Colonoscopy and Down syndrome

A colonoscopy is performed with a long scope that is passed through the anus into the colon. The test is performed for one of 2 basic reasons:

1. Diagnostic
2. Screening

A colonoscopy performed for diagnostic purposes is done when the person with Down syndrome has an abnormal symptom or finding. For example, sometimes they are done when a person has bleeding from the rectum, persistent diarrhea or constipation, or abdominal pain. Colonoscopies done for symptoms or abnormal findings are not the ones that are the subject of this post.

This post will focus on colonoscopies done for screening purposes. Colon cancer detection and prevention is the primary reason colonoscopies are done for screening purposes.

The definition of a good screening test includes:
- a test done when there are no symptoms
- the test can find the disease early enough in the course of the disease so that a better outcome is achieved (lower mortality, improved health, etc)
- a test for which there are interventions available that can improve the outcome
- a test for which the costs are acceptable when compared to the benefit (the cost includes financial cost as well as the potential complications of the test and possible follow-up tests).

The recommendation for colon cancer screening generally includes:
- start at age 50 (earlier in people with close family with colon cancer or colon polyps; in person with inflammatory bowel disease; and in certain genetic conditions associated with colon polyps).
- Testing can include:
  - Testing the stool for occult (hidden) blood
  - A rectal exam (with the examiner’s finger)
  - Flexible sigmoidoscopy (a scope shorter than a colonoscopy)
  - Virtual colonoscopy (CT scan)
  - Air contrast barium enema
  - Colonoscopy.

The colonoscopy seems to be becoming the standard. If the other tests are abnormal, a colonoscopy is usually recommended. A colonoscopy looks at the whole colon and if a polyp or abnormality is found, a biopsy can be performed and in many instances the abnormality (eg a polyp) can be removed.
That certainly sounds like something we would want to offer people with Down syndrome just as we do others. However, there are a few issues that have led us to generally not recommend screening colonoscopies for our patients.

We should clarify that this recommendation is for screening colonoscopies. If there is an abnormality or symptom, we do refer patients for colonoscopies.

1. The incidence of colon cancer is much less in people with Down syndrome. Solid tumors (including colon cancer) are less common in people with Down syndrome (see links below).
2. Our experience is that most people with Down syndrome require significant sedation and many even require general anesthesia to complete a colonoscopy. People with DS have a greater risk of complications when it comes to anesthesia (see links below).

Therefore, for those reasons, we do not generally recommend screening colonoscopies for our patients. (The risk is higher and the benefit lower).

Here are some additional questions we have been asked:

**Why don’t we recommend testing the stool for blood or some of the other tests?** The reason is that if the test is abnormal, then a colonoscopy is generally recommended. The same issues that we just addressed would then be present.

**People with Down syndrome are living longer. Will we see a higher incidence of colon cancer that will justify colonoscopy screening?** That is a good question for which we don’t know the answer. At this time, the information that we have is that solid tumors including colon cancer, are less common in people with DS. We will need to continue to monitor the situation into the future to see if this recommendation will change.

**Are there other anesthesia risks?** We have seen and other centers/clinics have reported a small number of individuals with DS who developed sustained additional cognitive impairment after anesthesia (they are noted to have more cognitive or behavioral impairment after anesthesia than they had before). There isn’t enough information at this point to conclude that the anesthesia caused the changes. This is being studied.

Lower incidence of solid tumors (including colon cancer):
Higher incidence of complications from anesthesia:

- Leukemia (especially in children and adolescents)
- Cancer of the testicle
- Cancer of the ovary
- Possibly lymphoma
- Possible malignant melanoma

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