



The Information Point
for Centronuclear and Myotubular Myopathy

★ Information ~ Support ~ Awareness ♡

Established March 2001

Frequently Asked Questions

This document has been put together with the aim of answering the most frequently asked questions of parents with newly diagnosed MTM babies. The responses have been provided by parents who have been through the early stages of this condition - it is **not** intended to replace medical advice but to augment it and to allow parents to make well informed choices when faced with difficult decisions. The information would also be of use to medical professionals who find themselves caring for an MTM baby for the first time.

1. Where can I obtain a molecular diagnosis?
2. My child was recently diagnosed with X Linked MTM, he is still in the hospital and has never been home. I need to learn a lot regarding daily care.
3. I am concerned about transitioning my child from breastmilk to formula, can anyone offer any advice?
4. Low Sodium Levels: Has anyone had experience with this?
5. I have seen in references to Chest Percussion Therapy. What is this, how you do it, when and why you do it and where I would learn it? Also what is physio? How do you know that they need CPT? What are the signs?
6. My son is 7 weeks, I would like to know what do you feel, looking back, what were some of the more valuable pieces of advice, things to do, etc...that benefited your child at this early age? What were your biggest problems with your child and how did you learn to deal, overcome, augment these?
7. Suctioning - how do you do it? I seem to be reading that most prefer to go thru the nose? Is one way better than the other? Should you not do it one way? Can you describe how, when and any additional suctioning tips?
8. Secretions - when your child was a babe were you able to correlate anything with the thickness / ability to manage their secretions? When did you see the secretions improve or become less of an issue?
9. Do you use humidifiers? Are they helpful? In what way and when?
10. Would like information about a Nissen. Did your child have reflux (do they all)? Do you regret the nissen or feel it was a good decision?
11. Constipation: my son pooped fine the first 4 weeks, now he doesn't. They have been giving him glycerin every 4 - 5 days to poop and has done it once on his own since...but it seems to have stopped. Why? Have you experienced this? Do kids with X-Linked MTM have issues with this as well? How do you manage and does it get better?
12. When my boy is in a deep sleep (as all 8 weeker are!) drifts and his respiratory can sometimes go as low as 5...they say he has central apnea...what can you tell me about this? What is your experience?

13. Continuous feeds: S right now has an ng tube and they switched him yesterday to a lower feed volume and it is on continuously. They thought maybe his belly wouldn't be as full, reflux less, secretions less bothersome...any thoughts / experiences around this?
14. Does swallowing improve with time? The doctors and ourselves are discussing the possibilities of a G-tube.
15. How do you manage siblings when you have a 'sick' child?
16. What types of therapy programs you did with your child from infancy? The state programs with PT, OT and speech were mentioned. What did you do, how often and at what ages? What did you feel was very important?
17. From the limited info that we have been able to gather so far it would seem as if you need to live in a bubble for our son's first year of life? Colds, infections, etc seem as though they could be devastating for him. I look at him struggling with his 'normal' secretions now and wonder what would happen if he got a nasty upper respiratory virus? How did you all handle life, I guess, is the question here. Family gatherings in the winter time, being around other children, going places? What types of precautions did you / do you take?
18. I understand that some of the boys have had tubes / grommets in their ears, and it has made me think that R could possibly need something similar, he is still not talking and I think this may have something to do with his hearing. What I need is more info as to how and when any hearing problems were picked up on in your boys?
19. My son seems to have irritated skin – is this linked with MTM?
20. I was wondering if anyone was aware of any contraindications for X Linked MTM to any vaccines? Has anyone's child had a serious reaction to any specific vaccine's, my child has just turned two months and the docs are asking us about vaccines.
21. I would like to know if anyone has experience of heart shadows and X Linked MTM. If so what was the outcome?

Q1:Where can I obtain a molecular diagnosis?

Answer 1 of 1:

The two best methods of confirming a diagnosis are a muscle biopsy or genetic testing. Usually a muscle biopsy is the first step, as a patient with negative genetic testing may still have a form of Myotubular or Centronuclear Myopathy.

In Europe Molecular Diagnosis is available from:

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Q2: My child was recently diagnosed with X Linked MTM, he is still in the hospital and has never been home - I need to learn a lot regarding daily care.

Answer 1 of 4

My son is 2 years and 3 months old diagnosed with the 'milder' form of X Linked MTM too and was always able to breathe in air but as with your son, my son's secretions were and have been the biggest issue. I always say that the physical disability is just a non issue in comparison to the trickiness of secretions. I don't know what Rubinol is - is it supposed to dry up secretions because if it is, we never found those kinds of drugs helpful - they just made secretions too thick to manage. And in fact our local (we're based in London UK) neuromuscular centre where the doctors are world experts on MTM told us they don't favour anything that dries up secretions in these children.

If my son had presented the way you're describing we would have assumed he had a virus, and the things that worked for him are as follows (and by the way to our great joy and eternal gratitude he has NEVER had a chest infection despite all dire warnings that it was inevitable:

