Family history – there is no way to change this risk factor – we don’t pick our parents.
2. Age – as we age, the risk of heart disease increases.
3. Sex – men have a higher incidence of heart disease than women.

The rest of the risk factors are potentially reversible/treatable but, to the best of my knowledge, these have not been studied in people with Down syndrome as to whether treatment reduces heart disease.

4. Smoking – we have very few patients in our practice who smoke. We certainly encourage them to stop smoking because smoking is related to many health problems. Stopping smoking has been shown to reduce the incidence of heart disease in people without Down syndrome.
5. Diabetes mellitus – although diabetes is a risk factor for atherosclerotic disease, there are mixed findings with regards to reducing atherosclerotic disease by treating diabetes in people without DS.
6. Hypertension (high blood pressure) – this is also quite uncommon in people with Down syndrome. Treating hypertension in people without DS reduces mortality from heart disease.
7. Inactivity – regular exercise reduces the risk of having a heart attack. It also helps treat diabetes, hypertension, and obesity.
8. Obesity – being overweight, particularly if a lot of the fat is around the waist, increases the risk of heart disease and losing weight reduces the risk of having a heart attack.
9. Hypercholesterolemia (elevated cholesterol) – lowering cholesterol with diet and exercise and/or medications lowers the risk of heart disease.

We encourage the treatments that clearly have no downside. For example, if someone smokes we encourage them to stop, if they are inactive we encourage exercise, and if they are overweight we encourage weight loss. We are more cautious about treating with medications that may have side effects.

For example, although we do prescribe cholesterol-lowering medications for some people, we are less likely to do so for a person with Down syndrome than a person who doesn’t have Down syndrome. With such a low incidence of atherosclerotic disease in people with Down syndrome, the benefit of treating with the medications is less clear. In addition, one of the side effects of the most common category of medications used for lowering cholesterol (the statins) is muscle pain. Many of our patients have difficulty telling us about discomfort. In that situation, we may be causing discomfort with the possibility that there is little benefit.

We certainly are strong proponents of providing high-quality health care for people with Down syndrome. We are certainly not in favor of short-changing a person with Down syndrome. However, the first rule of medicine is “Do no harm.” We want to provide optimal care for people with Down syndrome. It is important that we continue to study what that means. Sometimes that means providing the same care as people without DS and sometimes that treatment should be modified based on disease patterns in people with DS.