Rare Parents

Stories of parents with children living with rare genetic diseases

In loving memory of our members:

Ching Man Hon,
Tai Chun Kit,
Ng Man Hin,
Chan Man Fong,
Ching Ying Yu,
Ng Man Nok,
Wong Wing Kan,
Cheng Mo Yan,
Chim Pun Lap,
Yeung Hiu Dan, Annie,
Cheung Yiu Shing,
Hamza Shahid,
Wong Tsz Ki,
Law Cheuk Yin

Message from the Chairperson

Dear readers,

I am very delighted that our third book about Mucopolysaccharidoses (MPS) and other rare genetic diseases is finally launched!

Thanks to the kind help and continuous support of many, Hong Kong Mucopolysaccharidoses and Rare Genetic Diseases Mutual Aid Group (HKMPS) has evolved from a little-known organisation to one blessed with love and attention by many in Hong Kong, in just a few years' time.

Our two previous books have raised public awareness on rare genetic diseases. We are grateful for all the love and support that gave our children the courage to open up themselves and tell their stories, such that the community can understand them better.

No words can express the bitter-sweet experience and surprises that we parents feel and face daily in this long battle. Despite our anxiety, we believe in breakthroughs and try to look on the bright side. We cherish every moment with our children, finding comfort in their optimism and inner peace which help us see what family love and caring are all about.

We sincerely hope that you will encourage your friends to read this wonderful book of stories about parents whose children have MPS or other rare genetic diseases. We hope that recognition from the community can speed up the provision of medical treatment to our disadvantaged children, empowering them with confidence to race against time.

Our dear children, never give up!

CHING MA Oi Lee, Ellie

Chairperson of
Hong Kong Mucopolysaccharidoses and Rare Genetic Diseases
Mutual Aid Group

Message from the Vice Chairperson

We are not extraordinary dads and moms

It is a gruesome battle to take care of our sick sons and daughters, especially when their diseases are rare, and any available support to us is minimal. Very often we go into the fight alone. In some cases, a parent could not face the reality of raising two sick children, and had sadly chosen to abandon the whole family.

Despite our hardship, we have absolutely no regrets because each child is a gift to us. The courage that they demonstrate in their battle for life challenges our value systems, expands our horizons, and makes us understand what responsibility and love really mean. We are as important to them as they are to us.

This book tries to tell everyone that we rare parents are not special, with no extraordinary ability; we are just like you. If we can face such adversities, so can you. We sincerely hope that our sharing can bring a refreshing breathe of strength and love to your life.

Stephen MA

Vice Chairperson of
Hong Kong Mucopolysaccharidoses and Rare Genetic Diseases
Mutual Aid Group

Warriors for their children

I write this foreword with admiration and a heavy heart. This book is a collection of experiences shared by parents of children who are sick with rare genetic diseases. These are real stories of real people, reflecting real suffering and real strength.

It is inspiring to read about the hope and perseverance of these parents despite the challenges posed by these diseases.

The emotional turmoil of the parents on realising that their children are affected by the little-known diseases such as Mucopolysaccharidoses (MPS), Pompe disease and other rare genetic diseases and how they turn it into strength to help their children is well reflected in this book. This book will not only enhance understanding among readers about the hardship faced by the patients and their loved ones, but also raise awareness about the diseases.

It is heartening to learn that several MPS patients are now receiving the Enzyme Replacement Therapy (ERT) in Hong Kong. We hope that the other patients will also get treatment soon so they can enjoy a better quality of life.

I commend the Hong Kong Mucopolysaccharidoses and Rare Genetic Diseases Mutual Aid Group for honoring the persevering parents, their dedication, and their endless love for their children through this book. Our lives have been enriched by reading these stories and learning about their immense strength and resilience.

LAM Woon-kwong, GBS, JP

Chairperson
Equal Opportunities Commission

Foreword

Blessings of a curse

Children with rare diseases are mostly not able to grow up and live long.

Not being able to grow up and live long — is this some kind of a sinister spell? To the innocent child and his family members, especially to the parents, is this a kind of torture? I am not quite sure. My daughter suffers from severe mental disability. We are always worried that she may live longer than we do. But we certainly don't want to see her leaving us as well. This is our eternal dilemma.

But I've never had the feeling of being cursed. Our daughter has brought the family plenty of joy. Because of her being special, we became a special family. Because of her being weak, our family became stronger and more united. Because of the sufferings and happiness we went through together, every member of the family has become more mature and courageous. This has made our lives more meaningful.

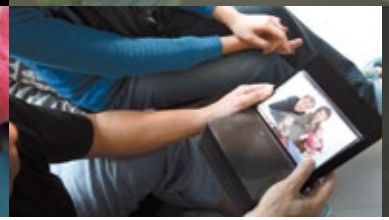
Not being able to grow up and live long — this could be a blessing in disguise.

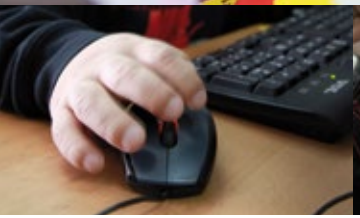
Fernando CHEUNG

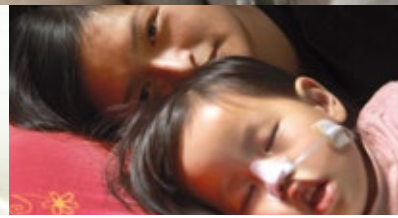
Consultant of
Hong Kong Mucopolysaccharidoses and Rare Genetic Diseases Mutual Aid Group







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互助大使特別大獎
堅毅精神獎
大國用主
黃詠勤女士
2006年1月8日

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From a mother

Little swallow in my heart

I never knew how strong I could be, until my daughter Hiu-yin was born. Her name means “understanding swallow”, my little bird.

She was born with a rare genetic disease called Mucopolysaccharidosis (MPS). I needed to take care of her every single day. At times, it was so hard. The disdainful look from many made it even harder. Once, things had become so unbearable that I thought about ending my life.

Fortunately, the support of parents of other sick children and my Hiu-yin’s optimistic personality helped me make it through, time after time.

Through countless adversities, I gradually came to understand that life is a test. Now I can say with certainty that Hiu-yin is not my burden. Instead, she is the one who strengthens my will and helps me grow.

Light in my lonely life

I was married to Hong Kong from mainland China. At the beginning, I was unfamiliar with the place here, and both my in-laws were ailing, so I had to stay at home to take care of them everyday, having no chance to make friends.

The birth of Hiu-yin brought vitality to my boring life. Her smiles were like rays of sunshine to me, helping me forget about the stress of taking care of the sick. Her every act became the joy of my life.

I remember when she was about one year old, she had to wear a plaster cast on her hip because her pelvic ligaments became loose easily. That made her walk with a limp, which was quite unsightly to look at.

The doctor told me that some kids of her age had similar problems, so I did not need to worry because they would grow out of it. I had no friends to seek advice from back then, so I believed in what the doctor said. It never occurred to me that the reason was not so simple.

Disasters at a young age

Whenever I had a chance, I would take Hiu-yin to play at the park nearby. That gave me a chance to get away from the apartment, and helped her to be in touch with things in the outside world.

However the park, a supposedly safe place for kids, would be full of danger to my Hiu-yin.

When she was three years old, she fell from the swing accidentally, and her neck hit the ground first. When I rushed to check on her, she was conscious and could cry and walk. I thought that she was alright and took her home.

Once we were home, I realised that she could no longer move the right side of her body. I was so shocked and scared, with no one to go to for help. I felt like I had fallen into an abyss. The doctor said that this accident had caused her neck bone to press against her spinal cord, making her unable to move. After a few days’ observation, her condition remained the same, so the doctor suggested surgery.

What worried me most was that the surgery would only give Hiu-yin 50% chance of recovery. If things went well, she could walk again, said the doctor. Otherwise she would be paralysed from the waist down for the

rest of her life. After discussing with my husband, the only person to talk to about this serious matter, we decided to follow the doctor's advice and let Hiu-yin have surgery.

Luckily her surgery was successful. However her nerves were already damaged with no chance of full recovery. Hiu-yin started to receive physical therapy (PT) everyday at the Duchess of Kent Children's Hospital at Sandy Bay on Hong Kong Island. We hoped that her damaged nerves could slowly recover in half a year.

Since then, my daily routine was to wake up early in the morning to take care of my two in-laws, then took Hiu-yin to the hospital for PT in the morning, followed by occupational therapy in the afternoon, and finally back home at night for house chores. Day in and day out, it had been like that, for over a year.

Those days were extremely hard on me, with enormous pressure. Not being a professional nurse, I had to take care of three patients at the same time. I did not want to be cruel to my husband by letting my air out at him, since he was already bearing the full load on his shoulder as the breadwinner for the entire household. So I buried all the sorrow and pain in my own heart.

So stressed, should I end it all?

One year had passed. Hiu-yin's condition worsened, and she would become tired easily after a short walk. The doctor advised for another surgery, this time to break a bone in her leg and insert a steel plate.

I was so taken aback after hearing the plan. How could little Hiu-yin bear such pain? After serious consideration for several days, we decided to take a chance, hoping that the heavens above would pity us and let my

daughter recover fully.

Yet, there was no miracle. Hiu-yin did not get better after surgery, and in fact, her legs had become weaker. Eventually, walking also became difficult for her. I felt so helpless, thinking that I would never let her have surgery again.

Since then, Hiu-yin's walking posture became quite strange. Other people on the street did not understand, so they gave us disdainful looks all the time. Once I took Hiu-yin out for a walk, an old lady followed us from our doorstep and looked at us with scorn. It was painful for me, not being able to explain what sickness my daughter had. So, every time we went out, I would rather carry her on my back so she did not need to walk. I wanted to avoid the hurtful look of other people toward us.

I often questioned why so many unfortunate things had happened to us. My family in the mainland did not understand my situation, and did not give me much support. My husband was busy at work, and could only console me and ask me to hang on. For a moment, I thought about ending it all. But when I looked at my daughter, I knew that it would be very irresponsible of me, so I brushed the idea away.

Later on, Hiu-yin went for special education at the Red Cross Kennedy Centre at Sandy Bay. It was there that I no longer found myself fighting the battle alone. I found supportive friends there, and I finally found out the true cause of Hiu-yin's numerous joint problems.

Support from parents of sick kids

When Hiu-yin first went to school, the school principal told me that there was another student there who looked a lot like Hiu-yin. I ran to meet the

parent of that child right away. Our two girls indeed looked very much alike, both with wide mouth, short neck and joint problems. I learned from the mother that her daughter was ill with Mucopolysaccharidosis (MPS). This was the first time I had ever heard of this disease!

I thought that since her symptoms were similar to my daughter's, may be Hiu-yin's sufferings were all because of this disease. So I went to ask our doctor to refer her to the genetics specialist. After examinations, we finally found out that Hiu-yin had MPS type IV, which was the culprit of her skeletal deformities.

At last, finding out what made Hiu-yin sick and meeting this family similar to ours were like finally finding a lifeline at the angry sea. I was no longer alone. That parent was very enthusiastic and helpful, she gave me reading materials about this disease and also taught me how to take care of Hiu-yin. Through her, I met more parents who were living with rare diseases in their families, which prompted our eventual formation of a mutual aid group.