- lots and lots of rest and quiet
- positional drainage i.e. moving him from left side to right side intermittently to make sure nothing drips into or sits on his chest. If he can't tolerate it for long, at least try it for as long as you can and then rest him in between.
- nasal suctioning - he keeps so much gunk in his nasal passages it's unbelievable, but a regular good naso-pharyngeal suction is really important
- lying on his tummy - I know that babies are not supposed to be put to sleep on their tummies but as he was being watched all the time and certainly on saturation monitors we always rested him on his tummy once we realised what a phenomenal difference it made to his ability to breathe when he's sick and weak. Now that he can tell us what he wants when he's sick he always wants to lie on his tummy, and if he's too weak to roll completely over by himself he'll just keep pointing at his bed or floor mat just in front of his face until he's put flat. He even does it in his sleep!
- when he was very small (under 1) and sick we would drain him over a big pillow on our laps - it seemed to really help to bring things up (but he needed a big rest after any such session)
- cutting feed volumes but just making them more frequent; the effect a full tummy can have on his ability to breathe is just astounding. We can have given him a feed (he has a gastrostomy) and he can be saturating quite low and obviously struggling and the minute we extract some of the feed the problems go away. It seems that he relies on his diaphragm so much to breathe that if a full tummy pushes up against it he can really struggle, particularly if he's weak and sick and his intercostal muscles are working even less.
- watching for constipation, again a full intestine can push up on his whole system to his detriment.
- when he was as little as your son, if a virus was particularly exhausting him, a little oxygen in a head box (a clear Perspex box that goes round the baby's head) with some added moisture really really helped. The last time he needed that was just before he was 1 and he had about four other instances. He never needed it more than 2-3 days.
- keeping fluids up - when I tried to find out about managing secretions I read a lot on the MDUSA website and read the descriptions from adults with other muscle weaknesses about how they managed their secretions and what it felt like not to be able to swallow them (I figured someone old enough to describe the sensations might provide useful advice) and every single thing I read seemed to talk about extra water, more and more water. Our boys lose a lot of fluids through suctioning - no one really knows how much you and I are swallowing that our boys get taken out but our physio was at a conference recently that said that we all produce 1.5 litres of saliva a day! Once we upped his fluids beyond the 'normal' age limits of total fluid intake his secretions seemed much much easier to manage, and he started having days and nights where he wasn't suctioned at all. That said though he still needs masses when he's sick - the minute he coughs something up when he's sick we suction it away so he doesn't have to exhaust himself by dealing with it.

Due to the wonders of the UK national health service we get respite care every week, which our private health insurance then tops up. That means he has a night nurse every night so he is never allowed to lie in the one position for too long so that nothing slips down deep into his lungs. (his position is changed hourly) We wonder if that is one of the big reasons why he has done so well from a respiratory perspective.

The other really important thing for your little boy at this time of the year is to get an immunisation against RSV - it's a virus that is really really hard on strong small babies, and disastrous for ours.

I so remember 7 weeks, he had been due to come out of hospital the week before and he got a virus and had to stay in an extra 2 weeks. It was a highly emotional time and since then we have had many ups and downs but the little man who's emerging from all this is so funny, strong, calm and so incredibly happy with life. They are so amazing these boys, and while there are no miracles (yet!) there's so much they can do.

Answer 2 of 4

I can tell when it is going to rain by how much secretions K has. When the weather changes to cold it gets thicker for a couple of days. When it changes to warm it thins out. When we go to the mountains, I have to increase his water intake or his secretions will be thick. He is our little weather man.

Answer 3 of 4

Our son (now 10 years old) needed a suction machine daily for about the first 2 years of his life, after which we introduced the nebuliser with "Saline" and "Atrovent" initially and then subsequently salbutamol (miracle drug) which helped him enormously, and gradually weaned him off the suction machine. You may find that your little boy is too young for salbutamol (which opens up the airways). In my experience, I have always felt it important to keep his chest as moist and loose as possible with nebulisers or even "steam" (steamy room/baths). I also found it very useful to learn chest percussion and positioning and gave it to him at regular intervals (minimum 4 times to begin with before suctioning). I also did regular baby massage which I felt made up for the time I had to physio him. I know a lot of physios will recommend positioning before percussion these days, but my physiotherapist was of the "old school" who thought that firm chest percussioning (and she almost seemed to be brutal with such a delicate, weak baby) would not only help shift secretions from sticking on the lungs but also help build up muscle/strength in his back. He is doing pretty well these days and I often wonder if she was right in her assumption! She also started doing physiotherapy with him to get him moving from an early age. You could start off with a baby gym (one that hangs over them) and try to encourage your little boy to kick it with his foot or bat it with his arm - do it for him to begin with until he one day has the strength or the inclination to do it by himself!



Q3: I am concerned about transitioning my child from breastmilk to formula, can anyone offer any advice?

Answer 1 of 3

We persisted with the less broken down formulas for longer because we didn't twig the connection with the desats, so T's system may have had time to become a bit more

desensitised and then he tolerated Neocate very well. However here are some ideas. You could try diluting it? We accidentally fell upon diluting T's milk 10 months ago (he had terrible stomach cramps from an antibiotic** combined with Dom Peridom) and half water half milk seemed to upset him less. We kept it up and since then he has never tolerated his food better. We basically combine his required milk and water intake for a 24 hour period and then make each feed 50% of each.

We also feed T every 1 and a half hours. I know that sounds mad BUT it works SO well. When he first came out of hospital I was DESPERATE to get him on a 4 hour routine so he could do things in between, but as we were constantly trying to push up the volumes we'd have to try and keep him still and lying down for ages, just to prevent reflux and aspiration. THEN again at the same time as diluting his milk, we stumbled upon giving his food so regularly. It meant he was getting an easy volume to digest and it was so little that we could feed him in 10 minutes and get on with things. In fact we calculate it so that the 1 and a half hours round the clock would give him too much milk, so it's perfect because we can take a longer break here and there and catch up later but not by having to give a bigger individual feed. For example yesterday I fed him at 8.30am and then not again until 12 because we went out for the morning. He still got all he needed in the 24 hours because we spent the rest of the day at home and he had a feed on the button every hour and a half. Overnight we give him an even smaller volume. Something between 100ml and 120ml every 90 minutes depending on how the day has gone. (He gets 160ml every 90 mins during the day, and when we were feeding him 4 hourly we were trying 220ml).

It is indicative of there being a digestive issue if the child is pooing less than usual. Because it has higher osmolarity the big risk of Neocate is that it is SUPPOSED to cause diahorrea (more osmosis of water into the bowel because there is a greater number of 'particles' per calorie because it's more broken down). HUH is all I have to say to that! We have even had to CONCENTRATE up T's Neocate - if his digestive system was operating like a regular child he'd be permanently pooing. Our medics were just stunned for ages that T could even be constipated on Neocate - never heard of before.

