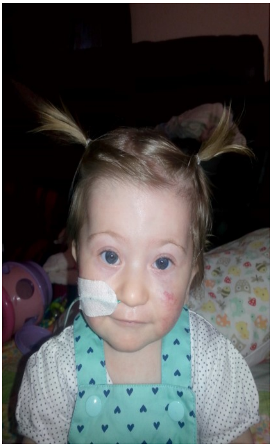
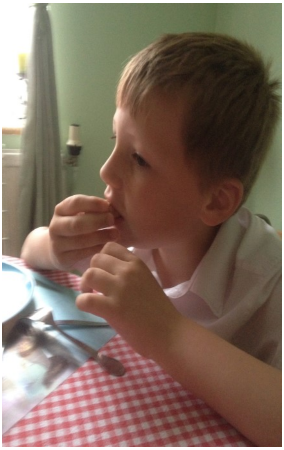
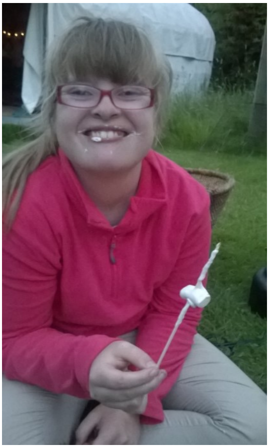
Understanding Chromosome & Gene Disorders

Feeding







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Feeding guide

Feeding problems can be common in babies and children but for those born with a rare chromosome or gene disorder it can be more likely. There are many possible reasons for these difficulties because of the different parts of the body and nervous system involved in this everyday activity. Sometimes the physical structures of the body are affected, such as when babies are born with a cleft lip and palate, or a high-arched palate. For some babies, it is challenging to organise the movements required for feeding because of muscle weakness (Hypotonia) or difficulties in co-ordinating movement. For others it is challenging to organise the complicated task of breathing, sucking and swallowing. Disturbances to the digestive system may result in babies having difficulty keeping their feed down (reflux, see page 7). Some children may overcome these early feeding difficulties as they get older. Some children can develop feeding and swallowing (Dysphagia, see page 9) problems in later years. It is important to recognise that even babies and children who are fed by tube when young, can go on to develop the skills needed to eat and drink.

This guide has been compiled to help parents of children with chromosome and gene disorders find further information and support.

Early Feeding

Breast feeding

Some babies may have difficulties latching onto the breast, this may be because of a poor sucking reflex or other problem such as cleft lip and/or palate. They can be fed expressed breast milk or formula milk. It may be necessary to use special bottles or teats (see page 6).

- **National Breastfeeding Helpline**

http://www.nationalbreastfeedinghelpline.org.uk/
0300 100 0212
The Breastfeeding Network (BFN) is an organisation based in the UK. They are an independent source of support and information for breastfeeding women.

- **National Childbirth Trust (NCT)**

https://www.nct.org.uk/baby-toddler/feeding
0300 330 0700 (Option 1: NCT infant feeding line)
Email: enquiries@nct.org.uk

Allergies and intolerances

Some babies have milk allergies, while others have milk intolerance or lactose intolerance.

Cow milk protein allergy (CMPA) occurs when a baby's immune system reacts to the proteins in cow's milk. The immune system identifies the cow's milk protein as a 'foreign substance' and activates an immune response that presents as an allergic reaction. Babies can experience an immediate immune response that involves an antibody called IgE or a slower immune response that does not involve this particular antibody (often called milk intolerance). Symptoms can include hives; swelling; trouble breathing; vomiting; abdominal pain; skin rash; watery, itch and swollen eyes; stuffy nose; bluish skin; eczema and in very rare cases anaphylaxis, a severe allergic reaction that requires urgent medical attention.

It is also thought that breast fed babies might react to breast milk if the mother has a diet that includes cow milk proteins. If a breast-fed baby is thought to have CMPA, a doctor may recommend that the mother cuts dairy products from her diet.

If a baby with CMPA is fed with a milk formula, soy milk or goats milk may be suggested as a replacement. The other options include hypoallergenic formulas that contain milk proteins that are partially or fully broken down. Many children outgrow their CMPA and go on to have a full and healthy diet that can include dairy products.

Lactose intolerance is extremely rare in babies, if this does occur, it usually happens later in childhood or adult hood. The immune system is not affected; it occurs because the digestive system is unable to digest a sugar called lactose found in dairy products. Babies with lactose intolerance might be irritable and have gas, a bloated stomach and diarrhoea. They might 'spit up' their feeds often and struggle to gain weight. Children with lactose intolerance may benefit from drinking a low or no-lactose containing milk.