Now I will call up different parents every now and then to share casual stories in our lives, and encourage each other when unhappy things come along. I have become a positive person, with greater determination to continue fighting against the disease with my Hiu-yin.

What made me most grateful was my sensible daughter. Whenever someone discriminated against us, she was the one to comfort me, saying, "Mom, don't mind them. They did that because they did not know I am sick. Please don't feel bad." I was very touched.

I am thankful for having such a loving and caring daughter, and I wish for more people to know about this rare disease and give greater care and support to this group of little fighters in their fight against their sickness.

Now whenever we go out, I always carry a few leaflets about MPS. When people look at us curiously, I will take the initiative to tell that person about my daughter's situation, hoping that I can help the weak in my own power.

Sixteen years ago, I thought my whole life was in darkness, with no hope. Now I believe that I live a meaningful life.

Mother **Chau Ho Fung-ming**

Daughter **Chau Hiu-yin**, 16 years old, patient with MPS IV


Father Reuben Cheng

From a father

Our angel

Arrival

January 3, 2006 was an incredibly joyful day. Mommy gave birth to a pair of beautiful twins.

Yan Yan, you were the big sister, born three minutes earlier than your brother Hei Hei. You two were precious little ones, bringing tons of joy and happiness to us. Holding you both in our arms was the greatest satisfaction and happiness in life.

April 14, 2008. Yan Yan, you left us. Mom and dad were heart-broken, and our tears ran dry. We will always love you, we miss you every single moment, and we are grateful to you for the rest of our lives.

You were a wonderful daughter who loved to smile. You were our blessed and gentle angel.

Yan Yan, when you were five months old, we noticed you did not respond to sound at all. When we took you to have hearing tests at the hospital, the results were not good. You were referred to the Hearing Centre. It was found that you had total hearing loss in your left ear, and your right ear could only barely hear. So we bought you hearing aids, in hope of getting you cochlear implant when you grew up.

We rushed you to take classes at the Hong Kong Association of the Deaf. True, you were only five months old at the time. I guess mommy and daddy were probably too much in a panic.

Yan Yan, when you were eight months old, after close observation, mommy noticed that you were getting better. That lighted up hope for us and we searched for every opportunity to treat your ear problems. Did you still remember? Daddy and mommy asked all gods and goddesses to help, and even invited master acupuncturists to give you *Qigong* therapy by "sending" their strong *Qi* all the way from Shanghai.

Your eyes started having problems when you were ten months old. Suspecting that you may have crossed eyes, we took you to have eye check-up as well when you had re-consultation for your ears at the hospital. The doctors found that your eyeballs were not moving at all, like doll's eyes, and suggested to have your bone marrow checked. The diagnosis came back — you had a rare genetic disease called "Gaucher's Disease".

What terrible news to strike us. How could this have happened to us? We had just had two lovely twins and our family was supposed to be a happy bunch of four!

The doctor said that there were three types of Gaucher's Disease. Type I was treatable by medication, and patients who had it could live to adulthood; Type II patients could not live beyond two years old, while Type III would not show symptoms till the patient was around nine years old.

Yan Yan, mommy and daddy were very scared and in agony, because you unfortunately had Gaucher's Type II, the most severe type. We were also worried about your twin brother Hei Hei. Could he be having Type III of this disease, even though he appeared to be a healthy baby at the time?

We did not dare to wait. We took your brother for check-up immediately. During the two weeks' wait for his report back, mommy and daddy lived in fear every single day, and it was the most depressing time of our lives. Luckily the report came back saying that he carried the recessive gene only, and we felt somewhat relieved.

Hand in hand

Yan Yan, the doctor said you lacked a natural enzyme in your body, and this affected the functions below your head, the growth of your arms and legs, gave you excessive mucus which choked you easily and made it difficult for you to swallow. Eventually, you could only take your daily nutrients with a gastric tube. Problems at your liver and spleen also made you suffer. Every time we saw your lovely face twist in pain, our heart also ached with you.

In order to treat you, mommy and daddy let you have the enzyme replacement therapy (ERT) for three injections a month, despite the high cost of this treatment which easily ran into twenty thousand dollars each time. In order to let you get well, we would do anything for you.

You surprised us all. No one could have imagined how courageous you, a little baby, could be. Despite the suffering and pain you went through, you simply gave a tiny "hum" sound, bore it all and did not cry. Our relatives, friends, doctors and nurses who had met you were amazed and impressed by you.

There was only this one time, when an inexperienced intern doctor

spent two hours before he could successfully give you an injection for ERT. You were crying in pain that whole time. However, when you finished your drips and saw mommy again, you immediately broke into a smile. Your smiling face simply melt mommy and daddy's hearts.

Around three months into your treatment, we learned about the option of bone marrow transplant, and decided to let you have this surgery to help you get better. It would have been the first case of bone marrow transplant for patient with Gaucher's Disease in the entire world. Instead of seeing you receive endless sessions of ERT that would never cure you, we would rather take a chance with bone marrow transplant.

At first mommy and daddy were full of hope, and had numerous meetings with your doctors about this potential surgery. However, the hospital eventually shut the door on this opportunity due to their perceived high risk of the surgery. The doctor said that there was only minimal chance of a match in bone marrow, and even if there were a match, there might be rejection after surgery. With no green light from the hospital, our persistence was in vain; we were left with no choice but to give up. We thought we had a chance, but ended up in despair. No words could describe the powerlessness and depression that we went though. It is as if we had fallen into an abyss.

February 2008. Yan Yan, your nap time became longer and longer. In fact, you seemed to show signs of unconsciousness. When mommy and daddy took you to the hospital, we were prepared.

Whenever you were asleep, we were worried that you would never

wake up again; yet when you were awake, you had terrible seizures that made you suffer. We were terribly upset. To lessen your pain, we gave you sleeping medication to let you sleep better. When you were in a coma, did you know that mommy and daddy were at the hospital to take care of you, to be with you, day and night?

Departure

Your condition deteriorated, and eventually you were not able to recognise us anymore.

On your last day, your breathing became heavy and you looked ashen, but you had a strong will to live. Your blood oxygen level fluctuated greatly, dropping and rising like a roller-coaster ride.

I still remembered your beautiful face turning rosy pink in your final ten seconds. You were at first drowsy, and suddenly opened your eyes to look at us. You smiled at mommy and daddy. Your face turned pale again, then you slowly closed your eyes. The doctor told us that pneumonia took you away.

Yan Yan, we knew, we knew that you did not want to go. You said goodbye with your smile, and took a good look at us before you left. You wanted to put us at ease, right? Mommy and daddy missed you too. We whispered gently into your ears our love, remembrance and gratitude. Did you hear us?

Mommy and daddy were saddened by your ailment, and we were in tears day and night. Our hearts sank every time we went in and out of the hospital. We often complained to the heavens above why it

gave us the greatest happiness in life, and then instantly crushed everything we had! When we were helpless and hopeless, we turned to the negative. At our most emotional and lowest moment, mommy and daddy even had the despairing thought of putting an end to all sufferings for the whole family.

We had fought numerous times over how to treat you and care for you. You must have known about that, right? But you do not have to worry about us anymore. Mommy and daddy have learnt to stop blaming each other. We are now more considerate and supportive of each other, and have become a strong team. It is all because of you. Did you know that?

Rebirth

It is you who have changed us. Previously we would pray to whichever god or goddess available, but never did we have any inner peace. Then, we gave it a try and went to a church gathering for the first time. It was there that we found inner peace, power and consolation. Mommy and daddy had thought we went to church for you. We did not realise that it was you who had actually led us to find a new life.

Yan Yan, mommy and daddy were baptised before you went into a coma. Daddy felt that you were waiting to make sure we did it. Mommy was touched by your consideration and gentleness, saying that you tolerated so much pain just to help us receive baptism at ease.

In the past, mommy and daddy only concentrated on making a

living, and we were so self-centred. Your sickness let us see a lot of sufferings and learn touching stories in the hospital. All these experiences taught us how to care for and console other people. Yan Yan, you changed us with your life, and we in return influenced those around us.

Looking back, mommy and daddy regretted at not doing enough for you. We were powerless in giving you a better life. We kept you at home most of the time, depriving you of playtime, for fear of giving you an infection if we brought you out. Perhaps it was just our excuse for avoiding to face our own weaknesses.

We overreacted and were very sensitive to the genuine love and support from all relatives and friends because we could not bear the way others think of us or look at us. While we told ourselves that all these were for protecting you from harm and discrimination, in fact this was just so that we can hide in our own shell. We had not yet learned how to truly accept ourselves and you.

We should have done much better. Mommy and daddy are very sorry. If only we could have the chance to make it up to you, anything that would make you happy, we would do so. Sadly, all these can only be empty thoughts now.

Yan Yan, our time with you was way too short. We wanted to give you more care and love. Why could we not be with you longer? Losing you was too much and too painful to bear.

Do you still remember what we told you softly when you were leaving? Our mixed feelings all came down to this simple expression. Mommy

and daddy love you so much, we thank you, and will miss you. We knew that you have heard us and will remember it, because you were smiling sweetly at us.

Yan Yan, our lives have become meaningful because of you. We now have faith and hope. Mommy and daddy know that we have not lost you, because you are always living in our heart and memory. We believe that, some day, when we arrive at the gate of Heaven, we will be greeted warmly and happily by a sweet little angel. We will meet again.

Father **Reuben Cheng**

Daughter **Cheng Mo-yan**, patient with Gaucher's Disease II, passed away in 2008 at the age of 2

Distressed papa, happy papa

Finally! Finally!

When I was only a teenager, I once saw a very smart little boy on the bus. He was about three or four years old, so articulate and with such wit, that I told myself how wonderful it would be to have a son like him.

Finally! Finally!

I have always hoped to have a son as my first child. Maybe it is because I was a firstborn myself, and I was raised in a single-parent family, so I understood deeply the responsibility of caring for my mother when I grew up. The idea of a big brother taking care of the entire family has long been ingrained in my mind. Some relatives of the older generation may not agree with me, saying an elder daughter would know how to take better care of one's siblings.

Finally I had a son! Lik Sang* (also known as Eric) was our first child. He was very articulate, and everyone who had talked to him would enjoy his company. So we just brushed aside his slow development at the limbs before he was two and a half years old. We thought the baby's slow physical development was just a natural part of growth.

The alarm only came later when my wife found that Lik Sang's short neck was close to none, and his back was crooked. After rounds of X-ray checks in over a year, we came to know that he was sick – he was diagnosed with Mucopolysaccharidosis (MPS) type VI.

"Bonus" of my life

This unexpected "bonus" brought me a colourful life from then on.

I came to know what rare diseases and orphan drugs are, was interviewed by the media, went on TV and radios, organised charity concert, published books, lobbied to legislators for help, contacted pharmaceutical companies, made connections with overseas patient support groups, presented talks on MPS to school students, hosted seminars and conferences, petitioned to the Chief Executive at the Government House, and so many things.

Patients with MPS were only generally around one metre in height. They could not straighten their short arms and legs, and their fingers were crooked and fat. So, all daily tasks would require the help of others in the family, even simple things such as buttoning the shirt, wearing socks, carrying the schoolbag, and getting on and off the minibus.

A few years ago, we changed the water tap at home from a twist tap to a lever tap. Lik Sang joked that he could enjoy using water like a king from then on! It showed how much he wanted to take care of himself. How could a careless father like me take good care of my son?! This heavy responsibility all fell on to the three ladies at home – my mom, my dear wife, and my daughter. My responsibility was to help him face this incurable disease, and especially to face the public.