I remember that battle so well, and in the end I stopped trying to explain myself, and used the broken record technique 'yes, I understand, but I believe Tom will do better on Neocate', 'of course yes I hear you, but I believe Tom will do better on Neocate', 'thanks for thinking that idea through, but I think Tom will do better on Neocate'. Always with a smile! It was a long meeting but it finally worked! (That was the same meeting that it was suggested I put T in a respite home to give myself breaks now and again, and when I said that things would have to be really bad for me to ever put EITHER (why should Tom go in a home, when no-one would ever suggest Sophie go) of my children in a home, they tried to explain to me why in 'their' opinion it would be better for me. It was a bit of an exhausting day! Although the paediatrician did apologise later!)

Other suggestions - why don't you ring the makers of Neocate and ask them if they can think of anything. Also keep a very very tight diary of what's happening - time of feed, volume, timing of respiratory issue, poos, anything else AT ALL.

** Hint: Never combine antibiotics with reflux drugs that empty the stomach quickly - antibiotics are supposed to be digested in the stomach and if rushed into the gut can be less effective and some can even be painful. Augmentin was particularly painful for T, he was in total agony

Answer 2 of 3

I can't offer much help or advice because Z was never bottle or g-tube fed, but I breast fed him until he was 14 months. All I can add is that Z obviously took in as much milk as he could cope with.... which was very little but very frequently. In fact, in the end he only fed on one side (which made me a bit lopsided at the time!) but I had a fantastic lady supporting me, who was doing research at Bristol University into benefits of breast feeding and she advised that this was fine because many women with single mastectomy can feed their babies adequately well on one side only. For the first few months of life, at least until Z was on solids, he was in the lowest centile of weight. I was given a lot of grief by my health visitor, but I used to lie about how much milk Z took in. (Very silly and naïve looking back – as I was so desperate to keep things as normal as possible and there just wasn't this fabulous information around when Z was a baby – we didn't even know of other families with MTM and we were definitely on our own!) My instinct is that breast milk is a lot “thinner” than formula but he coped with as much as he could and somehow, managed to thrive and get through his first year. When we moved to the Hammersmith (when Z was 18 months) they advised that Z would never have been allowed to breast feed and that he urgently needed a gastrostomy (which we resisted because he had coped for so long without one). Fortunately over the next 6 months, his swallowing improved and they agreed that we could persevere with feeding by mouth. The advice you have been given is absolutely excellent – it sounds like thinner, little and often is the answer.

Answer 3 of 3

The feeding issue is a frustrating one. We had similar issues with J. After trying a variety of formulas (when we were transitioning him from breast milk), we finally landed on on Pepdite 1+. It is great. J tolerated it from the beginning. It is one that is not that well known, but it has been a lifesaver. We make it at 3/4 strength which helps. Don't give up. Keep insisting. Obviously this is an issue with the boys! Once we put J on this formula, we haven't had any problems. He tolerates it great, has grown! He gets 150cc 4x a day and 59cc per hour at night for 8 - 10 hours.



Q4: Low sodium levels: Has anyone had experience with this?

Answer 1 of 1

I talked to the doc and then I dug back in L's old charts and found the low sodium problem you were wondering about. Turns out that infants not being able to process sodium and showing low sodium levels is quite common - especially when / if premature (they end up with premature kidneys but the kidneys look normal 'cause the rest of the body is also premature). The doc then reassured me that it wasn't something that was dismissed but it did take care of it's self relatively quickly (a couple of weeks). So apparently, not to worry! I just remember overhearing something one time about L having this sodium deficiency and sure enough...I found it. If you have / your doc has questions you can always call other hospitals

that have dealt with kids with this (MTM) and sometimes those docs can seem so reassuring when all of a sudden you hear some speak of MTM like they know what's goin' on!



Q5: I have seen in some references to Chest Percussion Therapy. What is this, how do you do it, when and why you do it and where I would learn it? Also what is physio?

Answer 1 of 7

Chest percussion (CPT - Chest Percussion Therapy) and physio are the same thing. It is when the respiratory therapist (or loving parent) pound (for lack of a better phrase) on the child's chest, back side to loosen secretions inside the lung. This makes it easier for suctioning. It works great because often the boys can't cough strong enough. This is a great assist!

Answer 2 of 7

Get a physiotherapist to show you how to do it properly...they will show you how hard to pat the wee one's chest and sides and back. Try using cupped hands rather than a flat hand..seems to work better and is less shocking to the wee one...rolling the child from one side to the other and suctioning alternately helps move the secretions also....know that after a physio session the crap in the lungs will continue to move for a while (1/2 hr or so of constant suctioning is not uncommon in L's case)... Keep up with the questions...it's the only way to learn quickly...it'll feel like an unbelievable landslide of info initially but it will settle down with a little time and be a little less overwhelming....

Answer 3 of 7

The first time CPT was done on our son it was with a CPR mask. The respiratory therapist at the hospital put a piece of tape over the opening and it worked great. Since then we've used the Vest, a hand-held vibrating device, and the little blue percussor cups. He doesn't require nearly as much CPT as he used to, which was 5 - 6 times a day for 10 - 15 minutes. Now it is usually done 1 - 2 times a day, depending upon how he is feeling.

Answer 4 of 7

Keep fluids up as they need to be well hydrated to maintain good lung function. Ensure that he has regular checks for night-time hypoventilation (under breathing at night due to the diaphragm being weak and intercostals (in-between ribs) being weaker. Its something that may develop or may never develop and can be easily maintained with oxygen overnight. Z has had a few "borderline" results which were scary at the time, but his lung function has improved dramatically with age (although we still have regular checks). He does lots of lung exercises now and even has a lung exercise machine which he's been using since about 7 years of age and uses for 20 minutes every day to expand his lung capacity. This has directly resulted in flattening his concave chest and increasing his lung volume!

