

Hope for Pompe

We have 3 lovely kids, each one of them are so special and different. The eldest one is Yen Ling, she is 10+ years old; she is a smart little girl with lots of wits. The middle one is Sze Hong, he is 9 years old; he is the most energetic one that keep on complaining about his boredom. The youngest one is Wei Ling, she is 7 years old; she used to be the most accommodating one once when she was a baby...It was due to their character plus our ignorant that have stopped us from seeing our girls were actually having very serious muscle development problem. We thought a boy is typically more energetic than a girl. We also thought that a boy will be more masculine. Besides, all 3 of them had normal delivery with average birth weight and termed with no complication. All these facts had prevented us to think seriously of any potential underlying problem. Due to our ignorant, we had only admitted Wei Ling to hospital for thorough check when she was 1 year old...



From left: Sze Hong, Yen Ling & Wei Ling. Our favourite photo taken when they were younger around 2005.

Yen Ling had started showing lower limb weakness problem when she was about 2 year old. She will fall easily and she would refuse to walk when we do our shopping when she was younger. She would always ask Yee Seng to carry her. Unlike Sze Hong, he will be running about in the supermarket. But due to the 2 had a close age gap of about 18 months, we thought she was jealous and wanted to get more attention. We never took the case seriously. We only started seeking doctor advice when she was about 3 years old after she went to kindergarten as she was the smallest one in the school. Besides, that's really the time when we observed her falling down very often with no good reason. Her knee would just get soft and then she would surely fall down. We consulted our family doctor, he thought she was only under nutrition as she has poor appetite. She was given multivitamins as her treatment. However, her condition did not get better after taking multivitamin for few months. So, she was then referred to a specialist. The specialist told us besides taking multivitamin, she will need to go for a regular occupational therapy to strengthen up her muscle and improve on the motor skill. Still few months of therapy, there was no major improvement. We started asking ourselves what was really wrong with Yen Ling, our instinct was telling us something was wrong and she needed help but we didn't know where else to go to...

While Wei Ling was delivered when Kian Foon was having flu, she came out with blocked nose. However, her stuffy nose got worse after 2 weeks and it seemed that she was having infection, later the doctor prescribed her antibiotic and she recovered after taking the medicine. In her early months, she would get sick from time to time but she would recover within a week. Even though she was weaker than Yen Ling and Sze Hong, she had the best appetite; she would be eating whole day. Though her muscle is softer than her sister, she could still sit up when she was about 8 month old. However, she never acquired the skill of crawling and standing up. All that she could do was rolling around. We thought her development was delayed due to her frequent sickness. A few months later, when Wei Ling was celebrating her 1 year old birthday that we suddenly realized something was really wrong with her. Any normal 1 year old baby can not have such a delay in



Wei Ling, before starting the treatment, lethargic & gloomy. My heart was pumping very hard until swollen, do you know that?

crawling and standing up. She could not sit up right. She was tired most of the time. More importantly was she had very poor weight gain even though she was eating almost non stop each day. Besides, she would get choked easily. There were few times that we almost lost her due to her swallowing weakness. At that time too, she was sick due to respiratory infection for almost 2 months. Her condition did not get better after taking oral antibiotics. She was finally admitted to Seremban General Hospital for a thorough check. It was then we knew that she had a huge heart & liver. We were scolded by the doctor for delaying her admission as she could not understand why we fail to capture the various symptoms. But when we told her that Yen Ling was quite like her sister too, the doctor was very concerned and asked Yen Ling & Sze Hong to be screened as well. After the screening, the doctor told me that Yen Ling had a similar pattern though her symptom is less severe and Sze Hong was totally normal. It was then we knew both of our girls are having a type of inherited disease even though we didn't know of the name yet. It was also then puzzle on Yen Ling health condition that had in our mind finally solved.



Breathing is such an difficult task for me, when can I go to beach and play with my brother & sister?

