

GASTROINTESTINAL TRACT KINDLER SYNDROME

1. Introduction

In some forms of EB formation of blisters on the mucous membranes of the mouth and esophagus can occur. This causes various problems; the most common are pain and difficulty swallowing.

Furthermore, it can also lead to reflux (rising up of acid, from gastric contents into the esophagus). The intestinal tract may also be involved, which then for example can lead to constipation.

We especially recommend in this chapter to read only the section that pertains to the EB-form, which you or your loved one is affected by. As this section is the only one really relevant for you.



Important points in a nutshell

- **In some forms of EB not only the skin but also the mucous membranes of the gastrointestinal tract is affected by blistering.**
- **The most common symptoms are: pain in the mouth and esophagus, dysphagia, constipation, reflux, and failure to thrive.**
- **In the chapter on “diet” you will find useful information on this topic.**

2. General Information

Those affected by EB can develop distinct blisters in the mouth and esophagus, to different extents, depending on the subtype. It may as a result cause pain when eating and swallowing however, it may not!

Also those affected by EB often suffer from reflux, i.e. a reflux of gastric contents into the esophagus. Reflux in infants is usually normal, because the muscle at the end of the esophagus does not work sufficiently and so the stomach is not properly "sealed".


If the baby is suffering from the consequences of this reflux or an older child or an adult has reflux, one speaks of a Gastro Esophageal Reflux Disease (GERD). In this case the leakage of acidic gastric contents into the esophagus leads to irritation of the esophagus. Pain can be caused, due to the presence of acid (gastric) in the esophagus. A well-known secondary symptom to this is heartburn.

It is important to treat reflux early, not only because it can affect food intake, but also because the stomach acid can damage the lining of the esophagus and later can lead to a narrowing of the esophagus.

Constipation is also common.

When a baby with EB has to strain to defecate, even with a rather loose stool, this can cause pain and cause blisters in the area of the sensitive skin of the anus. The fear of pain during a bowel movement may lead to a suppression of the urge to defecate and sets a vicious circle: The retained stool is always drier and harder, causing even more pain when you move your bowels. To avoid this pain, the baby or the child suppresses the bowel movement and so on.

Constipation in babies and young children is often the result of a low fluid intake. In addition, iron supplements can cause constipation or worsen it. Therefore, it is usually wise to take measures to prevent constipation when beginning the use of an iron supplement.

For more information, refer to the chapter  „Nutrition“.

3. Problems and Treatments in Kindler Syndrome

Those people affected by Kindler-Syndrome often develop various problems of the gastrointestinal tract.

Some of those affected with Kindler Syndrome have difficulty swallowing. This is manifested in the fact that the person eats less and a soft or liquid diet is preferred. How do you ensure the best possible diet especially during these phases, please read the chapter "diet".

Moreover, if adequate nutrition is not possible orally, after consultation with the treating team a so-called PEG tube may be a good way to help replace oral food, but still allows the child a normal meal. More information can also be found in the chapter "diet".

The swallowing difficulties are caused by a [narrowing of the esophagus](#). When blisters heal in the esophagus, this can lead to scarring. These scars tend to contract, resulting in strictures of the esophagus. Such a restriction makes it harder for the food to glide into the stomach. Then, only a soft or liquid diet can be eaten.

If the narrowing is very pronounced, which is sometimes the case; individuals often spit a lot because they cannot swallow the saliva.

When a narrowing of the esophagus is present and this significantly impairs food intake, the constriction can be stretched under [general anesthesia](#). First an X-ray examination needs to be made, to detect how pronounced and where exactly the constrictions are located.

In this study, the so-called upper GI (gastrointestinal), the patient swallows a contrast medium enriched, thickened drink, and the way this drink flows through the esophagus into the stomach is saved on X-ray film. On the basis of this recording you can determine whether strictures exist in the esophagus, where they are located and how pronounced they are.

Depending on the nature of the restriction, there are different [methods of surgical correction](#). Common to all is that the constriction is dilated.

Common to all methods is that the constriction is dilated. A typical method is the so-called [balloon dilatation](#). In this method an empty balloon is advanced down the throat over a guide wire. The balloon is then very carefully filled with an amount of

air to apply a specific pressure on the stricture and thus dilate the constriction. Under fluoroscopy, (movie- X-ray) the correct position of the balloon is guaranteed. Since the constriction cannot be stretched to the desired width in one session, several dilations over a few weeks is often necessary. Also, despite successful surgery a renewed narrowing can often occur. For more details, please discuss this with your physician.

Due to the involvement of the mucous membrane, some of the people affected with Kindler syndrome develop an inflammation of the colon. This manifests itself in abdominal pain and a mild to severe diarrhea. In severe cases, you may see blood in your stool. The inflammation of the intestinal mucosa may cause slight bleeding; iron is lost through the blood, which can cause anemia due to iron deficiency. This can also happen if bleeding is not obvious.

If you suspect that you or your child an inflammation of the colon may be present, please consult your treating physician or hospital.

Just as there may be strictures in the area of esophagus, they can also occur in the region of the anus. These so-called [anal strictures](#) can lead to constipation. In this case, it makes sense to keep your stool as soft as possible, either by eating stool softening food or if necessary by taking medication. If you still have problems if you take these steps, please contact your treating physician.