> *“ When my son was first introduced to milk formula at about 5 months old, he had an immediate allergic reaction. Within seconds he started to scream and his face and torso were covered in hives. We weren't 100% sure if it was a coincidence that this happened immediately after drinking formula (we had no idea about CMPA at the time) so we tried again a few days later and the same thing happened. A quick trip to the doctors confirmed that he had CMPA. I breast fed him for longer than planned and attempted goats milk when he was about 9 months old, he did not react. He grew out of his allergies after a few years and now loves eating dairy products! ”*

There are many different types of food allergy, not just milk but: nuts,

fruit, shellfish, wheat, gluten, etc. Children can develop rashes and lumps on their skin, or swelling of tongue, lips or mouth.
Here are some additional links to support:

- **NHS** https://tinyurl.com/2p9d9f43 and https://www.nhs.uk/conditions/food-allergy/
- **Allergy UK** https://www.allergyuk.org/about-allergy/allergy-in-childhood/
- **Children with Coeliac disease** https://www.facebook.com/groups/2374049168/
- **USA based allergy support** https://www.kidswithfoodallergies.org/

> “ *J lived off PaediaSure then PaediaSure with yoghurts and completely smooth puree until 7 years of age when we started an intensive feeding program. One of the best pieces of advice from our SALT for severe intolerance to lumps in food was to VERY slowly build up - adding wheatgerm (from a health food shop) to the puree. Literally starting with a 20th of a teaspoon and building up over weeks and weeks – I was very doubtful J would tolerate this but he did, so we got to stiff purees which helped to exercise the mouth. The next clever thing was to add completely soft bits into this solid puree, the texture being such that you cannot detect the difference between puree and bits. J is still learning to chew, but now can eat a wide range of soft mashed foods which is massive progress. To encourage chewing his IEP has a target to chew soft food 5 times on each side in front of a mirror and he gets stickers for each chew. He also has a chewy tube.* ”

Failure to Thrive

Some babies are described as having failure to thrive. This means that a baby is too weak to drink enough to meet their own nutritional requirements and is very common, and can be helped by enriched formula, such as Paediasure ™
https://patient.info/doctor/faltering-growth-in-children

What causes feeding and swallowing disorders in children with rare chromosome and gene disorders?

- Cleft lip
- Cleft palate
- High-arched palate
- Submucous cleft palate (hidden cleft)
- Conditions affecting the airway
- Gastrointestinal conditions (e.g. reflux)
- Head and neck anomalies
- Nervous system disorders (e.g. cerebral palsy)

- Medications that may cause lethargy or decreased appetite
- Muscle weakness and low muscle tone in the face and neck (hypotonia)
- Multiple medical concerns
- Prematurity and/or low birth weight
- Respiratory difficulties
- Other – refusal to eat certain food/textures – gagging, spitting out, being sick etc. in some children diagnosed with Autism or other neurological conditions for example: Sensory Processing Disorder (SPD).

Cleft lip and palate

Babies with a cleft lip or palate or submucous (hidden) cleft, which isn't obvious, but often causes feeding problems, because it's usually accompanied by abnormalities of the mouth muscles, have greater feeding difficulties until their condition is stabilised or surgically corrected. Babies with a cleft lip/palate might have to be bottle-fed but not necessarily with formula. They could be fed with expressed breast milk using a specialist bottle such as the Habermann Feeder. There are huge numbers of different bottles, teats and ideas that can help overcome the difficulties associated with feeding babies with clefts. The UK organisation CLAPA (Cleft Lip and Palate Association) has a wealth of advice and information.

- **CLAPA** https://www.clapa.com/

020 7833 4883
Email: info@clapa.com

Specialist bottles and teats

- **The Haberman Feeder** https://mandyhaberman.com/ and https://habermanbaby.com

The Haberman Suckle Feeder looks similar to standard baby bottle feeders but is designed to better mimic breast feeding. Milk does not flow under gravity as it commonly does with other feeding bottles; the baby needs to suckle on the teat and so has greater control over its fluid intake. This bottle was designed by Mandy Haberman over 30 years ago when she struggled to find a suitable feeder for her child with feeding difficulties. There are other specialty bottle and teats available:

- **Medela** https://www.medela.com/breastfeeding/products/collecting/bottles
- **Dr Browns** https://drbrowns.co.uk/

Dr Browns has a speciality bottle with cleft palate valve, some other bottles are wider/narrower, which can help alternate between breast feeding and bottle feeding; others have a vent which may help with colic.

- **NUK** https://www.nuk.co.uk/catalog/category/view/s/special-teats/id/16/

Produce specialist teats for babies with a cleft lip/palate.

Reflux

Gastro-Oesophageal Reflux (GOR) occurs when feeds frequently and forcefully return up the food pipe (oesophagus) from the stomach. The oesophagus is a long tube of muscle that runs from the mouth to the stomach. Sometimes, but not always, the contents of the stomach may continue to travel into and out of the mouth (vomiting, spitting-up, regurgitation). GOR can occur due to a number of different reasons. It is very common in babies and young children, but can also occur in older children and adults. GOR can affect babies who are breast-fed or bottle-fed, with or without a rare genetic disorder. GOR is more common in babies who are born earlier than expected and those with a low birth weight. It is also common in babies with a food allergy or a physical anomaly (E.g. Hiatal hernia, cerebral palsy, problems with the lower oesophageal sphincter); it usually improves over time without treatment, but this is not always the case. Reflux may be misdiagnosed or dismissed as colic (excessive crying, stiffening of the body and possible digestive problems) since babies can appear to react in a similar way.