While Wei Ling was in the hospital, she would cry every time any hospital staffs that wear a white uniform or gown near her. To her, they looked like aliens...Due to her body condition, she was staying at home most of the time. She would take a longer time to get used to her environment. Her saturated oxygen level in the body was poor at about 88% when she was admitted. She was asked to wear the oxygen mask or the nose nasal tube. She refused and finally the doctor had to put her into the square box cut out just right for her head. It's kind of funny and mother on the next bed was very cheeky and called her "aquarium baby". Guess rarely any baby would want to be put into the box. She was discharged after 1 week as soon as her condition stabilized. Due to the critical heart condition of Wei Ling with EF of around 30%, she was referred to National Heart Hospital (IJM) 1 week after discharge. Since Yen Ling was showing similar enlarged heart, she got to meet the specialist at the same time. Since then, they were like twins, they went to investigation, any medical check and treatment side by side. At IJM, we were told that nothing much could be done on the girls as the root cause of the problem was not at the heart but rather genetic problem. The only thing they could do was to perform heart transplant but the question would be how many hearts could they change? The pediatric cardiologist then immediately referred them to geneticists in Hospital Kuala Lumpur on the following week. We could still remember that we had admitted them on 29 Jun 05 for the investigation. They spent 3 weeks in the hospital for various blood and urine screening. During this time, Wei Ling was very sick after 1st week of admission. At that time too, the geneticists told us that they were most likely having Pompe. That was the first time we had ever heard of this disease. So, we started searching through the web for info. We realized that if they were Pompe, they would belong to Infant type of Pompe. When we looked at the typical life span, it said only 90% lived till 18 month old. Our Wei Ling was about 13 months old then and was very sick. We thought we were going to lose her as her condition was really bad in the hospital. Each day seemed so long as great fear was haunting us. We just could not stop thinking if she could make it...It was like we were in the hell...The mental torture was just beyond any words....

There was a short while that we ourselves could not take the facts. We did not know what to do. We were keeping all the info by ourselves, we were so scared that we would just

collapse when we open up to others and when others started talking about it. It was the darkest time in our life...But luck was with us. During the 3 weeks investigation, all the required blood samples, urine samples, liver biopsy and muscle biopsy were taken one shot. Both biopsy result indicated that our girls had elevated glycogen both in liver and muscle based on the test done on Yen Ling. Initially, only liver biopsy shall be taken. However, Yen Ling collapsed when the doctors tried to do a local sedation. Luckily the doctors had the reverse drug ready, she was able to be resuscitated immediately. Later, an well monitored OT was arranged with all team members carefully informed of her earlier collapsed. The OT went on well as planned and muscle biopsy was done too to increase the investigation reliability, it also speeded up the result finding. It might be a blessing that Wei Ling was sick, else she might just pass away in the OT as she was just too weak. After getting the muscle biopsy result in 2 days, the doctor had asked us to send both sisters back to KL Hospital for a dried spot blood test to confirm for Pompe. In 2 months times, we were told that both our girls were confirmed to have Pompe. It was the time when we knew of Myozyme, the miracle drug that would save our girls' life. When the geneticist told us the estimated cost for a year, we almost fainted. Our real challenge had just begun...It was also then we knew we could no longer keep this with ourselves anymore. We started telling our friends and relatives about what happened to our girls hoping to educate and also to get the blessing.

Guess all the kind blessing work. Luck again was on our side. The geneticist was extremely kind and fought so hard for 2 spaces in Expanded Program by Genzyme. The timing was critical too as it was almost time Genzyme tried to pass the drug with US FDA. They were offered the free medication just about 4 months before US FDA approved Myozyme. Even though we got the free drug confirmation, still that was almost when Wei Ling was 18 month old. We were counting days as Wei Ling was getting weaker, we were so scared that we might lose her before the arrival of the drug. The thought of losing her was killing us each day as if we are sitting on the electric chair. We were so desperate yet nothing much could be done except waiting for the 'day'. In April 2006, finally after waiting and fighting (to get the legal documents to import the unregistered drug) for months; finally the infusion started. We could still remember how hard were our heart beating for the excitement and fear (worried for any negative reaction) the day when we was told for their first ever infusion. She was almost 2 year old when she started the infusion.

