

Down syndrome (DS) and Hirschsprung's Disease (HSCR)

David Croaker

Down syndrome (DS) is the most common condition associated with HSCR. The occurrence of these two conditions together raise many problems for parents and other people who care for these children. Some of these issues are addressed in these four pages.

How often is HSCR associated with DS?

As mentioned in Chapter 3, DS is found in about 10% of HSCR. Looked at from the other point of view, about 1% of children with DS have HSCR.

What other conditions can be associated with DS?

A selected list of conditions commonly associated with DS are given in Table 1. HSCR occurs in DS affected children fifty times more commonly than in the general population. The reasons for this are not clear. DS is associated with a number of conditions. Essentially 100% have a degree of learning difficulty for instance. Other conditions are present at an increased frequency in DS.

Table 1. Some major conditions associated with Down syndrome, and their frequencies.

Condition	Frequency
Learning difficulty	100%
Congenital Heart Problem	30 - 50%
Duodenal atresia	5%
HSCR	1%
Oesophageal atresia	1%

Is HSCR more severe in DS?

Children with DS and HSCR have about the same ratio of long and short segment disease as those children with HSCR who do not have DS. If there is any difference, it is towards DS affected children having shorter segment disease. HSCR is more common in boys with DS, as it is in the rest of the population.

Children with DS and HSCR seem to be more likely to suffer from a form of bowel infection called enterocolitis than other children with HSCR. This condition may cause diarrhoea, fever and swelling of the belly. It affects up to one third of children with DS and HSCR. Prompt treatment is important. Fortunately it seems to get less severe with increasing age.

Can we toilet train our child with DS & HSCR?

Assuming that a pull-through operation has been possible, toilet training is still prolonged and frustrating in children with DS and HSCR. Only a minority of children will achieve full social continence of faeces. Continence seems to depend on the general level of disability of the child, as well as on whether she or he has long or short segment disease. Long segment disease children have more problems with continence.

In the best of circumstances it is probably unreasonable to expect DS and HSCR affected children to be clean before 4 years of age. It is possible for things to continue improving at least up to puberty however.

What are the treatment options in children with DS & HSCR?

The treatment options in the child with DS and HSCR will be determined by a number of factors which will be considered by the team looking after him. These factors are likely to include at least:

- ❑ The length of affected bowel.
- ❑ Whether there is severe congenital heart disease (for instance, a “hole in the heart.”) which might need more urgent treatment.
- ❑ Whether there is severe enterocolitis, or a perforation of the bowel.
- ❑ Whether there are other associated conditions that might make surgery or anaesthetic difficult or dangerous.

Once these factors have been considered the main choice is between:

- 1) Performing a pull-through operation to remove the obstructed bowel.
- 2) Forming a colostomy, without a pull-through.

If a pull-through operation is performed the child will have a possibility of defaecating in the normal way at some time in the future. If the child is left with a colostomy, his surgery and care will probably be simplified, but he will have a bag that will need to be looked after. This may be easier than looking after an incontinent child.

A third possibility, which may be applicable in the shortest segment cases, is to cut the anal sphincter muscle (“sphincterotomy”). This is often combined with an operation to leave the child with a tube into the bowel (the “ACE procedure”) which will allow regular enemas. The tube is placed through the abdomen on the right side, and is fairly inconspicuous. The aim of this tube is to achieve a regular complete bowel emptying, and thereby reduce the soiling the child suffers.

Whichever option is chosen, HSCR does add significantly to the effort required in the care of children with DS. The operation to save the child’s life is only the start of the story.