What medications are available to treat reflux?

The vast majority of children do not need any treatment. Medications like Omeprazole, or sodium alginate (which work by reducing the actual amount of acid produced in the stomach) might be prescribed if necessary.

Great Ormond Street hospital (GOSH) have a page on their website all about reflux and its' treatment: https://www.gosh.nhs.uk/conditions-and-treatments/conditions-we-treat/gastro-oesophageal-reflux/

Gastro-Oesophageal Reflux Disease (GORD)

Gastro-oesophageal reflux disease (GORD) is a common condition which can affect anyone. It is caused when acid leaks out of the stomach into the oesophagus.

Common symptoms of GORD include:

⇒ Heartburn – burning chest pain or discomfort that occurs after eating
⇒ An unpleasant taste in the mouth
⇒ Dysphagia – difficulty swallowing

A lot of people experience occasional episodes of GORD, but if people have persistent and reoccurring symptoms it is normally regarded as a condition that needs treatment. Taking antacids to help neutralise stomach acid can help.

This is a useful document highlighting the difference between GOR and GORD which is probably important to emphasise e.g. GORD (Gastro-Oesophageal Reflux Disease) is a diagnosis reserved for those infants who present with significant symptoms and/or faltering growth.
https://tinyurl.com/mujahfyj

Some babies with GORD have poor weight gain and can be more unsettled than normal. Occasionally, babies may have wheezing if the condition is more severe. It may be the case in children with rare chromosome or gene disorders that a lot of the children have silent reflux that is painful, but they have no way to express their pain. They could make aggressive noises like growling, and some self-harming behaviours may also be attributable to reflux.

- ♦ **Infant reflux** https://www.infantreflux.org/
- ♦ **Forum** https://www.infantreflux.org/forum/

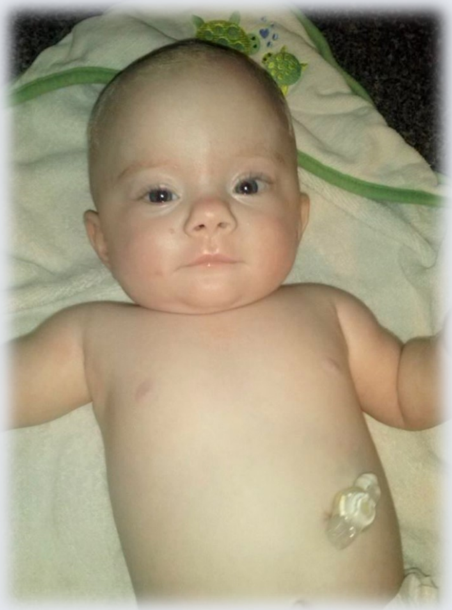
Support forums to chat with other parents

- ♦ http://www.reflux.org/

- ♦ https://www.netmums.com/local/l/reflux-and-related-food-allergies-support-group-1

- ♦ **Living with reflux** https://www.livingwithreflux.org/

Facebook: https://www.facebook.com/groups/livingwithreflux
Offers support for families who have infants, children and young adults who experience GOR, GORD and other associated conditions.



Dysphagia

Dysphagia is used to describe swallowing, eating and drinking difficulties. Some people with dysphagia might have a problem with one, many or all foods and/or liquids and some people might not be able to swallow at all. Further information: https://tinyurl.com/bd5wupwu

- **NHS**

https://www.nhs.uk/conditions/swallowing-problems-dysphagia/treatment/

Eating, drinking and swallowing difficulties can occur at the different stages of eating and drinking:

- Bringing food or drink to the mouth - you organise and co-ordinate breathing, locate and identify food and drink, mouth opening and getting ready to take food or drink in the mouth; you may need help bringing food and drink to the mouth, or may bring more food to the mouth when the mouth is still full.
- When food and drink are in the mouth you make movements to suck, bite, chew, and move food or liquid from side to side and from the front to the back of the mouth ready for swallowing, breaking up bits of food so that they are small enough for swallowing or holding liquid in the mouth until ready to swallow.
- When food and drink enter the throat you need to make some voluntary and involuntary movements to start the swallow which squeezes food down the throat, closes off the airway to prevent food or liquid from entering the airway (aspiration), pushing the food and drink in to the tube which opens in to the stomach. There are protective mechanisms at this stage such as coughing and gagging to keep the throat clear of food and liquid. The muscles of the oesophagus (muscular tube leading to the stomach) relax and then contract in a co-ordinated way to quickly squeeze food and drink all the way down to the stomach.