Lik Sang has been a good kid, and was rarely naughty. An anecdote I fondly recall was this one time when he first learned to walk — we were taking the bus, and this excited little boy insisted on babbling and walking around in the bus. I did not know whether I should laugh or to cry.

Since it is more feasible for me than my wife to take leave from work, I am often the one to accompany Lik Sang to his medical appointments. Because of this, we were able to spend much time together. Whether

it was taking the bus, waiting outside the doctor's office, even at afternoon tea time, we would talk about anything – audio-visual products, school life, news, even the topic of death.

We are very glad to have a consensus on funeral arrangement. We both agree to just use newspapers as wrappings for our bodies at our own funerals when the day comes – no need to waste money on fancy coffins, since everything would be cremated anyway. It is better now with paper coffin available; looks nicer that way.

A life of surgeries

Surgery seems a necessity for MPS; it is of course, inevitable to my son. He underwent his first surgery when he was only six months old, to treat his hernia. My most vivid memory of this surgery was seeing him cry his heart out after the surgery, yet we were not allowed to hold him, for fear of impacting his wound which would then require repeating the surgery. However we saw other parents in the room holding and comforting their crying children. That pain still lingered with me today.

My most self-satisfying moment was right after Lik Sang had hernia surgery on the other side of his tummy, when he was three years old. He was returned to the ward and the hospital staff told me that he had never seen any child in pain crying for his daddy instead of his mommy. He even praised me for the deep father-son bonding that we had. At that time, I was so happy that I felt like walking on air. Now I feel so childish when I come to think of it. I am usually the one to accompany Lik Sang for surgery because my wife needs to go to work, so of course it was natural for Lik Sang to call for me!

This incident however demonstrated how much a simple word of praise could bring great warmth to families of chronic illness patients. So, please do not hesitate to show your appreciation, while we shall also learn to accept it with pleasure.

My most torturing moment was on 15 May 2009. It happened to be the International MPS Day, what a coincidence! Lik Sang's doctor had booked him for surgery on that day because his spine was pressing on the nerves at his neck. The surgery was to remove a few pieces of bones there, so as to remove the pressure at the nerves, in the hope of preventing him from becoming paralysed. Lik Sang also expressed that his balance was getting worse lately. So we decided to let him have the surgery.

For four months after the surgery, he needed to wear a protective gear called "Halo", which was a shirt with a metal head frame attached to it, for stabilising and supporting his head. We were told that he would need to stay in the hospital for a bit over a month, similar to what we heard from friends who had this surgery before (with one having only to stay in the hospital for five days).

We all knew these before the surgery, so both Lik Sang and I were prepared.

However, after the surgery, his condition with MPS required him to have intubation at the stomach and a temporary breathing tube. Two weeks later, he needed a permanent stoma (an opening created by surgery) at his throat to help him breathe. This totally unexpected "bonus" brought him unprecedented challenges from then on. For example, he had to learn how to use an electrolarynx at the side of his neck to talk, and how to suck out sputum from his

windpipe through the stoma by himself. In the early days, he even needed suction once every few minutes. Every time I saw how he suffered during suction, my heart twitched too, as if I was the one going through suction.

There were times when he complained that he could not speak, eat or walk because of this surgery. He cried in pain that the suffering was too much for him. In order for him to hang on, I could do nothing but to hold back my tears and tell him all the advantages that the surgery brought along. Thanks to God, he did not give up on his inconsiderate father, and he was a fighter to his disease, in hopes of getting well soon so he could go home and continue with his academic study.

In July, he no longer needed his feeding tube, and his handsome face appeared again. He was even able to eat some porridge. We bought him a cake at his request that day. How he enjoyed the cake! That was probably the happiest moment of his hospital stay, which lifted our sadness from this depressing period. Father and son were able to smile and chat again.

I had always regarded myself a good-tempered and level-headed person, until Lik Sang was vanquished by this surgery. My mood started to fluctuate with his condition – if he was fine when I visited him at the hospital, I would go home with a brisk gait; if he was feeling depressed, my mood also went downhill, and sadly my dear mom and sister would be reprimanded for no reason.

Others would praise me as a good father who was willing to do everything for his son. Honestly I was just putting up a brave face. When Lik Sang was in the hospital, the visiting hours between 6 and 8pm was the most precious time of the day to a boy with nothing

to do at the hospital all day. Despite his many reminders that I should be there to visit him on time, his daddy would often be late because of the temptation of afternoon tea, keeping him waiting and afterwards giving him all kinds of lame excuses.

I have always been his moral support. He would always respect me, thinking that my decisions were always right. I would like to take this chance to say, “Son, please forgive me for all my wrong decisions, which had caused you unnecessary suffering and pain. There will still be challenges and suffering ahead, but trust that your family will face them with you. Let us be there together with you to lessen the pain, turning them into common little things of life.”

* The name “Lik Sang” means “experiencing rebirth” in Chinese, so is the wish of all Christians. In reality, he has experienced a lot in his life. I wish that the enzyme replacement therapy (ERT) that he is having now will also grant him rebirth in his physical self.

Father **Stephen Ma**
Son **Ma Lik Sang, Eric**, 17 years old, patient with MPS VI

Three letters

Here are three letters. They are for those who influenced me a lot in life. They gave me hope when I was down, gave me help, and let me see the true beauty of love. To me, all of them are my angels.

.....

Dear sis,

Do you still remember? Although Kei Kei came a week later than my due date, she was born a healthy baby in late November 2006 at Queen Mary Hospital. But she needed to stay in the hospital for observation because the doctors could not explain her low oxygen level and why she kept shivering. At first I was optimistic, hoping that I could take my daughter home soon.

One week later, my nightmare began.

I was at mom's place when I got a call from the hospital, saying that Kei Kei was sent into the Paediatric ICU (Intensive Care Unit) because of hypertrophy of her heart. It was then that I began to feel scared, sensing that things were not right, and tears started to stream down my face. I remembered the nurse told me Kei Kei's bed number over the phone, but I was too confused and worried to remember it at all.

When I finally calmed down and rushed to the hospital, they told me that Kei Kei still needed more tests, and asked me to wait patiently.

Night came, but the doctor could only tell me, "There are five possibilities that make Kei Kei sick. You need to be prepared. There may be bad news."

A chill went down my spine. Luckily, I had my husband and you, my good sister, beside me.

You searched the Internet right away, hoping to find out for me every possible cause and treatment. When I was helpless, you comforted me, making me feel grounded and supported. Finally, Kei Kei's results came back in mid-December. My husband and I went to the hospital and were met by the Attending Physician, Senior Physician, and Medical Professor. Things did not look good.

Their initial diagnosis was that Kei Kei had Pompe Disease. The doctor said that patients with this disease would have weak muscles, may never be able to walk, and would end with heart failure at around one year old.

We were crying like mad after hearing what the doctors said. The doctors saw that, and further said that there were medication to extend a patient's life then, but the cost would be very high. They asked us to consider and discuss whether to use that medication or not when we had calmed down.

When we had cleared our heads, my husband and I both agreed that we would do everything to save Kei Kei. We would go for anything that could treat her, even if it means exhausting all our savings.

We went home and told you the news. You and everyone at home were supportive of our decision, and were willing to lend a helping hand in pooling money for Kei Kei's treatment. That was a booster for us, supporting Kei Kei to fight for her life against this serious illness.

In order to raise funding for her treatment, you learned to develop a website for Kei Kei. You hoped that kind-hearted people would see Kei Kei's story through the Internet and help our family.

I once asked you why you loved Kei Kei so much. You smiled at me and said, "That's because I love you."

Thank you for not giving up on me at hard times. Thank you for supporting me all the way.

Love,
Your younger sister

.....

Dear families of patients with rare genetic diseases,

I believe that aside from my family, you are the ones who understand my difficulties the most. You face the same hard times as I do. You dedicate yourselves totally to take care of your sick children, while you are hopeless with no treatment in sight.

It was because of you on our side, lobbying to the government together, that Kei Kei is now able to have full funding for her medication.

I remember it was May 2007 when we first met. My husband and I went to the annual gathering of the patient group, the Hong Kong Mucopolysaccharidoses and Rare Genetic Diseases Mutual Aid Group, for the first time. That was the first time to know a group of parents who were experiencing the same ordeal as us. It was also our first time to see so many cute yet ailing little children like Kei Kei.

Although your children looked different from others, with their big heads and short limbs, they always wore a bright smile on their faces, which made me feel warm and welcomed.

Your experience gave me much encouragement, telling me not to give up easily.

At that time, Kei Kei had started to use medication. We still needed to worry about our finances, because we could only hold on to the end of the year despite a three-month drug sponsorship by the pharmaceutical company as well as support from our family.

After knowing you, we learned that we must stand up and fight for our children's medical treatment rights. In July that year, my husband and I went for our first-ever public demonstration with you to urge the government to

provide support and recognise the needs of patients with rare diseases.

Media coverage helped more people know about Kei Kei. We were pleasantly surprised by so many kind-hearted people in Hong Kong. They were willing to donate without asking for anything in return. In just a short time, we were able to collect a sufficient sum of money to pay for Kei Kei's treatment for the following year.

In October, we went together to the Legislative Council Complaints Division to ask the government to include drugs for treating rare genetic diseases into the Hospital Authority's Drug Formulary. That way, treatment costs for patients would be fully covered.

We caught the public's attention, and the government had to respond. In January 2008, the government announced that the Hospital Authority would have an extra recurring funding of 10 million dollars to support patients with rare genetic diseases. We were the first few parents to benefit from this.

Things will not be so smooth without you. If not for you, we would still be scraping around for Kei Kei's medical fee.

Kei Kei has improved much after taking the biweekly medication. She is no longer a "Tarepanda". She is now turning three years old; she can stand up and walk. We are thankful that her heart has not been giving her any problems too.

We know that you are still waiting for the government to lend you a helping hand. We sincerely hope that the day will come soon.

To all other family members of patients with rare genetic diseases that I have not known yet, you have my full support as well.

Yours truly,
Mrs Chung, mother of Kei Kei

.....

My dearest Kei Kei,

The fact that you are sick has indeed brought some sad days to daddy and mommy. However, this also exposed us to more things and people that we had never expected knowing, seeing and meeting. Our view towards life has also been changed.

We used to believe that all things can be planned in advance. For example, before you were born, we planned to bring you to your grandma's house in Cheung Chau for the first month after your birth. We planned to send you to childcare and bring you home from work everyday when my maternity leave was over. However, your illness had totally changed our plans. It also showed us in our face that there are things in life that cannot be planned.

In order to take care of you, we need to make much time. These instead became precious joyful family moments beyond our plans.

When we were home, we played with you everyday, and also took the opportunity to train your muscles. We bathed you ourselves. Seeing you splashing the water in joy, we knew that you were enjoying it very much even though you were too young to tell us yourself. We did not have time for social gatherings anymore, but we did not mind at all.

Do you know that mommy will now take the initiative to chat with children who look special? Most patients with rare genetic diseases do not look attractive, but now that I have had more contacts with them, I will say hi to them and talk to them. This is something mommy would not do before.