It did not take long for us to know the drug was working well. The first little thing that we noticed was Wei Ling would smile more easily. Before that, she hardly smiled; she was gloomy most of the time. Kian Foon always thought that it was her fault as she was not happy during the whole pregnancy. After 2 months of treatment, we noticed Wei Ling showed to have extra energy every time after infusion. She started to acquire a new skill to move around that was bottom chauffeuring. Slowly her legs got stronger and she started to stand up. Initially she needed to hold on to something and her legs would wobble. She gained more and more energy, her legs muscle also getting stronger. Soon, she was able to hold on to chair or sofa and started walking. After 8 months of infusion, Wei Ling finally could walk on her own. We could still remember the happiness on her face when she did that. She was laughing all the way when she made turns after turns. It was the most beautiful moment of her life that she was given a second chance to live. Too bad, we just too happy for her and forgot to snap down the moment. Besides, Wei Ling weight gain problem solved after the infusion, she started putting on weight slowly and steadily. She gained 2.5 kg within a year, that would never happen if she did not go for the infusion.



See I could stand holding on to the sofa!

After almost 2 years of infusion, doctor told us that her liver and heart had strunk to almost normal size. In fact, one of the cardiologist was very concern on her progress. She got worried if her heart would shrink down too much that another a different problem might appear. We explained to her what we knew about Myozyme and she was relief after our input. And the audiology testing showed her hearing improved and she only suffered minor hearing loss below 20dB which was negligible.

Life goes on, she continued eating almost non-stop and steadily put on weight, however in 2008 we noticed her teeth started to change color. A tooth decay sign. But she refused to let us get near her teeth. No way for us to stop the decay, it had turned from bad to worse. Finally she was referred to the dentist. Due to her frequent fever as the result of her gum swelling, an operation to remove her decay teeth was arranged end of Oct 2008. All went well, Wei Ling had changed into a 'no tooth' little girl, 7 of the slightly better teeth were left behind.



Wei Ling teeth were spoilt badly and had to be removed. 13 teeth were removed one shot in a well monitored operation. Pity her. This photo was taken in the waiting area of an OT before the operation.

In 2009, when she was 4 year 8 month, we sent her to a nearby kindergarten. We did not want to stress her too much, so had decided to put her in 4 year old class. She was happy joining the children 1 year younger than her. In fact, her height was more at par with 4 year old kids then. Her social life began, to our surprise she adapted well into her kindergarten life. She had always showed strong spirit to participate regardless of her weakness in the lower limb muscles. In the kindergarten, she would still fall down quick easily but she would find her ways to stand up on her own without any help. Even if the teachers offered help, she would insist of getting up on her own. All the kindergarten's teachers adored her spirit and gave their full encouragement.

2 years passed by quickly. Soon Wei Ling had reached the age of compulsory standard 1 government schooling this 2011. Even though we would like to delay enrolling her to a year later but Malaysia education system does not allow us to do so. So, we started sending her to normal standard 1. All seemed to go well initially. Nevertheless, there was some problem after a month. She started to have lots of phleme in her throat. It was very stubborn and won't go off, it changed from watery to thick and sticky. Not a very good sign. True enough, the doctor ordered for phleme culture and found the existence of "Klebsiella" bacteria. We were told that it was a very dangerous kind of bacteria and intra vein antibiotics need to be taken immediately. She was ordered to stay behind after her ERT on 19 Feb 11. A full 10 days course of antibiotics were ordered. At the end of the treatment, all her phleme was gone and we were happily home.

But the happiness did not last long, after less than 3 weeks; she had another serious viral attack. Her spiking fever would not subside and was once again admitted to hospital. This time, the doctors were very upset on her frequent admission and started probing into more details. Later it was found out that the school that we sent her was not suitable for her. Due to termite's problem, the classroom was made up of container (a kind of temporary building made up of metal pieces). Air quality was very poor in the classroom. She was ordered by the doctor to switch to another school immediately after she got discharged from the hospital. So, we had sent her to another school. Till now, she showed to be fine in her new school.

Now at Jul 11, she is at 112cm tall and weighted 15kg. Her non-stop appetite on food had finally stopped with the removal of her teeth. We found that it was getting tough to make her finish her food nowadays. So, her appetite had a direct impact to her weight gain. Though weight gain is not satisfactory but she is growing up at her own rate. Doctor suggested G-tube but we do not buy in the idea and hope she will start to catch up her weight after her permanent teeth started to grow. Too bad, there is still no sight of her new tooth. Though challenges are still ahead but we believe with her strong fighting spirit, she would be ok eventually.