Signs and symptoms of feeding and swallowing problems in children:

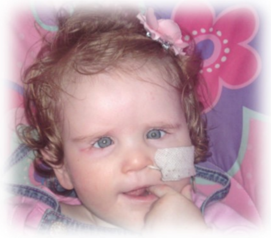
- difficulty breast feeding
- refusing foods or liquids
- taking a long time to feed
- unusual body movements during feeding
- irritation during feeding
- spitting food out
- unwilling to accept different textures of food
- difficulty chewing
- coughing, choking or gagging during meals
- food or saliva coming out of the mouth or nose
- frequent vomiting
- changes to the sound of someone’s voice or breathing
- difficulty coordinating breathing with eating and drinking

Difficulty with swallowing can lead to food or drink going in to the lungs; it is likely that this will trigger respiratory infections or pneumonia. This is called “aspiration”. Sometimes it will be obvious that someone has aspirated because they will cough, their eyes will water, they will change colour. Sometimes it will not be obvious and is called “silent aspiration”. If aspiration is suspected, it is recommended that the child has a dynamic video X-ray of swallowing called a videofluoroscopy. They may also have problems gaining weight or reaching growth milestones.
Children may be at risk of:

⇒ dehydration
⇒ poor nutrition
⇒ constipation
⇒ aspiration pneumonia or repeated upper respiratory infections

Dehydration occurs when your body loses more fluid than is taken in. When the normal water content of your body is reduced, it upsets the balance of minerals (salts and sugar) in your body, which affects the way that it functions. Some of the early warning signs of dehydration include:

⇒ feeling thirsty and lightheaded
⇒ having dark coloured, strong-smelling urine
⇒ passing urine less often than usual



Dehydration signs to look for:

⇒ few or no tears when they cry
⇒ drowsiness
⇒ a sunken soft spot (fontanelle) on their head
⇒ fewer wet nappies

How are feeding and swallowing disorders diagnosed?

If you suspect that your child is having difficulty feeding, contact your GP or paediatrician right away. A GP or paediatrician will examine them and address any medical reasons for the feeding difficulties. They might need to be seen by a Speech and Language Therapist (SALT) who specialises in the management of eating and swallowing disorders. They may also need to be seen by an ENT (Ears, Nose and Throat) consultant, a Physiotherapist or a dietitian. You will be asked questions about your child's medical history, development and symptoms and they may:

♦ observe feeding to see your child's readiness to feed, posture, behaviour, and oral movements during eating and drinking
♦ look at the strength and movement of the muscles involved in swallowing
♦ perform a test to evaluate swallowing, such as: a videofluoroscopy, where your child will be given food and liquids to eat and drink (those items will have barium added to them) and then the swallowing process is videoed on an X-ray
♦ further additional tests may also be necessary.

Your child's self-feeding abilities and their health and nutrition will be discussed by the team and they can then make a plan on how to improve a child’s feeding.

They may recommend:

⇒ medicine for reflux
⇒ changing to different foods, and/or adding calories to food
⇒ adding new foods
⇒ postural or positioning changes (different seating)
⇒ referral to other professionals

The Speech and Language Therapist (SALT) can help with:

⇒ improving chewing
⇒ making the muscles of the mouth stronger
⇒ thinking about the sensory experiences of mealtimes
⇒ increasing acceptance of different foods and liquids
⇒ improving sucking and/or drinking ability
⇒ altering food textures and liquid thickness to ensure safe swallowing.

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Parents can help by providing information and asking questions to help health professionals understand the problems their child has with feeding and swallowing; this includes making sure that they understand the treatment plan for their child and following any recommended techniques at home. The SALT may be able to go into school or other short breaks care to help teach those supporting the child, so that the techniques are followed by carers other than parents. Parents can also contact their SALT to tell them about what is, or is not working at home/school/respite. Information from: https://www.asha.org/public/speech/swallowing/feeding-and-swallowing-disorders-in-children/

◆ **Dysphagia cookery book in pdf format**
http://gutscharity.org.uk/wp-content/uploads/2016/09/Speech-Nyu-Dysphagia-Cookbook.pdf

◆ **Dysphagia cookery book**
https://swallowingdisorderfoundation.com/which-dysphagia-cookbook-is-right-for-you/

Medical treatments, including tube feeding, gastrostomy, Nissen's fundoplication.

Tube feeding

Naso-gastric intubation is a medical procedure that involves the insertion of a plastic feeding tube (nasogastric tube or NG tube) through the nose down into the stomach. If your child needs to be fed through a tube that goes directly into their stomach (it is relatively common and often temporary). Information and support from: https://pinnt.com/Support/Half-PINNT.aspx

- **USA based website**
  https://www.cincinnatichildrens.org/health/g/g-tube-care
  A video and useful cleaning care tips for g-tubes.