Answer 5 of 7

Would ask the respiratory therapists and / or the doctors to train you to listen with his lungs with a stethoscope and get to know what is "normal" for him....this will be invaluable. I can hear when J's lungs are "tight". Usually the best indicator that J needs CPT is increased secretions with difficulty clearing them. J has never had a fully-productive cough, although he is getting better.

Answer 6 of 7

My advice is that you need to get into a routine of doing CPT every day at least 3 times a day, whether they need it or not in the first couple of years! And listen to their chest every day and you will quickly learn what is normal and what is not!

Answer 7 of 7

The PEP treatments combined with Chest Percussion Therapy (CPT) and suctioning will definitely help with the rattly sounding chest!



Q6: My son is 7 weeks, I would like to know what do you feel, looking back, what were some of the more valuable pieces of advice, things to do, etc...that benefited your child at this early age? What were your biggest problems with your child and how did you learn to deal, overcome, augment these?

Answer 1 of 1

Our son is now 10 years old and his chest was always our main issue: it was a case of “just coping” for the first couple of years, but things did improve for Z as he grew older and stronger. You are in a great position because:

There is the internet and internet support groups with goodness knows how many families’ experiences (not around when Z was a baby – we thought Z was the ONLY boy in the whole world affected by MTM – we were very much ill-informed and “in the dark”).

The prognosis used to be very “doom and gloom”, but doctors are better informed about the condition and medicine / treatment has moved on tremendously and also you will get a lot of useful information from other families. (Our boys are longer surviving due to modern medicine and well-informed parents). There is a fairly recent paper entitled “A long term view of MTM” written by Prof Bart (Netherlands) and Prof Dubowiz (Hammersmith, London) which talks about a family of Dutch adult males, more mildly affected by MTM and still surviving in their 50’s. There are several informative sites: USA and Europe based which will give you an all round perspective of the condition. Advice I would give is:

1. If possible, ask for a referral to a specialist in MTM and possibly a lung-specialist hospital. There are 2 hospitals in the UK, but I'm not sure of the situation in USA? In the UK you can have your child seen by both specialist hospitals and local hospitals (for acute care), but again, don't know the format in the USA.

2. Always to go with your gut instinct. If you are not sure whether something the doctors are saying is correct, give it full research and time before you make any decisions and try to get a second opinion. If you are well-informed your doctors will query their own actions too. Above all, don't worry about writing things down, asking doctors to repeat something you don't understand or asking the doctor to give you a better explanation - they are not Gods and you have not just enrolled into medical school!

3. Seek the best physiotherapist you can who will not only teach you how to look after S's respiratory needs, but also his physical needs. She will teach you how to suction (down the nose) and give chest physiotherapy. Ask them to show you how to use a stethoscope and listen to his chest every day. At first you will wonder what you are listening for: but if you do it every day you will hear if / when something is abnormal.

4. This might sound weird too, but I tried not to treat Z as if he had a "disability" and did everything I would if he was "normal". I took him to a baby massage class which also helped babies to get moving and exercising. Sometimes it was utterly hopeless as he couldn't do half the things his peers could but I think it has given Z that determination to try everything that his friends do in later years. There are plenty of ways around things / equipment to hand which will help them lead a normal life. When you start exercising him, don't worry if nothing happens to begin with and you might have to move his limbs for him.... it will soon come! Exercise in a warm bath, followed by a massage with "sweet almond oil" is THE BEST!

5. Live for today. Try not to worry about the future. You probably wouldn't think so long and hard about it if you had a child who was "normal". If you take care of today, and enjoy every little progress and thing about your gorgeous little boy, then you won't have to worry too much about tomorrow.



Q7: Suctioning - how do you do it? I seem to be reading that most prefer to go thru the nose? Is one way better than the other? Should you not do it one way? Can you describe how, when and any additional suctioning tips?

Answer 1 of 1

Suctioning is an important thing to be proficient at! When J left the hospital we were suctioning frequently. Luckily, we had a great nurse (who I have thanked profusely over the years!) who taught me how to NT suction (through the nose). J used to carry so much stuff in his nose, I was surprised he could breathe. We would also suction his mouth, but keeping his nasal passages clear is essential!



Q8: Secretions - when your child was a babe were you able to correlate anything with the thickness / ability to manage their secretions? When did you see the secretions improve or become less of an issue?

Answer 1 of 2

We never really made any correlations although keeping on top of the secretions, doing CPT is essential.

Answer 2 of 2

The secretions started getting better as he got older and better able to manage them. They are always worse when he is sick (like now!) Sometimes I think we will float away!



Q9: Do you use humidifiers? Are they helpful? In what way and when?

Answer 1 of 1

We use a humidifier now that J is trached. He also used bipap before and we humidified it.



Q10: Would like information about a Nissen. Did your child have reflux (do they all)? Do you regret the nissen or feel it was a good decision?

Answer 1 of 1

J got his G-tube (which has been a lifesaver although frustrating now that I want him to eat..Cross that bridge as we go across it!) when he was a few weeks old. He did not get a Nissen at that time, although I am not sure why. He has reflux and finally got a Nissen about a year ago after MANY bouts of aspiration pneumonia!



Q11: Constipation: my son pooped fine the first 4 weeks, now he doesn't. They have been giving him glycerin every 4 - 5 days to poop and has done it once on his own since...but it seems to have stopped. Why? Have you experienced this? Do kids with X-Linked MTM have issues with this as well? How do you manage and does it get better?

Answer 1 of 2

Z has had "Domperidone" (Motilium) since he was a baby to help with constipation. This improved with age and he's absolutely fine now.

Answer 2 of 2

Something that really helped with S's constipation was flax oil. We didn't discover this until he was older - around 9 months - but it is a fish-based protein oil that you can buy in the refrigerated section of an organic grocery store, like Whole Foods or Wild Oats. Since we have discovered it, we've always mixed it in with his formula, and I also mix it into his vegetable and fruit purees. Not only did it help with constipation, it also is loaded with calories, and since putting on weight has always been an issue with S, we were grateful to have found this supplement. Once your little boy gets a little older, prunes and prune juice also work just as well as you would imagine they would!!