Wei Ling is a God child, her survival proved nothing is impossible in this world. Determination, perseverance, compassion, gratitude are the key things in determining success in our daily life.

We would always tell Yen Ling how lucky she was to have Wei Ling whenever she bullied her younger sister. Else, we would not know what would happen to her if she grew up without being diagnosed with Pompe. We saw too many stories of wrong and late diagnosis. Normally it was till the patient was wheelchair bound or ventilator dependent then the right diagnosis came about. Having 3 closed age children, we just could not stop them from fighting for attention and love constantly.

Now, let us put the focus back to Yen Ling. After the infusion, her lower limbs muscles became stronger. She could walk up the stairs now without support. Before the treatment, she could not walk up the stairs nor could she jump or run like her brother. She would need to pull herself up, so she was like a weirdo in her kindergarten. She was in her 5.5 years old when she started the treatment. After the treatment, she started to participate more in the outdoor activities whenever we brought them out to the park. Earlier, she would only be an observer; standing at 1 corner and admiring how the others running and jumping about. Now, she would compete with her brother in all the activities, though she would still lose out but at least she is more willingly to get involved. Now, she could handle all the 3 actions without any help. We still remembered her 4 years old kindergarten's sportday. During the 30m run competition, she didn't know how to run. Later, her teacher and Yee Seng held her side by side and carried her all the way to the finish line. Now, she is going to a normal school for her primary education. Most of her friends do not even know she has Pompe, to them she is as normal as any other student. They don't quite understand why she was missing in action every 2 weeks though. Only 1 of her classmate knows that she is going for infusion as she has always getting help from the boy to jot down her school homework.

But the only set back was her haunch back problem did not improve. This had indirectly made her feel inferior as compared to her peers. With the help of Myozyme, her height's growth is pretty good given her poor appetite as compared to her brother. Again, she too had got weight gain problem though not as serious as Wei Ling. Now at Jul 11, she is 137cm tall and weighted 25kg. At this height, she should weight at least 30kg.

For Yen Ling, she had grown up into a pretty little girl. If I didn't tell others about her disorder, no one will suspect her having any problem. People will think that her haunch back problem is due to lack of exercise or poor posture. Though I think the latter does play a role as she will never sit up right while doing her homework. Nevertheless, this young lady had developed the interest to dance. We saw her dancing away with her 2 other friends in the recent camp fire in her old school. She was so engrossed and happy during her dance. Even though her movement was not as accurate as her peers, it did not deter her to enjoy the dance.

We felt bad the recent change of school had affected her. Though she & Sze Hong had never fall sick in the container school. But we have to transfer them as well, else either side will be late to school. At the same time, we hope the elders can help take care of their little sister as well. She missed her old friends very much and had voiced her request for us to send her back to her old school. Luckily Facebook has come to rescue. Besides, we would bring them back to one of their old friend's house for any gathering event. We told her too that she was just learning to depart with her beloved friends 2 years earlier as after primary schooling, they will be parted anyway when she gets into her secondary school.



Myozyme has freed Yen Ling from tracheostomy, wheelchairbound as normally happen to untreated pompe patiets, now she can pursue her education like any other normal child.

Myozyme is such a wonderful gift for our girls. Without it, we could not imagine how would they be now or if they are still around. We pray everyday that our Malaysia government will continue to fund their medical fee as long as they need it. We hope more Pomes in Malaysia and around the world could be saved by Myozyme.

Seeing great improvement of health in both our daughter had made us join hand with other LSD families last April 2011 to setup a new society. The name of the society is Malaysia Lysosomal Diseases Association. Our main objective is to advocate for a sustainable medical system especially in the area of ERT. In Malaysia, there are still some eligible patients waiting to be treated. The bottleneck is mainly the government budget is never enough for the consistently increased patients. The number of patients continues to outgrow the funding allocated. We hope by having more awareness programs, the public and the policy makers will hear our voice and support us.



We hope you could help us spread the info on our new society. Let's work hand in hand for a better tomorrow.

By Parents of 2 Pompe girls from Malaysia
 Lee Yee Seng & Chia Kian Foon
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