- **NoTube** https://notube.com/
  NoTube was founded to help children around the world to over come the severe condition of tube dependency. The ultimate goal is to teach the child how to eat and to finally remove the feeding tube. They also have a facebook group:
  https://www.facebook.com/groups/404270672987962/

- **Oxford Academic Science Network**
  https://tinyurl.com/4t4yu6m5
  Has some useful guides on PEG care and button care.

- **Oxstar**
  https://www.oxstar.ox.ac.uk/more/supporting-parents/watch-the-videos
  Videos on PEG care and button care.

There are also several other Facebook groups run by parents which might also help with tips, here are just a few:

- **Tube Feeding your child in the UK support group**
  https://www.facebook.com/groups/177590912307248

- **Feeding tube support UK**
  https://www.facebook.com/groups/feedingtubesupport

- **(USA based) Pediatric feeding disorders / FTT/ GERD/and kiddos with feeding tubes** https://www.facebook.com/groups/feedingdisordersandkiddoswithfeedingtubes

> “ L was tube fed from birth until 16 months of age. He was nil by mouth and the doctors/specialists told me he would never eat enough to sustain himself, ever. He was ng fed and they suggested he have a peg and a Nissen’s at the same time as L suffered from reflux. I refused and after doing some research I took him to Austria (https://notube.com/) and they had him eating orally within 9 days. He is now 8 and eats for England. Had I listened to the UK specialists, L would still be tube fed. They told me he was “orally aversive”, which we found out was untrue. His reflux also went as soon as the tube went, so the Nissen’s would’ve been for nothing (it’s very invasive surgery). ”

Gastrostomy

A gastrostomy is a feeding tube inserted surgically through the abdomen into the stomach. This allows a child to be fed directly into his or her stomach, bypassing the mouth and throat. You can find out more about this procedure on the Great Ormond Street Hospital website: https://www.gosh.nhs.uk/medical-conditions/procedures-and-treatments/living-with-a-gastrostomy/

> “ *The thing that helped A was her gastrostomy. It means she has all the calories and nutrients she needs and I don't have to worry myself sick and means we have had less hospital admissions and I can keep her hydrated. A can eat as much or as little food she chooses, it's more a sensory/ pleasure, it’s not a battle for us anymore.* ”

Fundoplication

What is a fundoplication?

A Nissen’s fundoplication is an operation used to treat gastro-oesophageal reflux. Some babies and children have a gastrostomy during the same operation. The fundoplication operation is usually carried out using keyhole surgery (laparoscopy). You can find out more about this procedure on the Great Ormond Street Hospital website: https://www.gosh.nhs.uk/conditions-and-treatments/procedures-and-treatments/fundoplication-0/

Weaning

Many of the children on the Unique support are developmentally delayed. Parents tell us that they might get the hang of bottle/breastfeeding with time, but then remain on soft/pureed food for much longer than other children. A health visitor can advise you about feeding and weaning your baby. As your child grows, it might be necessary to ask for a referral from the GP or health visitor for further specialist help.

- **NHS link on weaning** https://www.nhs.uk/start4life/weaning/

> *"Our little girl had infant Nutrini milk from the hospital to help her gain weight, despite being breast fed as well. When she was able to eat, eating at home was easier; as she wouldn't eat a thing when we were out and was very unsettled, always having to be on my knee if we were out. Routine was and still is key for her. Textures of food were also an issue."*

- **Weaning a premature baby** https://tinyurl.com/bdd7ryew

Chewing

Chewing is far more complicated than you might think. Skilled chewing and biting requires sensori-motor learning, where the child learns to use sensory information to develop appropriate oral movements to manage different food textures in the mouth. Foods react differently in the mouth, some need prolonged chewing and others just fall apart. Many children with chromosome or gene disorders might have a physical reason why they can't chew. Others might have sensory issues including oral defensiveness that makes it difficult for them to accept certain foods into their mouth, particularly those with lumps or unusual textures. Some children do not know how to chew using their teeth and tongue correctly.

Developing chewing skills

The introduction of solid foods into your baby's diet is important for the development of oral motor skills. Oral motor skills describe the actions of the tongue, mouth, lips, cheeks and jaw as they chew, suck, bite and lick. These motor skills work the same muscles in the mouth as those that are needed for the development of speech.