Q12: When my boy is in a deep sleep (as all 8 weeker are!) drifts and his respiratory can sometimes go as low as 5...they say he has central apnea...what can you tell me about this? What is your experience?

Answer 1 of 1

J had some slight apneic (sp) episodes when he was very small, but it improved and was not an issue when he came home.



Q13: Continuous feeds: S right now has an ng tube and they switched him yesterday to a lower feed volume and it is on continuously. They thought maybe his belly wouldn't be as full, reflux less, secretions less bothersome...any thoughts / experiences around this?

Answer 1 of 5

My experiences with:

Suctioning:

Nasal suctioning should be limited as the membranes are thin and susceptible to damage (causing nose bleeds)...maybe twice a day or after a physio session...stick to oral suctioning and keep the depth roughly the distance from lips to ear +1 cm ...this should get the secretions successfully without going into the esophagus (suctioning there is usually fruitless)...as for thicker secretions you may have to go with two different diameter catheters for suctioning as we do (right now we use a #8 for thinner secretions and a #10 for the thicker ones-thickness seems to depend on sicknesses, dry weather, diet etc...) when suctioning push the catheter in to max depth (lip to ear depth) without the vacuum plugged(helps get the tube down without getting stuck on something on the way in...like his tongue, cheek etc...) then plug vacuum and withdraw slowly ...when you see the tube starting to get fluids moving stop and let it suck 'til no more come and let withdraw some more until the fluids come again...repeat til tube is all the way outthen repeat this process....listen to his breathing in

between insertions for the "rattly" sounds...if rattly try some percussion or repositioning to get those secretion plugs loose and moving out of the lungs or wherever they are stuck....goal is a clear breathing sound...(not always possible)...humidifiers or nebulizers will help with loosen the crap in the lungs and throat and make suctioning a little easier...when the secretions are thick they are really hard to suction G-tubes

L has a GJ tube, which is an advanced G tube to specifically avoid reflux as the food is past the point of where it can reflux. Funduplication (Nissen) is something that, it seems, is more common in the States than here in Canada...was never even talked about with us.

L is on a 21 hr feed schedule and it seems to work well for him...I think it might help with keeping him hydrated as well, which keeps his secretions less thick (when people are dehydrated our secretions become quite pasty and thick) although my thoughts on this constant feed thing are only a theory...ask a doc.

Apnea:

L had lots of apnic episodes in his first 4 - 6 months but are occurring less frequently now as his muscle control is a little better co-ordinated....with other kids where the disease isn't quite as severe the diaphragm gains strength and co-ordination and once this happens the apnic episodes also abate...this is also directly linked to viruses...more apnea with a cold. etc...apnea is controlled (although not stopped completely) with (initially) cpap and then when kids gain a little strength a bipap machine (need strength to trigger a bipap machine) these machines assist with breathing so that when resp. drops below a certain #/ min. then the machine takes over....

Answer 2 of 5

J does bolus feeds during the day and continuous feeds at night. He takes Prilosec (Omeprazole) for reflux and that has helped immensely!

Answer 3 of 5

S was really gunky for the first 6 - 8 months of his life and required daily deep mouth suctioning and sometimes suctioning thru his nose. I should say that we were not taught how to suction him through his nose initially, though, and I did feel that it got much more gunk than the throat suctioning did - especially when he was sick and stuffed up - but by the time we knew about it, S was old enough to really protest when we did it, so we reserved it for times when he was ill. The hard thing with suctioning is having the gumption to really get the tube down far enough to make a difference, and making sure that the tube is large enough to get the really thick secretions. I can't remember now what size tube we used on a daily basis with S, but I want to say that it was almost twice the diameter of the infant one they started with in the NICU. For us, the bigger diameter worked MUCH better. I don't know if anyone else has mentioned this, but we also found having a portable suction machine a godsend. It is only recently that I don't travel with this in the car, no matter how far we went from home. We left the hospital with this, an apnea monitor and a pulse-ox machine, and we needed all of them for at least a year (and like I said, we still have the suction machine just-in-case, but haven't used it for a year).

Thinking of car travel makes me also want to mention that when S was under a year old, I was constantly worried about him not being able to support his head when I would need to stop quickly (or sometimes even when I would stop gradually, his head would fall forward

and he couldn't move it back himself). This was really scary for me, and we quickly realized that he needed to have additional support around his neck in the car. At that time, those little 'mini boppy' head protectors had not come onto the market, so we manufactured our own, and also bought padded neck guards that could be wrapped around the straps of the carseat restraints to support his head. Now I've seen in some baby stores that they sell a little 'mini boppy' that looks like a boppy but is really small and goes around an infants head in the car. I think this would be perfect, and save your piece of mind on the car trip home from the hospital!

Humidifiers are really helpful for us all of the time (it is very arid in Colorado), but especially when S is sick. When he is ill, I often just run a really hot shower and we sit in the bathroom and let the steam loosen him up. We have found that a warm mist humidifier works best for us.

S has a G-tube and a Nissen, and we do not regret the Nissen, because S did have really terrible reflux. I think that even if we didn't have the tube, we would have needed the Nissen. (And, I'm still not sure about the tube - I think I'm glad that we have it!).

S was on continuous feeds for about 3 months, and then we switched him to bolus feeds (where you use a really big syringe and the tube, and let gravity take the food in). We did bolus feeds during the day, and used a kangaroo pump (ask about this at the hospital, it is really hard to describe) at night until S was just over 9 months old. We also started giving him a bottle around 3 months, which was great for his oral development, even though it wasn't a big source of nutrition. 3 months is really the 'magic' number in regard to oral skills (so we were told), in that it is important to try to have them try oral feeds by three months, or it is harder to learn the skills. We also encouraged S to use a pacifier (dummy), and I think this really helped with his oral skills (but he still has it a 2 1/2, which we don't mind - it is only for sleep - but my mother is always asking after it and if we have had the 'pacifier fairy' take it away!! Ha!!) But, in regard to the pump feeds: we feel that the pump feeds contributed to the failure of his Nissen, because even though they went in really slowly (I can remember having the setting on 80ml per hour for forever), I think that the fact that they were being 'pumped' in contributed to pressure on his Nissen, and helped it to fail. This is just my opinion, however, and the doctors didn't feel that it made a difference. Regardless, once S had the Nissen and G-tube redone, we switched totally to bolus feeds, and now S eats three meals of purees, yogurt, cheese, crackers, milk, etc, and we only give him a 6 oz bolus of 'tube food' at night.