I remember I was once waiting in line for the bus. There was a child with Down Syndrome. He was very articulate, and I told him that he was

very smart. I believe that simple words of encouragement would help patients feel loved and cared for by the community.

Kei Kei, you need to go to the hospital for medication once every two weeks. Other people may think that mommy and daddy are having a very hard time. However, we do not feel that way at all. We can still go home with you, play with you and sleep with you. What more can we ask for? We are so happy with you around.

We see kids who are smaller than you in the hospital. Their illnesses are not as rare or as severe as yours, yet they are not able to go home. How terrible and worried their parents must feel.

Because of you, mommy and daddy treasure everything that we have ever more. And because of you, our relationship with our friends and family members are closer than ever.

Please remember to be a happy, healthy person, always.

Love,
Mommy

Mother **Chung Man Shuk-man**
Daughter **Chung Wing-kei**, 3 years old, patient with Pompe Disease

Mother Wong Lai Wing-jun

Walking our own paths together

I am content with my family life, with my two daughters and a son. My husband is a truck driver going between Hong Kong and mainland China. He is rarely at home due to his busy work schedule. Being an unambitious mother, I am happy to take up the role of a father at the same time.

Although we are not well-off, we can lead a happy life — that was what I thought at first. It turned out that true happiness is not ensured; it is hard to find and hold on to.

It all began in 1983…

I found out that my lovely second daughter Wing-kan was a bit different from other kids when she was one year old: her forehead was more protruded than others, the joints at her arms and legs were crooked, and she was often sick.

So I took her for medical check-up at a clinic nearby, but the doctor there did not know what was wrong either. He just hastily did a check-up and jumped into conclusion that her deformity was incurable.

It was about the life of my daughter; there was no way I would give up so easily. I began seeking medical consult everywhere with my daughter. After almost a year of searching, it was at the Queen Mary Hospital that she was diagnosed with a rare disease called Mucopolysaccharidosis (MPS). This piece of information did not make us any more hopeful. It turned out that the very first doctor who checked up on her was not completely wrong. It was many years ago, and medicine was not as advanced as now, so this type of illness really could not be treated. I was told my daughter only had about ten years to live.

Since then, Wing-kan was treated as a patient with MPS. It had been like that for twenty years, until she was thoroughly checked again at Prince of Wales Hospital in Shatin and was found that she actually had Mucolipidosis (MLS)! Same as before, this finding did not give my daughter any hope; it's just another incurable disease.

Although the disease was not curable, we still wanted to treat all the problems that my daughter had. Frequent visits to the hospital became my routine. Sometimes I even slept at the hospital. Every time I heard about a new treatment, I would let her try it, including Chinese medicine and acupuncture. Other people may regard me as messing around with medicine and wasting money, but I just wanted to try every possible way that may help my daughter. I did not want to give up on the slightest chance of hope that may help her.

Wing-kan had begun to require all kinds of surgeries to her bones, eyes and ears since kindergarten. She endured all these sufferings and pain with grace at her young age. It was also hard for me to believe that she even needed to use a breathing machine to help her sleep. As a mother, I could not do anything to reduce her suffering. All I could do was to make myself useful by taking care of her and staying beside her day and night, and to encourage and support her all the time. I would always stay alert when taking care of my daughter, no matter how exhausted I was. Never did I complain or give up on her.

Persevering a splendid life

Wing-kan's disease caused her height, look and built to be very different from others. Ever since she was small, people often looked at her with a peculiar stare. She was also discriminated against when she applied to school and for work. All these gave us very hard times. I had once cried in front of teachers and school principals because they did not let my

daughter go to school. It was Wing-kan who suffered the most. Because of all the discrimination, she had a low self-esteem. Trying to contain the pain in me, I could only comfort her that everyone's life is different, and the paths we walk are different. When we face things that we cannot change, we can only submit to humiliation and move on.

Later on we became accustomed to it. In fact, we could not afford to be bothered by it anymore. Wing-kan also slowly learnt to accept the reality, and to one's surprise, she began to take on a stronger and more positive attitude.

Although my daughter was sick, it was only in her body. Her mind and IQ were no different from others. I would like her to go to school and have a normal childhood, so I took her around to seek admission. We hit the wall everywhere we went. What angered me most was that it was the school principals and teachers who were the most judgmental. In those days, there was no Equal Opportunities Commission, nor was there the Anti-Discrimination Ordinance, so we had nowhere to go for help or to complain. In the end, I could only find admission for Wing-kan at a village school.

Being a very clever and hard-working student, Wing-kan was always top in class. After a smooth transition to secondary school, she met some very good classmates and teachers who were considerate of her condition. Thanks to them, she received much support and accommodation, so she was able to study well and complete the HKCEE public examination.

Shortly after graduation from secondary school, my tireless Wing-kan continued to take courses in design, computer, accounting, office administration, etc. But all these hard work did not help her land a job. She did not give up, however. She turned her attention to learning to make clay flowers. I was both proud and amazed at her talents and strong will!

Because of her persistence and perseverance, and despite of her crooked hands and fingers, she managed to master the art of clay flower making! She also designed her own website and displayed her beautiful artwork for sale on the Internet. I was joyfully encouraging her and cheering for her all the way.

So, we paired up as an ace team – she led the design, production and online sale, while I helped purchase materials, make deliveries, and collect payments. We had great times then. Sometimes we became so excited and busy and yet we did not want to stop. What comforted me most was seeing her persistence and hard work paid off. She truly enjoyed and loved her work, and found her self-esteem again. She was smart in academics, talented in design, and had won several scholarships and design awards. I was so proud of her.

Valuing our short time together

Children with rare diseases were destined to race against time. It is a countdown to the end everyday. I was also quietly counting the days of Wing-kan. I could only wish that she would lead a happy life; no matter what she wanted, wished for and dreamed of, I would try my best to help her fulfill them. She was eager to learn, so I encouraged her to study and further educate herself; she also loved travelling, so I took her to trips whenever we had a chance. During our trips, she would get tired of walking easily, so we used the wheelchair; when the roads were bumpy, I would carry her myself.

I regarded her happiness as mine, and her sadness as mine. My only worry was not doing well or doing enough for her. As a mother, I willingly did all that I could.

In 2006, one year before Wing-kan passed away, her heart muscles continued to enlarge, and her condition worsened. She was so weak that the doctor needed to inject her with a cardiotonic needle to sustain her life. The doctor said that this needle could only be used once, to extend her life for a year. At first my daughter was so scared of the pain that she refused. It was after rounds of encouragement and persuasion that she was willing to take the medication, but she still needed lots of pain-killers to feel better. However, these made her drowsy and unconscious most of the time.

Soon after that, Wing-kan made a request – she wanted euthanasia! I knew in my heart that she must have suffered enough to make this bold request, as it would take more courage and determination to ask for death! I was so heart-broken and confused then. Fortunately the doctors referred us to the Palliative Care Department at Tuen Mun Hospital, which unprecedentedly provided us with counselling that was previously available only for cancer patients. While the doctors and I tried to talk her out of this, we also told her that it was illegal to have euthanasia in Hong Kong. She eventually gave up on the idea of ending her own life.

Wing-kan knew her time would soon be up. She treasured every moment with us in the family. She also requested not to die in the hospital but rather at home, so she could spend her remaining days with us. She said that it was her last wish. The Tuen Mun Hospital held a birthday party for all patients who were born between April and June. Wing-kan was born on June 2, so she was invited to the party too. She had always liked to laugh. She was so happy that day and I could still remember her brilliant smile. That was her last birthday party.

Two days later, in the afternoon of April 22, Wing-kan left peacefully while watching TV at home. Just like that, she did not make a scene for anyone, and silently reached the end of her life. With a persevering will and positive, optimistic attitude, Wing-kan has bravely lived a good 25 years of

wonderful life, instead of the declared ten years of hopeless life.

Although I had had a lot of counselling in advance, and was prepared to accept the reality of losing her any minute, nonetheless I was the one who gave birth to her and had been with her every moment. How could I really accept it that she had left?

I was heart-broken and in deep sorrow. My life lost all purposes, and I did not see any reason to live on. I was left with emptiness and loss. Grief did not go away easily. I also kept blaming myself for not making all her dreams come true. It was only when I received psychological counselling that I slowly began to feel relieved.

Drawing a short straw in life

The idiom “troubles never comes singly” truly reflected the first half of my life. Caring for Wing-kan had languished me in this long haul. However, the heaven above did not pity me. Instead it kept on fooling me, especially in the last few years of Wing-kan’s life. Besides being worried about her condition and running around for her day and night, I also tasted the bitterness and sorrow of my own life.

My husband turned out to have another woman in mainland China. They even had children. In 2002, he wanted a divorce with me. No matter how much I begged and asked, things would not change, and I could only accept it reluctantly. Luckily he still supported this family financially. Yet, two years later, he died of a heart attack. With the head of the family and the breadwinner suddenly gone, and my close-to-zero work experience and skills, add to that the 24-hour care necessary for Wing-kan, how could I possibly have the ability and any energy left to shoulder the entire family? The only choice I had then was to apply for Comprehensive

Social Security Assistance (CSSA). Our whole family also had to cut down on living expenses. Days were not easy then.

August 2005. I had not yet even recovered from the loss of my husband when I was diagnosed with the deadly cancer. I felt as if I have fallen into the lowest point in life. Yet, when I thought of my children, that they would become orphans if they lost me, I told myself that I could not die. So I took the courage to accept surgery and undergo electrotherapy.

Wing-kan was a very good daughter. She was filial and so understanding. Although she herself was suffering much due to her disease, she was the one who often engaged her elder sister and younger brother to comfort and encourage me. I was so glad to have my children around. They gave me comfort when I was in pain, giving me the strength and reason to stand up and fight against my own illness when I was down.

One year had passed after my diagnosis of cancer. But bad news continued to come. In 2006, Wing-kan's condition took a bad turn suddenly. She needed much more caring, making me even more worried and tired. In April 2007, my daughter passed away, and her grandfather also left this world soon after she died. The departure of my love ones one after another was too big a blow to bear. I could only sigh and cry silently. Why did it have to be me?

Embracing the past for the future

Fortunately, all the sufferings and sad happenings did not bring me down. In the past twenty years of accompanying Wing-kan to fight for her life, my will was strengthened like her. This helped me to have the courage to face adversities alone, and to clench one's teeth to solve all problems. I want to thank Wing-kan. She has inspired me, teaching me what it means to be strong willed and courageous.

Wing-kan's departure made me understand that we have no control over life. She helped me understand that as long as we try our best and come to terms with what we have, we can enjoy a good life.

Yes, I still need to take care of her sister and brother, who both love her very much. I am able to continue to participate at the Hong Kong Mucupolysaccharidoses and Rare Genetic Diseases Mutual Aid Group as a veteran, helping other patients and families by openly sharing my sad journey and my experience in caring for Wing-kan.

Two years had passed and Wing-kan's room remained the same. The paintings she drew, the poems she wrote, and the clay flowers and embroidery she made were all kept in their same places, as if their owner had never left us and was still living among us.

Time has eased the pain. Or more so, the pain I felt has precipitated. However, i still miss my daughter so much. Although my body and mind were tired during the days we had together, she left me with more happy times and memories than sad and painful ones. Her bright and strong smile is what I remember of her. She is always on my mind.