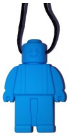
- **Chewing and speech development**

https://tinyurl.com/2p8fsf9m

Some children like to chew inappropriately, like clothing for example. There are items available to help stop that, like the items sold here:

- **Chewelry** https://chewelry.co.uk/
- **Fledglings**

https://www.fledglings.org.uk/collections/oral-care-chewing



Chewy Man Blue

> “ A is always raspy we haven't done the swallow study yet, but our play therapist recommended thickening. Keeping in mind A’s tummy doesn't do well with oatmeal (constipation), we thicken with baby yogurt and it works GREAT, plus extra calories, and she loves it. ”

Teeth

It is important to think about your child’s teeth. Painful or crowded teeth and sore gums can contribute to feeding problems. We also know that feeding problems can affect the teeth. Please read the Unique practical guides on Teeth and dental problems on our website:
https://rarechromo.org/practical-guides-for-families/

> “ We have issues with our son’s over-eating. As a small baby/infant, he was fussy with texture. We thought he was 'lazy' as he wouldn’t chew and wouldn’t eat anything with bits in until he was over 12 months. We had to simply persevere and keep trying things. I don’t have much advice, except don’t give in, you have to de-sensitise them as much as is humanely possible, without stressing them out. M still has some sensory issues with textures, smells, colours, etc, but on the whole is a good eater. ”

Choking and swallowing

Chewing difficulties and choking often go hand in hand for some children with chromosome or gene disorders. Structural difficulties, hypotonia (low muscle tone) and sensory difficulties can make biting and chewing food quite challenging. Reflux, can cause choking, coughing or aspiration. Keeping your child properly positioned during eating and for at least half an hour afterwards, will aid the digestion process. Helping your child to pace their mealtimes can also help, for example, encourage your child to swallow one mouthful before putting more food in their mouth. Some children with chromosome or gene disorders are known to over-stuff food into their mouths and swallow without chewing properly. Watch them closely when eating, making sure they don't throw their heads backwards, whilst they have food in their mouth.

♦ **NHS link to how to stop a child from choking**
https://tinyurl.com/mvxdf7yz

♦ **UK RESUS Council**
https://tinyurl.com/34w4vp6t

♦ **First aid for life**
https://tinyurl.com/4k2rzm62

Postural alignment, with body and head held straight, can improve swallowing and decrease feeding difficulties. Specialist seating is available e.g. chairs with a tray and/or footrest that allow children to sit upright at the dining table, or close to the dining table while they eat, which also helps to include them as part of family meal times. If a child doesn't have the motor control to keep their body and head upright and straight while eating, they are more likely to choke. Some Unique children have found their own positions for feeding comfortably, which can also reduce the risk of choking. Some children may not be able to feed themselves because of constant body movements, or needing to use their arms or hands to keep themselves balanced. You can always get positional seating advice from an Occupational Therapist (OT); in some circumstances they may be able to supply specialist seating on loan from the NHS.

> “ My son had a Videofluoroscopy. It was discovered he was aspirating trace amounts with every mouthful - but he didn't cough like you or I would. The fluoroscopy showed that he wasn't chewing his food, but rolling it around in his mouth with his tongue and then sucking it down his throat, he even did the same with drink. With the help of a specialist feeding Speech and Language Therapist, we devised an eating plan for him. Eating with his head in a downwards position works better for him, it is his way of coping. ”

♦ **IDDSI framework** https://iddsi.org/framework
The IDDSI Framework provides common terminology to describe food textures and drink thickness.

Oral Defensiveness

Babies who spent long periods of time in a Special Care baby unit (SCBU) or Neonatal Intensive Care unit (NICU) gaining nutrition through a feeding tube, often have oral defensiveness. As a result, they won’t have learned to co-ordinate sucking and swallowing at the proper developmental age and they will have regularly had to deal with people putting tubes in their mouth or nose. Overcoming oral defensiveness takes time and patience. Parents can use touch by play, either with fingers gently touching a child’s face or using soft objects, or their own fingers. Singing games can help with body awareness, so they can also be used for facial awareness. If they can tolerate some of these touches, try the area around the mouth and lips before feeding. Then bring food to their mouths slowly and gently using only plastic or rubber spoons, as metal ones are cold, sharp and hard. Gum massage can also help oral stimulation.

> *“ My son has low muscle tone and sensory processing disorder. I have him drink yogurt through a straw to address these issues. We also have to give him small pieces of food one at a time, because he stuffs his mouth. Again, it's a sensory thing for him. As an infant we had to rub the inside of his mouth to help with his gag reflex. He is 3.5 now and continues to have OT therapy to help him use a spoon. ”*

♦ **Sensory Processing Disorder (SPD)**
https://www.sensory-processing-disorder.com/
A website for everything connecting to sensory processing disorder, also known as sensory dysfunction, including information for oral defensiveness and picky eaters.

Pica

Pica refers to eating objects which are not meant to be eaten. Young children are often curious about everything and put things in their mouths. Some children and Adults with a rare chromosome or gene disorder can carry on doing this; putting things in their mouths like stones, poop, coins, acorns, leaves, etc. Whilst some objects pass through the body without harm, pica can be very dangerous.

♦ **The Challenging Behaviour Foundation** has a very useful information sheet on Pica: https://tinyurl.com/2knycsrs

Avoidant restrictive food intake disorder (ARFID)

Avoidant restrictive food intake disorder (ARFID) is a condition characterised by a person avoiding certain foods or types of food, having restricted intake in terms of overall amount eaten, or both.