Answer 4 of 5

J, now 3 yrs 8 mnts (WOW!!) was born at 40 weeks and spent 6 weeks in the NICU. He came home with a trach, G-tube, and nissen fundoplication. J spent several weeks the exact way you described your boy - very gurgly, "wet", junky - all of that. He would aspirate, code, they would ventilate him, wean him a few days later, then the process would start all over. Eventually the only thing we could do was trach him. I'm certainly not saying that is the answer for your boy, but it was for J. I believe it helped him to thrive as a baby and now he is rarely sick. Of course regular ole' childhood illnesses do get the best of him, but he is a fighter. Now we use a ventilator, which was started when he was about 11 months old. He isn't ventilator dependent, but he uses it at night, and it helps so much when he is sick. It is kind of a preventive measure as well. Others here may / may not agree with this, but giving

these kids oxygen is not always the best bet. Yes, there are times when it is required. But most of the time it masks the underlying problem which is hypoventilation, which requires vent support in these kids. For instance when J is sick and his oxygen saturation begins to drop, putting him on the ventilator almost instantly brings his saturation back into a normal range. J started on Robinul when he was about 6 months old and has been on / off since then. Currently he is getting a very minimal dose - 3cc per day - to help with his oral secretions. However, this is counterbalanced with regular nebulizer treatments and CPT to prevent any thick secretions from collecting in his lungs. We have always suctioned through J's mouth. In the beginning, we did do some nasal, but mostly his secretions would pour out of his mouth. Once he got his trach there was lots of tracheal suctioning, which can be draining on parents and baby. It took 4 - 5 months for J to get a handle on those secretions and then it wasn't so bad. Changes in weather have always had an effect on J. We live in Florida so it is very humid and the weather is crazy, especially this time of year. Spring is actually worse for him because he has allergies as well. We started him on 10mg Claritin this year and that helped. J does have a G-tube, along with a Nissen. I can only say I wish I had done more research before I agreed to have the Nissen done. Within 6 months, J had a hiatal hernia resulting from the Nissen "slipping."

Answer 5 of 5

We give K extra water when his secretions are thick. The weather has a lot to do with how thick K 's are unless he is sick. K has been on Robinul since he was 4 weeks old and it works great for him. We just have to adjust how much he gets by the weather. We also have a humidifier in the house and a extra one in his room. As far as sodium levels, K takes 1/4 teaspoon of lite salt everyday to help with electrolites, this is through his nutritionist.

Our other son affected by MTM, C, is getting a G-tube and Nissen soon, because he has reflux. Now he is breathing on his own, no assist. But they are talking about him being on a vent at night because his blood oxygen levels drop and his heart rate goes up and down. This seems to be a hereditary thing, because K does it too when he is mad or doesn't want to do something. C is in Childrens Hospital in Washington, DC. K also sees the doctors there.



Q14: Does swallowing improve with time? The doctors and ourselves are discussing the possibilities of a G-tube.

Answer 1 of 4

One thing the Hammersmith doctors told us about T 's swallow when he was a month old, was that in their experience some 'milder' children experienced a big improvement in their swallow over the first 3 months. If by that time it hadn't improved then it may take much longer.

Answer 2 of 4

With regard to feeding issues, these are my experiences:

I was lucky with Z (ignorance on my son's hospital's part and my determination too) in that I was allowed to breast feed him. We never had any feeding observations done until we moved (under specialist hospital The Hammersmith, London) when Z was 18 months old and we were told that his feeding was diabolical and he should have a gastrostomy.

We had managed for 18 months so I wasn't going to give up feeding him by mouth easily and although its been massively scary and a complete nightmare at times, he's now eating perfectly normally. Z, like every boy with this condition, had a very, very weak suck. Fortunately I had enrolled on some breast feeding research programme (linked with Bristol University) and had the best advice and support available from a fabulous woman who came to visit me in SCBU. Fortunately, she arrived the day after Z was born, when I was about to give up the option of breast-feeding in favour of bottle feeding, but she really encouraged me, so at first, my breast milk would go into the freezer until I was ready to try Z on the breast. The only thing I would say here is that Z was my second child, I'd no problems feeding my first child and with all the expressing, Z didn't have to make much of an effort to suck! I was a bit silly and naïve of me because I wanted Z out of SCBU as soon as possible and convinced myself that he was feeding okay. Z did manage to get some milk down him but would frequently vomit and go blue, but it did improve a little bit. He only actually fed on one side – which made me a bit lop-sided (and which I did not tell my health visitor either).

Fortunately, my own sister is a health visitor and she said that women with mastectomies can feed successfully, so I took comfort in that. Sadly, at 6 weeks, then 10 weeks (then 4 more times before he was aged 1) he was back in hospital with pneumonia and left lung collapse (probably from aspirating his milk). As soon as he was 3 months old I started giving him baby rice and then yoghurts (which he tolerated much better because they were heavier and stayed down). I added calorie supplement which started to help keep his weights up. Z suffered 7 episodes of lung collapse and numerous chest infections before the age of 3 years.

Since then, I think his swallowing has really improved. You will have to weigh up the pros and cons of the possibility of your child having a gastrostomy, it all depends how he's been feeding already (by naso gastric tube or otherwise?). If he's not already doing so, try to get him to suck a dummy to strengthen his suck and swallow and if possible, when he's at the appropriate age, tasting foods from your finger and playing with foods.