Of course [constipation](#) can also occur when there is no anal stricture

Constipation in babies and young children is often the result of a low fluid intake, reduced appetite and/or increased fluid demand (e.g. in hot weather). However, this can evolve as well for no apparent reason. A hard stool can cause pain during bowel movements, resulting in the subsequent fear of pain during defecation which leads to a suppression of the urge to defecate and sets a vicious cycle: The retained stool is always drier and harder, thereby causing more and more pain when having a bowel movement. In addition, the appetite and general feeling of well-being can be reduced. Therefore, prevention is most important: Make sure that your child drinks a lot. If your child refuses water (in infancy it should be boiled and cooled) offer him well diluted fresh fruit juice or bottled baby juice, diluted at least 1: 1. Once you start feeding your baby solids, make sure to feed it vegetables and pureed fruit daily. Please be aware that bananas, blueberries and rice can cause

hardening of the stool. In contrast baby food with apple or pear, act the opposite by softening the stool. Whole grain products should not be offered to a baby! Please read more about this in the chapter "diet".

Be consistent in the treatment of constipation because the longer the above vicious cycle exists, the longer it takes to break it again.

If stool softening food and adequate fluid intake alone is not sufficient to resolve the constipation, there are other options: For example, to treat constipation, Optifibre® can be given. Optifibre® contains soluble, tasteless fiber from the Guar bean. It is used for the regulation of intestinal activity.

If your child is under the age of three, please talk with your pediatrician as to whether he recommends Optifibre® and if so, what dosage you should give your child. Optifibre® is given over several days in increasing dosages. It is important that you drink enough fluid while taking it! When over a longer period of time the digestive tract is again working properly, Optifibre® may eventually be discontinued, however it must be "weaned" by slowly decreasing the dosage.

If, despite all these measures constipation persists, a laxative should be given. Consult your pediatrician as they will prescribe the most appropriate laxative for your child. At EB we have had good experiences, for example with Macrogol (Movicol®). When using Movicol® you should also drink enough fluids.

It is better to give these digestive enhancing agents regularly and proactively, rather than when constipation occurs. Because the vicious cycle of "pain during bowel movements - fear of pain - suppressing the urge to defecate - Pain" is already set in motion.

After a period of stubborn constipation, the rectum needs a "recovery period" of several months to adapt again to a smaller and softer stool mass. Therefore, you should give Macrogol, even after the stool returns back to normal, for at least half a year! Otherwise, you'll return back to having the same digestive problems within a short period of time.

Treatment with Macrogol is the same as with Optifibre®: It should be given over several days in increasing dosages and when discontinuing it you must be "weaned"

by slowly decreasing the dosage. Of course, there may be [constipation](#) as well in older children and adults.

It is the same for them:

You should [drink plenty of fluid](#) (preferably water, diluted fruit juice, unsweetened fruit juice and herbal teas) pay attention, especially on hot days.

Physical [exercise](#) is beneficial for digestion. A walk or other exercise will contribute to good digestion and physical well-being.

Some [foods](#) should be avoided because they are constipating, such as white flour products, husked rice, cocoa powder, chocolate, gummi bear candy, bananas etc. In contrast, there are other foods that act as stool softeners, e.g. fiber foods such as whole grains, vegetables and fruits, prunes that were soaked overnight in water, naturally cloudy (unfiltered) juices, etc. For more information, please read the chapter "diet".

If these measures alone are not sufficient to help the constipation Optifibre® can be given. Optifibre® contains soluble, tasteless fiber from the Guar bean, and is available at the pharmacy. For the exact dosage, please refer to the package insert, Optifibre® is "titrated" over several days by increasing the dosage. You also need to drink enough while taking it! When the digestive tract starts working properly for a longer period of time Optifibre® may eventually be discontinued, it must be weaned slowly by daily decreasing the dosage.

If the measures mentioned so far are not sufficient, additional laxatives should be taken. At EB we have had good experiences with Macrogol (Movicol®). Please contact your doctor; he will prescribe the most appropriate laxative for you.

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A [pain relieving ointment](#) (e.g. containing Lidocaine) can be supportive in both children and adults; it should be applied before defecation. This is especially useful if small, bleeding lacerations of the mucous membrane of the anus are present.

If the constipation persists and the laxative medications are not effective, a doctor can give an enema, emptying the rectum and thus giving pain relief. **Warning:** however, enemas should not be given too often as it may make the problem worse or amplify it!

Such an enema should be carried out for those affected by Kindler Syndrome with great care to ensure that the mucosa is not injured. It is advisable to apply cream to the anus and insertion piece of the enema with Vaseline or a similar fatty ointment to keep the friction to the area as low as possible.

In addition, iron supplements can cause constipation.

Therefore, it is usually wise to prevent constipation early especially when beginning an iron supplement. You should also start using Optifibre[®] or macrogol at the same time you start using Iron supplements, if you already are using Optifibre[®] or Macrogol you should increase the dose when you start taking iron supplements.

[Anal fissures](#) are tears in the anal mucosa, as mentioned briefly above. They extend in the longitudinal direction of the anal canal and cause pain during and after defecation and can cause bleeding. If you are affected by an anal fissure, analgesic ointments are used as a local pain reliever, however, it is also important to keep the stool as soft as possible.