Hirschsprung disease is a congenital defect which affects around 20-25 children each year in Sweden. The disease is characterized by a lack of neurons in the intestinal wall. This impairs the intestinal movement which ultimately leads to severe bowel symptoms and mortality if no diagnosis is made and treatment given. Diagnostics today is achieved by microscopic analysis of biopsies from the intestinal wall and treatment consists of surgical removal of the affected part of the intestine. The surgical procedure is performed as soon as possible, when the child is only some weeks old. During surgery, the central clue is to resect only as much bowel that is affected by Hirschsprung disease, not more or less. However, too much of healthy intestinal tissue is often removed because there is no tool to exactly determine where the transition from sick to healthy bowel is. Of course, frozen biopsies are sent to the pathologist during the operation in order to confirm that no sick bowel is left, but the method is inexact and waiting for the results adds hours in anesthesia for the child. To address this issue, Pernilla Stenström, Specialist Physician and senior consultant at the Department of Pediatric surgery, Lund, is currently evaluating the use of a modern high frequency ultrasound as a method to distinguish between healthy and sick intestinal tissue during surgery in patients with Hirschsprung disease.

Pernilla addresses the initial idea of an ultra sound project in Hirschsprung disease to her PhD-student Christina Graneli, who also is a resident in pediatric surgery. She insisted on finding an application for ultra sound in her thesis about Hirschsprung disease. A kind donation (Hans and Kerstin Perssons fond for research in Hirschsprung disease) by a relative to a patient made it possible to invest in the ultrasound technology. The establishment of a collaboration with researchers at Lund Technical Highschool, who have the edge knowledge about high frequency ultrasound, has also been extremely important for the project and its translational approach. Currently, Pernilla Stenström is collaborating with associate professor Magnus Cinthio and post doc Tobias Erlöv (Biomedical engineering, LTH, LU) and Prof David Gisselsson (Department of pathology, SUS Lund, LU) to develop a safe and effective procedure to identify bowel wall with and without neurons with help of high frequency ultrasound.

“It’s not possible to distinguish between healthy and sick intestinal tissue in Hirschsprung disease through plain visual observation. The standard procedure today is to analyze biopsy samples to confirm the presence or lack of neurons in the samples”, says Pernilla Stenström. Since the method is still in its early stages, the declared status of the tissue is done by both ultra sound and pathological analysis of biopsies. Post-surgery, the ultra sound is used on the resected bowel to validate the results. So far, 5 patients have been analyzed with the ultra sound and the results have been very promising.

“With ultra sound, not only can we reduce the amount of healthy tissue that is removed, but also the time we keep the infant asleep when waiting for biopsy results”, says Pernilla Stenström. In addition to the ultra sound project, Pernilla Stenström is involved in another collaborative project with Biomed engineering (LTH) and Child and Family health (Health sciences). This project
emphasizes Electronic health (E-health) and safe communication between the family of a child who has undergone surgery, e.g. for Hirschsprung disease, and the Department of Pediatric surgery after the family has left. This project focuses on the use of an “e-pad” (an iPad like device) which allows the user to get in contact with hospital staff and send information, such as pictures, to the hospital. The e-pad has a highly encrypted connection to the hospital which makes it safe to send sensitive information. A pilot study has shown that this approach is good for families and their well-being due to the medical care is easily accessible and continues even after discharge from the department. It is also a good alternative for families who live far away from the hospital, since the e-pad could help avoid long trips for issues that can be solved over encrypted internet communication.

“It has been shown that parents to babies who are offered contact, whenever needed after surgery, reduces their need of contact with professionals in the long term compared to those who are offered only regular limited counseling. Also, medical care should be equal for everyone, even at home after surgery, and for those who live far away from the hospital. I think the e-pad constitutes a potential option for safe medical communication, including pictures, and reductions in long travels for the patients and their families”, says Pernilla Stenström.

Lund University Hospital and the New Karolinska Hospital will from 1/7 2018 be the National Specialized Centers of Medicine (Rikssjukvård) in Pelvic floor malformations and Hirschsprung disease, which means they will be the only Swedish hospitals which will offer surgery to patients with Hirschsprung disease. Pernilla Stenström sees this with optimism and as a big demand; the responsibility to take care of all patients with Hirschsprung disease and pelvic floor malformations on a nationwide basis pushes the limits forward with regards to treatment and research.

“Within one year we will treat twice as many patients with Hirschsprung disease as today, about 12-15 patients, each year. The responsibility to take care of a patient group on a nationwide basis needs to involve development of both treatment and research”, says Pernilla Stenström.

In the future, Pernilla Stenström is hoping ultrasound analysis of Hirschsprung disease will be helping bringing surgical methodology forward. In addition, she is working to improve the reception of patients by involving parents and patients, letting them help guide the research to suit their needs better.

“Research should be a question of what patients need and ask for. The direction of research should be pointed out in close collaborations with patients. That is what I work for”, says Pernilla Stenström.

- Joakim Hising