♦ **Beat eating disorders**
https://www.beateatingdisorders.org.uk/get-information-and-support/about-eating-disorders/types/arfid/

Constipation

Constipation is when you're not opening your bowels as regularly as you should, or you're unable to completely empty your bowel. Constipation can also cause your stools to be hard and lumpy, as well as unusually large or small. There may be other symptoms as well. It can be acute (sudden and severe) or chronic (ongoing and constant). It is a very common condition that can affect people of any age. For some people they can develop faecal impaction, which might need further treatment.

Causes of Constipation

There are many triggers that can cause constipation and associated symptoms, which for children with a rare chromosome or gene disorder can include:

- ⇒ Lack of fluids
- ⇒ Diet e.g. not eating enough high-fibre foods
- ⇒ Some medicines, especially pain killers
- ⇒ Ignoring or inability to understand the urge to open the bowels
- ⇒ Lack of exercise
- ⇒ Emotional upset.

How to Prevent Constipation

Speak to your GP or child's paediatrician or health visitor, before making significant changes to their regular diet. If necessary ask for a referral to see a dietitian, who will be able to offer advice and support.

Constipation overflow

Constipation overflow is when the poo in your bowel is so hard that you can't push it out in the usual way. So your bowel leaks out watery stools. The watery stools pass round the poo causing the blockage and out of your rectum. The leakage can look like diarrhoea.

How is constipation overflow treated?

Your GP can prescribe laxative medication e.g. Macrogol, which is a powder that is mixed with water. If this doesn't work, your child might need an enema, which is special liquid that your doctor inserts into the rectum to soften the stool and help your child to pass the poo more easily.

- ♦ **NHS** https://tinyurl.com/3w3kr8ne
- ♦ **Patient** https://patient.info/digestive-health/constipation/constipation-in-children
- ♦ **Faecal impaction in children** https://www.nhsaaa.net/media/1580/20170222faecimpac.pdf

Massage

Abdominal massage can help children and adults with constipation.

- ♦ **Baby Massage For Constipation wind and gas (video)** https://www.youtube.com/watch?v=Fcn6gVX5y4I
- ♦ **Digestion massage** https://www.babycentre.co.uk/l25014847/how-to-massage-your-baby-aiding-digestion-photos

- ♦ **Self-abdominal massage for adults** https://www.wchc.nhs.uk/content/uploads/2019/12/Self-abdominal-massage.pdf

> *“ I hate giving my son laxatives. I only do it in extreme circumstances (when he hasn’t opened his bowels for 3/4 days). Several years back, I asked our continence nurse advisor if there was a more natural solution and she said to give him 4 or 5 spoonfuls of ground linseed (or flaxseed) in with his morning cereal, which I did. It must be the seed and not the oil, as the body absorbs the seed better than oil apparently. It has helped a lot, I give it to him every morning (I buy big 3kg bags off amazon but to start off you can buy 500g, 1kg etc. I buy ground golden linseed. Also available for health food shops). ”*

Drooling (dribbling)

Drooling (dribbling) is when saliva comes trickling out from the mouth. It occurs naturally in babies, but as a child gets older it usually stops. Daytime drooling stops within the first few years of life, though it will still occur in an older child/adult during sleep.

Drooling can be caused by:

- ⇒ Low muscle tone (Hypotonia)
- ⇒ Abnormalities in swallowing
- ⇒ Difficulties moving saliva to the back of the throat
- ⇒ An unstable jaw
- ⇒ Limited jaw and lip closure
- ⇒ Tongue thrusting
- ⇒ Reduced awareness
- ⇒ Seizure activity

Drooling is fairly common in children and some adults with chromosome and gene disorders. Children born with a cleft lip and palate, a high-arched palate or hypotonia can be affected by drooling. It can also cause a sore mouth and chin and clothing often needs to be changed regularly because of the wetness. There are treatments available to help with drooling.

Treatments may include:

- ⇒ Oral skills activities
- ⇒ Medication, where drugs might be used to dry up salivary secretions (Hyoscine hydrobromide patches [also used to treat travel sickness] or Glycopyrronium Bromide tablets/liquid are two of the most common medication treatments used for drooling)
- ⇒ Botulinum toxin (Botox) injections
- ⇒ Salivary gland surgery

- ♦ **Scope** https://www.scope.org.uk/advice-and-support/drooling/

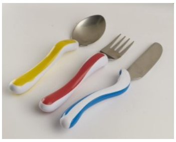
Growing up……………

For many children who have problems feeding as a baby, these problems may continue as they grow up, but not always. Several of our parents have found help and solutions of their own.