I have begun to walk out of my dark phase, and my body is beginning to recover. I want to be stronger and more positive to face the unknown future. My dear daughter has left me with courage and energy, and I am ready to embrace whatever comes ahead of me.

Mother **Wong Lai Wing-jun**
Daughter **Wong Wing-kan**, patient with MLS, passed away in 2007 at the age of 25


Mother Yeung Yeung Siu-foon

Twists and turns

Having sick children is worrisome. It is even harder with the disease Mucopolysaccharidosis. People say that well-being is worth a pot of gold. I would say health is worth more than that; it's priceless.

My two children and I immigrated to Hong Kong from Guangzhou in 2003 to reunite with their father. My son Johnny (also called Tak-chun) was eleven years old then, while my daughter Annie (also called Hiu-dan) was eight. Since then, we had been in and out of the hospital most of the time.

Twist and turn — hospital

Looking back when they were born, no one noticed any problem with them. As they grew up, we found out that their hair and eyebrows were coarse. Also, they could not straighten their arms and legs. When they were five or six years old, they were not as tall as other children. So, we took them to the Children's Hospital for medical check.

We were still living in Guangzhou at that time. We even had to visit three different hospitals in one day.

The doctor at the Children's Hospital said that my children had Mucopolysaccharidosis (MPS), but was unsure of which specific type. After doing a urinary test, they were said to have MPS type IV.

Doctors at the Children's Hospital asked us to visit the Women and Children's Hospital of Guangzhou for medical check. When we arrived, doctors there asked us to go instead to the First Affiliated Hospital of the Sun Yat-sen University for assessment. Finally, at the First Affiliated Hospital, the doctors said they recognised my children's illness, and said they had also researched on their disease. But, they told me that there was nothing available to treat them, and this disease had no cure.

How I wish them to be just normal children! That would be enough for me. However, they turned out to be special. Would I give up on them? Never! We understood that it would be more and more difficult down the road. We also knew that they would not be able to grow as tall as other people, and they would have a short life. However, the doctors never told us how short a life, or how fragile they would be. For example, we thought that Johnny and Annie would only grow up like people with dwarfism, being short like five- or six-year-old but still agile in movements. It was totally out of our expectations that their physical condition should gradually worsen, especially their cardiac and respiratory functions.

Twist and turn — days of darkness

It was only after we had moved to Hong Kong that both of my children were diagnosed with MPS VI.

When we first came to Hong Kong, we lived in one of those old walk-up buildings. I would carry Johnny on my back and let my daughter walk alongside us by herself, and would have them swapped every now and then. The social worker Ms. Woo at the Assessment Centre was kind to us. Seeing that I had to care for the two children by myself during the day, she provided us with a waiver form, so that we did not need to worry about medical bills.

About a month after Johnny's arrival in Hong Kong, he complained of a very bad headache one night, and went to bed early. When he woke up the following day, he asked us, "How come it's so dark?" It was only then that we found out he could never see anything again!

It happened all so suddenly! But Johnny remained calm, and he accepted the reality, which made it less difficult for me and my husband. It was our daughter who was the most frightened and kept on saying, “Oh no, oh no, big brother can’t see anymore!”

Twist and turn — school-hunt

My daughter Annie came to Hong Kong a few months earlier than her brother, and had studied in a regular school for about a year. She was a conscientious and hard-working student. I did not need to worry about her or help her with school work. She completed all the tests herself. She would only ask me for help occasionally if there was a word she did not understand.

I had planned to take Johnny to a regular school as well. However, when I took him to the Ebenezer School for the Blind, they would not accept him, saying that he was physically disabled; when I took him to the Kennedy Centre[1], they rejected him as he was blind. With nowhere to go for help, I took him to seek assistance from social worker at the Education Bureau (EDB).

During that time, Annie was having a hard time with school work because she needed to skip class once every week to have physical therapy at the Queen Elizabeth Hospital. So the social worker suggested to transfer her to a special education school[2], where in-school resources such as physical therapy would be available so she did not need to make a trip to the hospital every week. In light of that, Annie transferred to a special education school. Later on, EDB also recommended Johnny to the same school, because there would be services and counselling for people who were blind.

1 The Kennedy Centre of the Hong Kong Red Cross — a school for the physically handicapped
2 The Margaret Trench School of the Hong Kong Red Cross — a school for children who are physically handicapped and with multiple disabilities

Twist and turn — Annie

Annie had attended special education school for about a month when we received news from the Prince of Wales Hospital (PWH) that there was a match for her to have bone marrow transplant surgery. So, she stopped going to school. For the following two years, I was the one who took her to PWH for follow-up consultation, so our bonding was particularly strong. Daddy, on the other hand, stayed home to take care of Johnny.

Whenever Annie was frightened or was suffering, she would think of me; when she could not stand the pain, she would call for me. After the surgery, she had to be intubated and take steroids. There were times that the medication was so strong that she once had to go pee twenty one times in a single day! It also made her unable to go to bed. In two to three days’ time, her hair had all fallen off. She was suffering, and my heart ached with her.

One day, when she was about to resume school, she cried out loud that her head was so painful as if it were going to explode. The doctor said she needed surgery right away to drain extra fluid from her head. I still remembered that the surgery was done near midnight. I was alone at the hospital, with no phone. It still scared me to think of it now. Luckily, the surgery was a success, and Annie could go to school again soon after that, which made her very happy.

Having gone through two major operations, we took great care of her daily beings. The doctor had also warned us that her bones could be injured easily, so we never let her run, to minimise any chance of her tripping and falling.

But to think of that day again…may be her legs suddenly become weak. She fell at home, hit her head, and in a flash she was gone, forever gone.

She could not be revived even after being rushed to the hospital.

It was early in the morning around 7 o'clock. I was alone in the hospital, panicking, not knowing what to do, thinking it must have been a dream.

Poor Annie! She had been through so much. She had just resumed class for a few months when it happened and we thought her suffering would soon be over. She left us so fast, so unexpectedly. True, we knew that those with this disease would make an early departure, but we never thought it was to happen all so suddenly.

I still feel that Annie is around, as if she has only gone to school.

Twist and turn — Johnny

Now we are only left with Johnny.

Johnny's cardiorespiratory functions had deteriorated slowly these few years. If he had bruises, they would not heal as easily as other people do. His bones were also deformed. The doctor said that bone marrow transplant would not help him.

Johnny is an optimistic boy. Otherwise he will not be able to deal with this disease. At first, whenever we went out with him, many people on the streets would discriminate against us. Now those in our neighborhood are used to us. Sometimes, people are curious and show pity on us, some will come by and say hi, some will ask if Johnny will likes to listen to their music or not. However, there are also some who will complain about us for no reason at all.

We try to fulfill what Johnny needs as much as possible. He needs to use

the breathing machine when he goes to bed. In the morning, he needs help on a lot of things such as wearing a hearing aid, taking medications, and taking eye drops. His lower eye lashes often poke into his eyes, making them red and swollen, and so he looks tearful most of the time. Because of that, he often brings a handkerchief with him. But other children and parents may think that he is crying whenever he uses it.

Recently, Johnny has had less seizures, which makes me less worried and can rest more too. I am not someone born with a silver spoon, but it is not easy at all to remain poised and good-tempered while taking care of children with chronic illness during the day and sleeping restless every night. When I do not have sufficient rest, I will become grumpy. I have come to understand that I must learn to take things as they are, despite adversities and challenges.

Twist and turn — treatments

Now Johnny can finally have enzyme replacement therapy (ERT). He needs to go to the hospital for it once every week. I will go with him and stay inside the ward every time, so that he knows that I am there whenever he needs me, making him feel at ease. This makes me feel better too, because otherwise I will be worried about him at home.

Usually Johnny cannot eat anything before blood test or treatment. He needs to do intravenous (IV) drips for his treatment every week. At the beginning, the nurses and doctors would need to try many times before they could inject him with the needle successfully. It was so painful for him. Generally, every doctor would need to try at least two to three times before giving him the injection successfully.

Once the doctors had suggested to implant a small tube below his

clavicle bones for IV, so that he would not need to suffer the intravenous needle every week. But to us, it was easier said than done.

I still remembered that Annie used to have a small tube like that when she was around. Although having the small tube required just one poke, and the patient would need to bear the pain only once, the tube required constant cleaning thereafter; also, after returning home, she would feel the tube being in her way; when she was at class, we were worried that her classmates would accidentally pull it out. Johnny could not take care of himself, so if he wandered around at school and had his small tube pulled out accidentally, what shall we do? There were too many concerns. I had asked for his opinion too; he would rather go through the needle-poking every week.

Since Johnny cannot see a thing, the physical therapist has provided him with a set of simple arm and leg exercises these few years. But, simple things like raising the dumbbells are difficult for him. May be these few months of ERT have really taken effect. I could not believe it when he was able to raise the dumbbells a hundred times just earlier today! He was panting afterwards, but the smile on his face showed how much he valued this little achievement. I was proud of him too.

Twist and turn — hope

Although the treatment is not painless, I tell Johnny that it is totally worthwhile as long as it is good for him. Now he has had ERT for half a year already, he does not feel tired as easily as before. His cardiorespiratory functions also seem to have improved, but we do not know whether he will be able to continue the treatment after the one-year drug sponsorship has been used up. This worries him, me, and also his daddy.

I hope he can continue to have ERT, wishing that it would help him. The most important thing is to prevent his condition from deteriorating, and for him to live a better life, perhaps a longer life too. At present, he cannot tie his own shoelaces. Even simple daily tasks like dressing himself and bathing himself would require others to help. I simply hope that once his condition has improved, he will be able to take care of himself, and will be able to move around more freely.

All in all, having treatment is much better than none. I wish that he can continue to have treatment, which will help him live.

Mother **Yeung Yeung Siu-foon**
Son **Yeung Tak-chun (Johnny)**, patient with MPS VI, 17 years old
Daughter **Yeung Hiu-dan (Annie)**, also with MPS VI, passed away in 2009 at the age of 13

Short days, long nights

My story began like a Cantonese melodrama old movie.

I am a Chinese who was born and raised in Indonesia. My elder sister once travelled to Hong Kong for vacation and visiting relatives. She came home very excited, telling me that we had a cousin there with very decent personality, who was totally different from the chauvinistic Indonesian boys. She also told me that unlike in Indonesia, Hong Kong did not allow polygamous marriage. So it would be good for me to have a husband that could be my partner for life.

My sister encouraged us to see each other. I thought that it would be good for my future too. So, my cousin and I began writing to each other. Two years later, I packed my suitcase and became a so-called “immigrant bride”.

We were married in 1982. My first son was born in 1985. We led a happy and wonderful life. Not wanting our child to be lonely, we decided to have another child to keep him company. Four years later, my younger son Kam-shing was born. We were so happy to have two sons in a row, and we thought life could not be better.

Unfortunately, good times did not last long. Our nightmare soon began.

Shortly after Kam-shing was born, I took him for a regular medical check-up. The nurse said that his thighs were a bit stiffer than other children’s. So he was referred to have ultrasound scan at the hospital. Fortunately, nothing seemed wrong.

When he was about two months old, he vomited whenever I fed him. What was more, he vomited out even more than what he had just taken. So I took him to the doctor, and we were referred to the hospital. This time, we had to wait for a month before we could see a doctor there. Fortunately, again, the doctor said he was normal. In order to reassure me, the doctor even arranged for Kam-shing to get an EEG brain scan. So, we were on the waiting list again, and things turned out alright.