Answer 3 of 4

My son, S has a milder form of MTM, and he was very floppy and weak at birth. He was on oxygen for 2 weeks, and then on room air for another two weeks before we left the hospital at just over 4 weeks. At 3 weeks, he underwent surgery for a Gtube (he had a nasogastral tube before the G-tube operation). Looking back, I've been trying to think about how I feel about the G-tube (which Sebi still has a 2 1/2), and the positives and negatives it has brought about. On the one hand, I firmly believe that having the tube allowed S to remain VERY healthy, with only one pneumonia to date, and only one chest infection that required hospitalization. I can contrast this with my brother, who also has a milder form of MTM, and who was VERY sick for the entire first year of his life. At the time, my brother was not diagnosed as having

MTM, and no one thought his illnesses might be connected to aspirating food. But, in hindsight, my mother and father feel that his illnesses could only have been caused by food aspiration, because he was always ill when everyone else in the family was fine. So, I guess that, on the one hand, I've been really grateful for the G-tube, because it has prevented S from getting sick, and when he has had smaller colds, we can easily keep him hydrated by putting water or pedialight or whatever he needs in the tube. This has been invaluable during a stomach virus, for example, when we could keep him hydrated and therefore healthier than w/o the tube.

On the flip side, now that he is 2 1/2, and we are struggling with weaning him from the tube, I often think what it would have been like to never have had the tube, and just allowed S to eat 'normally'. It is a hard place to go, because when we made the decision to have the tube placed, the doctors told us that it was the only thing that was keeping us from not being able to care for him ourselves at home. At the time, we just really, really wanted him home, and didn't really think that we would regret our decision. And, I'm not sure that even now I regret it, but when I look back over the first year of S's life, and how quickly he improved and grew stronger, I think that he could have been okay without the tube, and would have fewer feeding issues now. I know that I'm not giving you a clear answer in regard to what choice I think is best, and that is because I really feel that it is a toss-up. One the one hand, you will probably have a healthier child if he has the tube, but do not underestimate how difficult it can be to wean a child from the tube, and how much patience and care it will require. You should also know that S had to have the G-tube surgery redone when he was 9 months old, because the tube grew too high on his stomach and was beginning to touch his rib cage. The Nissen also was failing, and this needed immediate attention, too, and so he had to go through the entire surgery again, which was heartbreaking for us. Bottom line is that the tube has helped S have a very healthy start, and I think that his cognitive and physical abilities have had the chance to really grow without having to deal with constant illnesses, but, we still have the tube, and he has some difficulties with eating that we are working on, and probably will be working on for some time.

Answer 4 of 4

Great that your boy is able to swallow and suck!! My feeling is that even if you do get the g-tube and nissen, just keep up with the oral feeds, and you may be able to transition very quickly from the tube to regular eating. We were given the tube and kind of left to our own devices, and since we aired on the side of caution, we started everything very slowly and carefully with S. In retrospect, I would have been MUCH more aggressive with transitioning him from the tube, and worked much more closely with a feeding specialist to make sure that we were doing everything we could. We came home with physical and occupational therapy, but our occupational therapist did not specialize in feeding issues, and I think this is very important for babies with feeding difficulties. Please do not assume that a G-tube is terrible - it isn't! And, with aggressive intervention and persistence on your part, I think that you could switch him to regular feeds in under a year (if you had the G-tube and Nissen). It is just so good that he is already getting something by mouth!!



Q15: How do you manage siblings when you have a 'sick' child?

Answer 1 of 1

While S is still in hospital one fantastic piece of advice I got was to have a very set routine for when his older sibling sees you. And added to that the other piece of advice that worked along with it was to ask friends who were parents or at least very happy with babies to come and hold or stroke or read to or sing to T when I was with his big sister so that I wasn't eaten up with anxiety about my baby alone in hospital.



Q16: What types of therapy programs you did with your child from infancy? The state programs with PT, OT and speech were mentioned. What did you do, how often and at what ages? What did you feel was very important?

Answer 1 of 2

Here in Florida we have the Early Intervention Program. Honestly, I think it is a joke, but we did get some therapy for J through them. It is a birth-3 years old program which offers OT, PT, and speech therapy. The last year and a half J was in he had only 1 therapist who consulted with the other therapists. We opted to have an OT because we really like her. Now, J is in school and is getting more therapy than he ever has. Our school district is awesome, though. I hope that NJ is better than FL as far as early intervention services.

Answer 2 of 2

Here in UK it varies from county to county, but generally children are monitored by a paediatric assessment team consisting of consultant, speech and language therapist, physiotherapist, occupational therapists, social worker, community doctor, GP and then later an education psychologist (for schooling). We have tried to get our son involved in as many programmes outside NHS as possible: riding for disabled has been excellent, and we've even paid a paediatric physiotherapist for hydrotherapy which was very successful! Otherwise, there are some services provided by NHS, and physiotherapists should be seeing your child at least weekly when they are discharged from hospital.



Q17: From the limited info that we have been able to gather so far it would seem as if you need to live in a bubble for our son's first year of life? Colds, infections, etc seem as though they could be devastating for him. I look at him struggling with his 'normal' secretions now and wonder what would happen if he got a nasty upper respiratory virus? How did you all handle life, I guess, is the question here. Family gatherings in the winter time, being around other children, going places? What types of precautions did you / do you take?

Answer 1 of 3

We handle things with LOTS of hand washing and hand sanitizer in every room!

Answer 2 of 3

My son M was born on new years day, and because of this he missed out on all the winter bugs until he was 9 months old when he was hit with a viral infection, this hit him quite hard but we were quite lucky in that he old enough to fight it, if he would have caught it as a very young baby I'm not so sure he would have faired so well. As soon as we noticed he was getting ill we put him on a course of antibiotics, which in them selves do not combat the viral infection but they do help prevent more serious things like bronchiolitis and pneumonia developing, its also a good idea to try and keep their weight up so they are better able to fight these things (easier said then done as most babies with this condition will be on the lower end of the weight chart).