> “ *Have a little patience! Easier said than done, I know! …. My daughter has never had a problem as such, with eating, as she has always loved food, but she has never had a 'stop switch' and just wants to continue to eat. As a baby she would always feed until she was sick and then want to continue again. So, I learnt very early on, that I had to take control. I would just stop feeding when I thought she had eaten enough. As she has got older and found her own voice (she didn't start talking until she was about 8) this has become harder because she loves her food. But I have remained strong (mostly!)! She is now 15 and accepts that she (and I realise no two children are the same; but this works for my daughter) cannot have second helpings of school dinners or puddings; she has learnt what is an acceptable amount of food on her plate at a buffet; she has also learnt to ask for healthier alternatives as snacks between meals; more importantly, she has actually learnt to say 'No, thank you.' to the offer of more food…! Anything is possible! Good luck everybody!* ”

Equipment to help with feeding

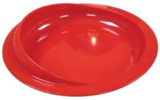
There are a huge variety of pieces of equipment that can help with eating and drinking including specialist plates, bowls, cups, caring cutlery, etc. Specialist seating may help to support your child's posture at mealtimes. Larger items of equipment like a feeding chair can be provided by an OT, but most of the small items like plates and cups, you will probably need to fund yourself (check with your OT as even if they can't fund items on the NHS in the UK, they should be able to advise you in finding the right equipment and direct you to funding to help pay for them). There are several different disability aids websites where you can purchase items like specialist plates, bowls and cutlery etc.



- **Fledglings**

https://www.fledglings.org.uk/collections/eating-drinking

Feeding aids and many more items, including oral and chewing aids, for children and adults.



> “ *K. has a special plate that's slanted and special cutlery from her OT. It really does help! She eats with little or no spillage, but only with these. For K it was soft plastic spoons, different textures and colours that helped. Don't set your own goals too high and at the start being prepared for lots of breaks. But persevere.* ”

Glossary

Aspiration - inhaling fluid, food or saliva into the airway or lungs.

Aspiration Pneumonia - occurs when food, saliva, liquids, vomit, or foreign objects are breathed into the lungs or airways leading to the lungs.

Barium swallow – Barium liquid (often fruit-flavoured). You stand or sit in front of an X-ray machine whilst X-ray pictures are taken as you swallow. This test aims to look for problems in the gullet (oesophagus). These include a narrowing (stricture), hiatus hernias, tumours, reflux from the stomach, etc. A barium swallow test takes about 10 minutes.

Cleft Lip - a split in the upper lip. A cleft occurs if, early in pregnancy, the separate parts of the developing baby's face (in this case the palatal shelves) don't join together as expected.

Cleft Palate – where there is a split or gap in the roof of the mouth.

Gurgly voice – voice sounds wet or bubbly.

High-arched palate – the roof of the inside of the mouth is higher than usual.

Hoarse – the voice sounds rough or harsh.

Metabolic - metabolism is the process your body uses to get, or make energy from, the food you eat.

Nutrition - the process of providing or obtaining the food necessary for health and growth.

Oesophagus – (also known as the gullet) - the muscular tube that conveys food from the pharynx at the back of the mouth to the stomach.

Oral – relating to the mouth (as in oral hygiene).

Pharyngeal – relating to the Pharynx - the back of the mouth and throat, with its surrounding membrane and muscles, that connects the mouth and nasal passages with the oesophagus.

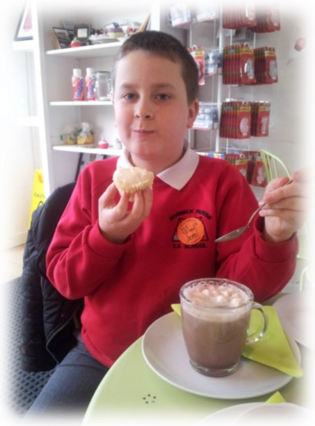
Pneumonia - lung inflammation caused by bacterial or viral infection, in which the air sacs fill with pus and may become solid. Inflammation may affect both lungs (double pneumonia) or only one (single pneumonia).

Videofluoroscopy (VFS) - a moving or dynamic X-ray that looks at the way your swallowing works. It is one way of confirming the presence of aspiration because of problems with swallowing.

Notes

> *"My son wouldn't eat anything much, or ate the same thing over and over again, he didn't drink much either. I bought a smoothie maker and started giving him fruit smoothies; he really enjoys them."*

Notes



> “ C.'s issues are mainly psychological. I read him lots of books about food and meal times. We also drew pictures of the food and talked about it before eating. He has tried 'caring cutlery' but it hasn't helped much. Numbers and letters are his motivators so I add Alphabetti spaghetti or similar to his food. We try hand-over-hand, where I hold his hand under my hand when feeding. We tried lots of desensitisation techniques around his mouth with cloth textures. Family meal times with us around the table seem to be counter-productive, so we are trying different places and using distractions, such as reading to him whilst he eats. ”

Inform Network Support

Unique
Understanding Chromosome & Gene Disorders

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Join Unique for family links, information and support.

Unique is a charity without government funding, existing entirely on donations and grants. If you can, please make a donation via our website at www.rarechromo.org Please help us to help you!

Our thanks to all of the parents that contributed towards this guide.

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