Soon, Kam-shing had turned five months old when he appeared to have a cold or flu. I took him to the clinic, and the doctor said that he had some shortness of breath, gave him some medicine, and asked us to visit again in two days. The doctor said Kam-shing would recover quickly, and he was fine.

Unfortunately we were fortunate

The many fortunate times have turned out to be Kam-shing’s misfortune. If his disease could be diagnosed earlier, he would have been offered better care and treatment, then his body would not incur so much damages, and he would not have to suffer so much.

I still remembered that night when it all started. It was the night before his follow-up consultation. Kam-shing drooled uncontrollably for no reason. He could not go to sleep the whole night and stayed up till the morning. His father just returned home from working overseas that morning and saw that Kam-shing looked quite different from normal, so we rushed him to the hospital. The doctors checked on him, did an EEG brain scan, and said that he had stomach cramps.

We wondered though, if he had stomach cramps, then how come his eyes were rolled up, his tongue protruded, and he was not able to smile or cry? I was so scared. The doctor later even told me that both of his eyes had gone blind! Seeing how my son was suffering, and thinking that he had become blind, I could not hold back my feelings and started to wail in the hospital.

I stayed with Kam-shing every single moment during his stay in the hospital. I took care of him carefully, and observed how he was progressing. I had a

feeling that his eyes would slowly follow me wherever I went. The weather was quite cold then. I sneezed one day, and Kam-shing smiled!

I was very certain that my son was conscious. I was so happy that no words could describe it. So I told the doctor what I saw with my own eyes. A series of investigations and serious examinations followed, and we were in and out of the hospital many, many times.

The reports finally came out, and so did the unfortunate news. My son had a rare genetic disease with a name that was so hard to remember and pronounce --- he had Glutaric aciduria. He had its type I, and was the first-ever person in Hong Kong to have this disease.

Because of this disease, Kam-shing lacks an enzyme that breakdown two kinds of proteins in his body, which affects his physical self and functions as he remains like a three-year-old child and cannot grow big. He lacks power particularly at his bones, neck, waist and legs. His pelvis was tilted, so he cannot raise his head up, cannot straighten his body, and cannot walk or stand. All he can do is to lean sideways against the back of a chair. Sometimes his limbs will also sway involuntarily. His body is very weak and he has fever, asthma, and shortness of breath easily. He is allergic to some proteins too, so we have to be extra careful when preparing food for him.

I was not afraid of the strange looks of others on me at all, now that we knew our son has this disease. However, I was disappointed that the relatives on my husband's side did not accept the reality that Kam-shing is sick. My parents and family were all in Indonesia, so no one was there to support and comfort me except my husband. I felt really bad, and often felt hopeless and helpless. All I did was to cry day and night. I did not want to carry on, and once I drastically lost ten pounds in a week.

Long-time struggle

The day that we took Kam-shing home from the hospital was the prelude of

my long-time struggle for my son.

Because of his illness and pain, Kam-shing's emotion was not stable. He would throw a tantrum easily, and he was often anxious and hysterical. I took care of all his daily living and feeding, and he relied on me more and more, to a point that he would not sleep alone in bed. I had to carry him in my arms so that he would sleep. I could only give in to him because the slightest nervousness or anger he felt would make him unable to sleep for three nights straight. That would make me feel even worse and more helpless.

The only way he would sleep every night was for me to hold him on the living room sofa. However, Kam-shing was very sensitive to how I hold him. When I sat on the sofa, I could not lean against it to ease my posture. If my body or my arms moved the slightest bit, he would be start from his sleep and cry, and then not able to sleep peacefully again. So I could only be very careful and not move at all while holding him, so that he could sleep well.

Since I needed to be on the alert all the time and to maintain a particular sitting position every night, eventually I had back problems and began to wear out.

Taking care of Kam-shing in daytime was not any easier than sitting and holding him to sleep at night. Tasks that other people would regard as simple, such as feeding him, were in fact difficult struggles of strength and endurance. Because of his illness, he could only "sit" by leaning on a chair, and his mouth could not open normally. He did not know how to suck, eat or swallow food, and he would vomit easily. Moreover, if he felt nervous, his body and limbs would swing around uncontrollably. So, when I fed him every time, it would require lots of energy.

When it was time for him to eat, both of us would sit on the floor. I would let his back and head lean against my chest, and then I would use both my legs to hold his arms and legs, so that he would not hit around and spill his food.

Then, I would use a specific angle that I discovered after the many years of trial and error to place food in his mouth. Yet, that did not necessarily mean a spoonful of food taken. Since he vomited easily, whatever he swallowed could come back out like a fountain all of a sudden. So I would need to clean up and start feeding him all over again. Each meal would take at least one to two hours.

Cooking for him, feeding him, cleaning up after his meal and cleaning him up consumed much of my energy every day. And days flew by quickly just like that. There was no time for me to care about other things. I had even neglected my elder son.

Only I could do it?

The nights were often too long, and the days too short. I wish there would be more time for me to spend during the day. It was hard to go on like this. The strain that repeated itself day after day, year after year, was taking toll on my body and mind, and I was slowly burning out. Despite warnings from my doctor and my worries about myself, I was most concerned about Kam-shing. Who would take care of him if I were not well? Who could possibly care for him like I did?

When he reached seventeen years old, I tried to talk him out. I told him about mommy's condition, asking him to try to go to sleep by himself. I was so glad that Kam-shing understood my hardship and loved me too. Since then, he would not need me to carry him to sleep, although he still needed me around so he could sleep peacefully.

20 August 2009. Kam-shing had a high fever of 39°C! I rushed him into the emergency room. However, the doctors there did not pay much attention to his very special needs. They just sent him into the Isolation Ward for adults as he was over eighteen. No matter how I explained and begged, no matter how

I went to the hospital everyday to plea to see my son, I was not allowed in. They did not allow me to stay in the hospital to take care of him either. They just insisted that my son was doing fine.

On the third day of his hospital stay, perhaps even the hospital staff felt that something was not right with my son, they asked me to go help feed him.

When I saw Kam-shing, I was shocked and was unable to recognise him! He was as thin as a stick. He looked dull and did not respond at all. All he did was drooling with his mouth opened, and he was bruised everywhere. I was so mad and worried! Later on the doctors checked that some of his thigh muscles were torn because of intense struggling. His skin and cardiac enzymes had also soared way up the scale. Just imagine how the medical staff had taken care of my son!

Besides feeling pain for my son, this hospital experience also made me understand that it was not enough to have me as the only one who knew how to take care of him. What would happen when I grow old? What if I were not around? With this new direction in sight, I began my "posthumous arrangement" about his care. I asked the hospital to help find him a suitable care centre. Ironically, all that the hospital could do was to refer him to a hospice care centre for the elderly. Should we try our luck by searching again, or pray for my own well-being? I had no idea.

Ever since Kam-shing was born, I no longer have a personal life. As a mother, I knew it was hard, but I was very willing to accept my life because he was my own son. I do not ask for much in life. I have already learnt to ask myself to have an open mind, that whatever saddened me, I should not take it to heart or think about it or remember. May be it is just a kind of helpless compromise, but it is the only way I know to make life easier.

Mother **Ngai Kong Lei-sun**
Son **Ngai Kam-shing**, patient with Glutaric Aciduria type I, 20 years old

Journeys in hope

Urdu is my mother language. I don't know any Cantonese.

I came to Hong Kong from Pakistan with my husband fourteen years ago. We had two little children, Mavish and Hamza. Every now and then, we went to Pakistan to visit our relatives.

Yet, good times did not last long. My husband left us, after finding out that our children had MPS when they were very young. Now I am left with only my children.

What happened?

It all started when Mavish and Hamza were about 5 years old and 4 years old. They had knee pain so badly that it was too painful for them to even put their heels on the floor. At the Queen Elizabeth Hospital Emergency Room, the doctors said, "Something is wrong with your children."

That's all they could tell me. In order to find out what was making my children sick, the doctors sent their blood samples to Australia for testing. It was found that they both had a chronic disease called Mucopolysaccharidosis (MPS), a rare genetic disease, and they had MPS type I. My heart sank even further.

Luckily their pain did not come often, and it did not last. They looked and played just like other children. No one would think they were suffering from a disease that was gradually taking them in.

When Mavish complained about knee pain one day, we were in Pakistan visiting relatives at the time. In my village, we have no hospital or clinic, and the nearest place for medical help is four hours away in Islamabad. Being a big city and the capital of Pakistan, Islamabad offers everyone treatment if he/she can afford it. So I took Mavish to the hospital there. Can you imagine how they treated her? They gave her the medication for TB (Tuberculosis)! Even I know that TB is in the lungs, not at the legs!

I also took my children to the Army Hospital, hoping it would be more advanced and would therefore know how to treat my children. But even they did not understand my children's health problem. "We've never heard of it in Pakistan." There was no MPS patient in Pakistan.

The choice

We decided to come back to live in Hong Kong, because the doctors in Pakistan did not understand my children's medical problems.

Another advantage of staying in Hong Kong is the culture. It is very common here for all adults in the family to work for a living. In Pakistan, only the men in the family work and pay for expenses of the whole family; the wives stay at home. So if we stay in Pakistan, with just me raising my children, I cannot afford their treatment.

The weather, healthcare and education system all prove that Hong Kong is the right place for us to stay.

Another factor is that the social welfare department, CSSA[1], has a rule that welfare recipients cannot stay outside of Hong Kong for more than 60 days in a year. So we cannot obtain regular treatment if we live outside of Hong Kong.

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1 Comprehensive Social Security Assistance (CSSA) Scheme

Reaching out for hope

Yet, living in Hong Kong is not easy, especially I was shortly left to sustain on my own with two young children.

I wanted to meet more patients and families going through life like me. My children also wanted to know more about why they were different from other children.

A social worker at the Assessment Centre in Kowloon City gave me the contact number of Mr Stephen Ma to find more support, saying that his child also had MPS and he was with the MPS patient support group. Stephen explained to us about the different types of MPS, introduced us to other families, and even helped us with Chinese/English translation.

One day I surfed the Internet and saw that there was treatment! It was called Enzyme Replacement Therapy ("ERT" in short) from a pharmaceutical company for patients with MPS. Mavish was around 7 or 8 years old back then, and Hamza a year her junior, both at the suitable age for treatment. However the treatment was very costly and we could not afford the cost of the drug which needs to be used weekly for the rest of their lives.

I contacted the pharmaceutical company to send information to me and my doctor about this treatment that we have been waiting for years. However, our doctor said that being a public hospital, they could not take the treatment from private companies, so we could not use it. Moreover, the Hospital Authority insisted that there was no treatment for MPS, but some investigations were still going on for ERT, so we may consider waiting for the ERT investigation results. All we hoped for is to use it as soon as possible, at least try it and see whether it can help my children.

As a single mother, I hope my children can get treatment soon, so they will not be hindered by the disease, and so they can take care of themselves and do not need to depend on me in case I am not around.

Other parents would ask me why my children are different. But no one knows the answer. My two children have different symptoms: Hamza has breathing problem but not Mavish, while Mavish has vision problems and Hamza can see clearly. Hamza needs to use C-pep (breathing machine) when he sleeps, but during the day he feels tired easily and cannot stand for a long time, so he cannot play or move around much.