Since M's illness he was put on a course of preventative drugs (I think there called RSV but don't quote me on that), these are given each month and there supposed to be brilliant at preventing things like bronchiolitis, the Dr's were a bit reluctant to put him on these as they are very expensive (£2,000 - £3,000 for a five month course). As I said M was quite lucky with the timing of his birth, R on the other hand was born in October and had a tough first few months, no sooner would he recover from one bug then he would catch another.

You are quite right in your belief that the first year and especially the first months are the hardest, but no matter how hard you try babies will catch things, the best thing you can do is be prepared.

Answer 3 of 3

VERY IMPORTANT - your son needs an RSV immunisation. RSV or as it is know more commonly bronchiolitis is a very tough virus on any baby, but particularly ours. Older children only get a cold, and adults might get a runny nose when they get RSV, but young babies get a really really tough virus. Babies who have weak lungs from being premature get this RSV immunisation automatically, but children with rarer issues sometimes don't get thought about. We luckily have a friend who is a neonatal consultant at the Hammersmith who jumped up and down about the fact that T HAD to have RSV and then our local hospital accepted it easily once it had been pointed out to them. This is T's 3rd winter and he is getting it again - just in case.

I could bore you to death with this point I keep repeating at any given opportunity - you need to see an 'expert' - someone who's familiar with MTM - it is most reassuring thing that has ever happened us since T was born.



Q18: I understand that some of the boys have had tubes / grommets in their ears, and it has made me think that R could possibly need something similar, he is still not talking and I think this may have something to do with his hearing. What I need is more info as to how and when any hearing problems were picked up on in your boys?

Answer 1 of 3

Z had “glue ear” until he was about 18 months and the move from Bristol (which is an area which is much more “damp”) to Bucks seemed to clear it up Z ’s hearing problems almost overnight! This is probably rather a drastic measure to take if R does have glue ear but I have heard a lot of success stories with cranial osteopathy (manipulation of the skull by a specialist osteopath). Certainly a lot of my friends in Bristol whose “normal” children also suffered from glue ear seemed to opt for cranial osteopathy and found it very effective. It’s probably worth considering before taking the more invasive procedure of having “gromits” fitted (which was an option for Zak before we moved away from Bristol). R will need to be assessed by your ENT consultant and it is worth asking for a referral if you are worried as it won’t be something that they immediately think of doing as standard procedure. Glue Ear is when the middle ear is filled with a glue-like fluid instead of air. Exact cause is unknown but it is more common in younger children. They think that it can be caused if Eustachian tube is not working properly or is too narrow, which causes an imbalance of air and fluid in the middle ear.

Answer 2 of 3

J has never had a hearing test because we've never had any reason to suspect he has any problem hearing. However, when he was between 9 months old and just over a year, he was treated for back-to-back ear infections. If I remember, he was on 4 courses of antibiotics in 3 months, and steroids. That is why he had tubes placed in his ears.

Answer 3 of 3

When J was little we noticed that he didn't respond normally to sound, but discounted it to the fact he couldn't really move. We finally had a thorough hearing test done on him when he was almost 2. We found out that he had some hearing loss in both ears. (He wasn't making any noise or attempting to talk either!) We had tubes put in his ears and it was amazing to see his reaction to sound, and try to talk. He definitely makes much more noise now!



Q19: My son seems to have irritated skin – is this linked with MTM?

Answer 1 of 2

K is a lite scratcher. His skin is dry and flakey. He has scratched at times and made himself really red. Kyle takes 5cc of Claritin every morning. I also use Keri lotion for sensitive skin on his whole body after every bath. The scratching has stopped. Now he likes to pat his stomach and hunt for his belly button.

Answer 2 of 2

I'm a carer for a 9yr old MTM boy in England, we had this scratching problem too for quite a few yrs. It came to a head when he developed Jaundice - turned out to be liver problems - he takes medicine for it and it helps a lot!



Q20: I was wondering if anyone was aware of any contraindications for X Linked MTM to any vaccines? Has anyone's child had a serious reaction to any specific vaccine's, my child has just turned two months and the docs are asking us about vaccines.

Answer 1 of 3

K has gotten all his vaccines. He never had a reaction to any.

Answer 2 of 3

The doctors told us that we could still give our boys their vaccines.

Answer 3 of 3

Z has never had any bad reactions to vaccines. Sometimes, like most children with vaccines, he's a little tired on the night that he gets them or has a slightly higher temperature, but other than that, no problem. For us, it was a "no brainer".... ie, having no vaccines was not an option!



Q21: I would like to know if anyone has experience of heart shadows and X Linked MTM. If so what was the outcome?

Answer 1 of 3

J was dx'd with cardiomegaly back in Jan 03 after a routine x-ray. A cardiology consult was called. He did an echocardiogram and an EKG, both of which revealed nothing. He also said that although it did appear "large" on the x-ray, that it was within normal range for his age and size.

Answer 2 of 3

K also has yearly ECG checkups. Just to monitor his heart. MTM babies are known to have large hearts. The heart is a muscle and has to work twice as hard as a normal heart which can make it enlarged. Kyle's is normal size for his body size. Kyle also has a caved chest on the right side, which is why I say he has problems with his right lung collapsing.

Answer 3 of 3

I was told by Z's doctors that the only organ unaffected by MTM is the heart! They still do routine ECG's as standard procedure, and sometimes the "readings" come back "abnormal" but I've been told that this is due to the unusual shape of his chest (he has a concave chest wall or slight "bowl" in his chest) which affects the reading. I would also be interested to hear if anyone has had any issues with the heart. Obviously, if Z has been on Salbutamol, (he did for a trial study to see if it strengthened his muscles as it works well with some boys with Spinal Muscular Atrophy), they observe the heart rate more closely because this drug causes the heart to race or "flutter".