Journey to treatment

My children and I have been going to Queen Elizabeth Hospital (QE), for medical treatment for many years.

When we wanted to get ERT, the doctor told us that it may be more likely to have ERT successfully at Princess Margaret Hospital (PMH). So I changed my children's medical appointments to PMH, and their doctor made their first application to Hospital Authority for using ERT. That was a few years ago.

PMH was closer to Mei Foo, where both of my children studied at the time. So it was more convenient for me too in case they didn't feel well at school.

But soon we realised that since we were making the request for ERT as individuals, our applications were not being considered as promptly as we have wished. So instead of doing it on our own, we applied for ERT again through the MPS patient group. Soon the Hospital Authority started evaluation on both of my children.

Recent surprise

We have had our medical appointments with QE for 12 years, and they have never investigated our family history or asked for genetic testing. Recently they wanted me to do it, and I was surprised, thinking what took them so long.

One week after the test, the doctor called me to the hospital and told me that the genetic defects in my blood and Hamza's blood were very similar, so it's very likely that we had the same problem!

Does that mean I may also have MPS? But I have never had the problems that my children have!

The doctor simply told me, "If you agree, we will send the blood sample of yours and of the two kids' to Taiwan to do a blood test to confirm if you have MPS."

Twelve years! And we are still waiting for another report to come back in two weeks. What we can only do now is to keep waiting and praying, and visit the doctors when my children are not feeling well.

Worries, and moving on

I know that I need to stay healthy so that I can take care of my children. Sometimes my neck, back and knees become so stiff, that recently I begin to practise yoga every week. I help Hamza to do some yoga at home as well, hoping to improve the flexibility of his body.

We live a simple life. My only hope is that with treatment, maybe my children can live their lives and enjoy things like other children. I know they won't be

100% cured and be like other children, but even if 90% similar to others, it will be good enough.

When I go to pick them up at school everyday, I really feel that I want my children to be comfortable and happy. When I look at other kids their age, I also wish that my children can play like them.

Mavish can take a bath by herself and do homework by herself. But at school she is very afraid that her pony tail will come loose, and no one would be there to help her tie her hair back up. She cannot do so on her own because of her stiff joints and bones. If her arms are a bit more flexible, she can take care of herself better.

Hamza only likes to play in two places in Hong Kong — Tokwawan and Kwun Tong — where he has friends and his own cricket team. Playing cricket, especially catching, improves his joints & arms, and he used to like it a lot. Now he cannot move as swiftly as before, so his interest in cricket is almost gone.

Hamza has once dreamed of being either a scientist or astronaut. Now, he wants to take a computer course and become a car designer, because he can just sit down at a computer to do it – that he can manage, so he calls this his "final decision". As his mom, of course I support his dream. He is happy when he talks about his dream, making me feel happy too.

Hamza usually has no strong feelings toward medical check-ups. But the last time when we came back from the hospital for a check-up, suddenly he cried, as he was upset that no doctor was trying to help and he was worried that his condition was getting worse. All that the doctors talk about every time is whether he needs surgery or not, without any treatment in sight.

It worries my child and I too. I love both of my children, and they are both important to me. But being the boy in the family, Hamza is special to me.

If my children can get ERT, may be there would be some improvement to their bodies. I hope one day they can grow up to be independent and can take care of themselves.

Mother **Nighat Sultana**
Daughter **Mavish Sultana**, 15 years old, MPS I
Son **Hamza Shahid**, also had MPS I, passed away in 2009 at the age of 13

* Post-script:

In August 2009, Ms Sultana was relieved by the genetic test result which confirmed she did not have MPS, although she carried the same genetic problems as her two children. Her doctor could not explain why this was so.

In September 2009, the Hospital Authority's "Expert Panel on ERT for Lysosomal Storage Diseases" approved for Mavish and Hamza to use ERT for treating their MPS I.

In October 2009, ERT was also approved for both children at Princess Margaret Hospital. Mavish began using ERT weekly and has been making progress since then, while Hamza's case was pending further medical evaluation and report.

In mid-November 2009, Hamza agreed to consider doing MRI after returning from his trip to Pakistan for a family wedding in December. The Sultana family had a joyful shopping day in Shenzhen before Hamza went to Pakistan to represent his family at the banquet and to bring holiday gifts to his grandparents and relatives there. He was scheduled to fly back to Hong Kong in early January 2010 for MRI, and to begin ERT soon. Sad news hit the patient support group on 3 December 2009. Hamza passed away - he fell on his back while walking in his home in Pakistan on 29 November 2009.

The world is for my child to see

I grew up in the sixties to seventies. In those days, women would get married and have kids when they turned twenty. I was no exception.

I met my husband when working at my father's shop. We decided to get married after dating for a few years. We were from poor families, so we decided to tighten our belt and save up a good sum of money before having children, in order for our children to have a better future.

When our days had become better, I also became pregnant. I thought life would be wonderful for us in the days to come.

I was almost thirty years old when I had the baby. My husband decided to call her Ka-po, as the name sounded like the two words "family treasure" in Chinese, and she indeed was the apple of our eyes.

Unfortunately, our wishes were not to be. No better plan could withstand the stroke of fate.

Coming to terms

Ka-po could speak only after turning one year old. When she called me "ma" for the first time, I was so happy that I kissed her over and over again.

At that time, her size looked the same as kids of her same age. So I thought it was just her speech ability that was falling behind.I took Ka-po to the health clinic for IQ test. At first the doctor said she did not have a big problem, so I thought it would be ok.

After several assessments, the doctor thought something may be wrong with her, so a referral letter was written for us to go to the Pediatrics Department at the Duchess of Kent Children's Hospital at Sandy Bay.

In my heart, I began to question what kind of illness could Ka-po possibly get.

On the day of the medical checkup, the doctors did a lot of tests but had no conclusion. They just asked me to keep bringing Ka-po in for follow-up checks, as well as for physical therapy and speech therapy.

I was worried. At that time, with no Internet available, information access was not as advanced as now. I could only enquire at many different community service centres, hoping that someone could give me some hints or pointers.

I was a frequent visitor at various hospitals because of Ka-po. In the beginning I was very worried, but eventually I was numb toward all the running around and unanswered questions.

Ka-po was already four years old when the doctors finally found out the cause of her illness. The doctor officially diagnosed her with Mucopolysaccharidosis (MPS) type III.

The doctor explained that this disease had no cure, and patients could only live to their twenties at best. Although she did not look much different than other people, she would have hyperactivity and intellectual problems, and as she grows up, she would slowly lose her ability to speak and comprehend.

Having already gone through several years of torment, I did not feel too sad when I heard the news. I thought to myself that we would raise Ka-po like someone with mental retardation. With hard work, I was sure that I could teach Ka-po well too.

Confronting hardship

We lived in North Point at that time. There was a pre-school and training centre that I frequently visited. The staff there said that the golden window to teach and train kids was between three and six years old. Ka-po was four by then, so I decided to grasp the limited time she had. I thought that if she could master eight out of ten things that she learnt, that would be good enough.

I quitted my job for Ka-po, so that I could give my full attention to take care of her.

Every morning around seven o'clock, I would wake up and bring Ka-po to kindergarten. The school promoted integrated learning, so that children with relatively slow intellectual growth could learn in a regular environment. That was why I did not mind taking her to this school at Wah Fu Estate in Aberdeen, which was far away from where we lived. I would then rush back to North Point to go to the market, do household chores and prepare dinner for the family. When afternoon came, I would rush to Aberdeen to pick Ka-po up from school. I would then take her to different therapies and training sessions.

Aside from receiving speech therapy and physical therapy provided by the government, the whole family cut down on expenses so that we could afford to let Ka-po have private sessions for additional speech and physical therapies, as well as sensory integrated training, which was not yet popular at that time.

All expenses for Ka-po's training took up one third of the family's income. Although it was a big burden, I did not mind at all.

In order to achieve the best results in learning, not only did I accompany

Ka-po in her therapies, I also jotted down all the main points from the therapists so that I could drill with Ka-po at home.

At the time, the teaching kits for speech therapy and sensory integration training were very expensive. We could not afford to buy a set for each, so we borrowed them from the therapists and made our own sets at home.

For example, when training Ka-po to identify different shapes, I home-made my own set of flash cards with different shapes drawn on them. I would point to a shape and told Ka-po at the same time, "Circle! This is a circle! Ka-po, look, circle!"

Other children could usually remember that after learning two or three times. However, Ka-po would need to have it repeated as many as ten times, even twenty times, before she could remember it.

I felt tired too in this long ordeal of training. Only when Ka-po could successfully recognise some words, I would feel better and have the faith to go on.

The hard work of these few years finally paid off. Ka-po passed the requirements for entering the Shatin Public School for primary education. I felt like I had been awarded the top prize and I was so happy, because that was the elite school among schools for disabled students. That was like being accepted into the Diocesan Primary School or La Salle Primary School for other parents.

After Ka-po came home from school on the first day of class, I was so shocked when I checked her homework!

Previously, it had taken me a year to teach Ka-po to know her name. Yet, her books from school were full of chapters with hundreds of words! They

looked simple to most people, but I knew in my heart that they would be nearly impossible for my Ka-po to learn.

With no alternatives, I created flash cards again for the words in her school book, then taught Ka-po to match them word by word. We studied again and again day after day, hoping that in the end she would have learned a little out of it.

Two years soon passed. Due to her limitations, she could only manage to learn a few words. The school principal figured that it would not be good for her growth if she stayed on to study like this. So the recommendation was to transfer her to a school for the moderately disabled.

I felt defeated by this, but I encouraged myself that I would support Ka-po to continue learning no matter which school she went to. Her IQ went downhill along with her condition. Now, she was like a newborn baby. She could not speak at all. She could not recognize anything.

Being discriminated

Actually what broke my heart most was not Ka-po's deterioration, but the look of other people.

Because of MPS, Ka-po's arms and legs would sway uncontrollably. Those with a kind heart would think of her as a bird who wanted to fly. But more often, people would regard her as a weirdo, a monster. This often made me feel hurt.

There were times when I took her to buy food at the market with me. We would walk in the middle of the road, so as to avoid the stalls on the two sides. I also had to make sure that Ka-po walked straight ahead of me, so

that I could prevent her from touching and pulling things around.

No matter how careful I was, there were still times of oversight. Ka-po had torn out a fish's tail and had damaged the leaves of vegetables before. When that happened, the hawkers would scream at us, "What's wrong with you? Didn't you teach your kid to have manners? What a bad kid! You come to damage other people's things. What a terrible family!"

At that point I felt so hopeless, feeling that no one on earth could understand our suffering and pain. The child was innocent and she did not mean it. She was only sick. Don't you have any sense of compassion?

Despite that, I am not going to hide Ka-po at home. I want my child to see many different things, so that she can feel the beautiful side of this world.

Ka-po is now 20 years old. My wish is for her to be well and can go with me to visit all corners of Hong Kong.

Mother **Leung Wong Pik-Ngor**
Daughter **Leung Ka-po**, patient with MPS III, 20 years old

Mother Tang Tsang Ka-lei

Still my big kids

I have four children. They were all born in the sixties in Hong Kong.

My first child Wai-man and my third child Wai-chi both have the rare genetic disease, Mucopolysaccharidosis (MPS). They are lucky to still be alive today, but I am still anxious and worried for them.

They have been tortured by this disease since birth, and their days are not easy.

This disease is not simple. It is caused by their bodies lacking an enzyme that can break down mucopolysaccharides, a type of complex sugar. Because of that, these complex sugars keep accumulating in their body, damaging their many organs. At first their joints became deformed and rigid, and their arms short, thick and bent. Later on, we found that their liver and spleen became very large, their abdomen bulged, and they had umbilical hernia. Their cornea became cloudy, and the disease also affected their eyes and ears.

It was in the seventies and eighties back then. The doctors had no idea how to treat them. The many efforts to check and examine were to no avail.

May be the heaven above was touched by my sincere intentions. In 2002, the doctors finally found out that my sons had the rare disease MPS type II.

Newborns with hidden disease

When Wai-man and Wai-chi were still babies, all our relatives and friends called them the lovely, cute little ones. They were very clever too.

I thought I was the happiest person ever, with four dear children. Little would I know that two of them were born with severe illness.

When Wai-man was two years old, he often had high fever and hernia. Each problem would take as many as three surgeries to resolve. Similar problems happened to Wai-chi, so we took extra care of them both since they were children. While they were frail, I had to make a living at the same time, so given our circumstances there was not much we could do.

As they grew, their joints started to have problems, becoming rigid and deformed. Their arms and legs also showed signs of bending even when my children tried to straighten them. Their fingers also appeared claw-shaped. I was alarmed because they were also shorter than other children their age. Wai-man and Wai-chi's own younger brother and sister were way taller than them.

I took them to see the doctor. The doctor just muddled through, saying he had not yet found out why they were sick, and comforted me that it was not a big deal. I did not know much, and I did have trust in the doctor back then. Now to think of it, if I were more vigilant, we might have known earlier what made the two brothers sick.

In search of a good doctor

Doctors with western medical training have been treating them by taking stopgap measures, dealing with the symptoms and not the cause. The real cause of their disease was not in sight. I was so anxious and worried that I attempted other ways to treat them.

My two boys were still in primary school then. I asked around for ways to treat them, and many people suggested bringing them to mainland China for help.

I thought to myself that it was worth a try. Being proactive was better than to sit back and wait. After discussing with my husband, we began to contact

our relatives in the mainland to see if anyone could refer us to any famous doctors.

Finally we had a reply that there was a famous bone-setting master.

Despite our knowledge that bone-setters are experts in joints and bones only, we still wanted to give it a try. So my husband and I took the two boys to the mainland during the holidays, in hopes of at least finding out what was wrong with their joints, how to treat it, and give them the proper remedy.

It turned out to be a disappointing trip. The master did not know what was wrong with them, why their joints were so stiff, and had no way to help them. We could not believe that our first trip for medical help had ended in vain.

Since then, we kept on trying other treatments and finding out the cause of their illness. We took Wai-man and Wai-chi to Guangzhou this time.

We went to the famous First Affiliate Hospital of Sun Yat-sen University in Guangzhou by the referral of my distant cousin. Despite our experience last time, we still brought with us some hope, thinking that the doctors in the big hospital could help Wai-man and Wai-chi. The doctors first examined the two boys, then gave them acupuncture and used Chinese medicine to clear their nasal.

Acupuncture was a very painful experience for children, and my two boys were not willing to do it then. They kept crying that they wanted to go home to Hong Kong. I tried my best to comfort them and somehow managed to bring them to the hospital several days in a row. I only hoped that the acupuncture and nasal clearing would take effect soon. The doctors there had a soft heart too and did not want the boys to go through this intensive treatment. So they asked us to return to Hong Kong first, and feel free to consult them again if the boys' condition did not improve.

The expenses of the trip cost us most of our savings then. Yet, it was worth it as long as there was a chance to cure the two boys and to help them become healthy again.

After we had returned home, their eyesight and hearing did improve, giving us hope that they were getting better. However, the alarm in their body went off again soon after, and they had to frequent the hospital again.

Since the treatment in Guangzhou was not effective, and it probably did not target effectively at the illness, we did not go to Guangzhou for follow-up anymore.

Turning away from my grandchild

In 1996, my other son became a father. His pregnant wife asked me to help take care of my grandchild. To many people, this would be a happy occasion. However, deep down in my heart, fear seeped in.

At that time, the cause of Wai-man and Wai-chi's illness was still a mystery. What if it was I who passed them the illness? Or was there a voodoo casted on me that made them sick? After considering long and hard, I declined the kind invitation of my son. Now to come to think of it, I regretted for missing the joyful moments of raising my grandchild.

It was until 2001 that we finally learned the truth.

My youngest child, my daughter, was afraid to get pregnant after being married for seven years, for fear that she would be in my shoes. She was persistent in asking around for the reason behind her brothers' illness. With great efforts, she found an organisation with information about treating MPS in Taiwan. So she travelled there hoping to find out whether it was MPS that made her brothers sick. After examinations by specialist doctors there, we

finally knew what happened. My two sons did have the rare genetic disease MPS type II.

Since she had found out that her brothers had MPS, the doctors insisted on having our whole family undergo special assessment when my daughter was pregnant in 2004. We all have our blood samples sent to Australia for detailed assessment. When the results came back, it was found that the cause of my two sons' illness was due to a defect in my gene.

I felt so bad! I was the one who made my sons sick for their entire life!

For a long time, I kept on blaming myself for implicating my two sons. Later on, my children all comforted me, telling me that I was not to be blamed, and that no one would possibly expect that defects in the tiny genes could cause such damaging effects.

I have now let go of my scars. I want to use all my energy to protect them and take care of them. It is way more practical than to have negative thoughts and keep blaming myself all day.

Wishing them a longer life

Wai-man and Wai-chi have the mild type of MPS II. They are lucky to have lived till now. Although they need some help on daily chores, they can take care of themselves, and their IQ are no different from others. I feel blessed for these.

Their disease does not seem dangerous or fatal, but as a mother, I am scared that they will suddenly die and leave me one day.

Just like that one time, when Wai-chi suddenly puked out blood and had bloody diarrhea, I was so scared and worried.

It was found out that his liver cirrhosis had led to esophageal variceal bleeding. Previously the doctor had also discovered a polyp at his vocal cord, which made it difficult for him to breathe, and the doctor had a hard time intubating him to stop his bleeding. At that time, Wai-chi was suffering so much. As a mother, my heart ached badly when I saw him like that. He kept on saying that he did not want to live anymore; he would rather end it all, to stop the pain and suffering.

Of course I was reproaching myself and I did not know how to comfort him. Aside from bringing him food and giving him a sip of water whenever he needs, I could only support him silently by his bedside.

Luckily the doctor was able to stop his bleeding in the end, and Wai-chi was able to get through this hurdle in life. But it took him two months to get better.

As a parent, all I wish is for my children to be healthy. I wish so much for Wai-man and Wai-chi to live a normal, healthy life from now on. I hope that the government can provide medical treatment for my sons. As long as they have a chance to receive enzyme replacement therapy, their condition will improve, and they will no longer need to count their life in days and can have new hope for the future. Why? Because they are human beings, and human beings are all equal. That is my greatest wish.

Mother **Tang Tsang Ka-lei**
Son **Tang Wai-man**, patient with MPS II, 46 years old
Son **Tang Wai-chi**, also patient with MPS II, 43 years old

Mother **Christina Hellmann-Ipsaryaris**

From a mother

Stormy course

Hong Kong has been a place I call home for some years. Being someone who has travelled and lived at various places around the world, I thought that I have seen it all. The least I expected was to undergo the turmoil in the last few years, especially in my role of a woman and a mother.

I am now on a mission that I totally did not anticipate in the first place. It was all for my dear son, Joshua, who had lived an active and inspirational life for fifteen years. He passed away in October 2007 after a courageous five-year battle with the rare disease — MELAS Syndrome[1].

A new journey together

My journey of understanding began on Christmas Day 2002, when Joshua suffered the first of his ten strokes. He was only a ten-year-old child then. After countless hours of questioning and medical tests, we were no closer to understanding the cause or finding a solution. As a parent, I felt powerless given the lack of knowledge, information and resources available. I was desperate to find answers.

While medical professionals did their very best to diagnose the cause of Joshua's illness, there were many frustrations which we, as a family, encountered along the way to a correct diagnosis. Eventually, we learned that he was afflicted with a disease caused by defects in the genetic material of the mitochondria, the parts of the cells that generate energy for the body.

1 MELAS stands for Mitochondrial Encephalopathy, Lactic Acidosis, and Stroke-like episodes

Even after diagnosis, we encountered many challenges in treating this rare disease. As MELAS is a multi-symptomatic neuro-degenerative disease, it became a full-time job for me to care for my son. It was particularly difficult even in a multi-ethnic city like Hong Kong. Being expatriates from Switzerland, we often find it not easy to communicate with the locals. The rare disease that Joshua lived with was hard to explain to anyone, and there was neither any cure nor ways to help him get better. I felt quite helpless and hopeless despite the kind support from our circle of friends.

In the end, Joshua was rendered blind, deaf and unable to speak. The strong character of my son, however, means he is not a person to give up easily. He lived courageously and with a vision to assist other people afflicted with orphan diseases. This gave me a new goal in life and I was to be the one to help him accomplish this vision.

The fight

Joshua's life was shorter than expected, but his courage was extraordinary. I was and still am very proud of him. He taught me what courage and optimism meant.

Here is a chronological account of Joshua's fight against MELAS:

Before the age of 5-6:
Joshua was a very intelligent child. He could speak three languages and could recognise over 200 airline logos by the age of two years old. Joshua had no problem with his health. He enjoyed physical activities and was just like any other child, probably even brighter than his peers.

From the age of 5-6 onwards:
Joshua suffered from vague symptoms including recurrent vomiting, exercise intolerance, easily getting tired and clumsiness. He no longer liked to exercise as other children did. We sought the advice of our family doctor on Joshua's conditions and were reassured of anything untoward. Some investigations were performed but the results were negative.

I kept on searching for answers that caused his illness but to no avail. It occurred to me that time well spent with my children was the most important. My two younger daughters were probably too little to understand my worries then.

At the age of 10:
Christmas Day 2002 in Hong Kong. Joshua had his first stroke, with headache and visual problems.

At the age of 10 and a half:
Joshua had his second stroke while our family was in Switzerland.

At the age of 11:
Joshua was genetically diagnosed with MELAS in Switzerland.

Joshua had recurrent strokes, prolonged seizures / epilepsy. He later became impaired at his vision, hearing, and mentally, as well as bed-stricken, which severely diminished his daily activities.

At the age of 14:
He was given an innovative treatment for MELAS – *Arginine*, and coupled with adjustments of anticonvulsants, Joshua's condition became much more stable.

At the age of 15:
Joshua passed away in October 2007 after suffering from severe aspiration pneumonia and multi-organ failure.

In his name

As with many hundreds of other rare disorders and illnesses, MELAS is deemed an "orphan disease". An orphan disease is one that has been "orphaned" or neglected by the medical community and the pharmaceutical industry in terms of diagnosis, research and development of treatments due to its rarity, thus generating low levels of profitability and unattractive returns on investment potential.

Joshua lived his life courageously, optimistically and with a clear vision to assist other people afflicted with orphan diseases. It is in his legacy that I founded a foundation for orphan diseases in his name, in hopes of turning my personal loss, profound grief and my experience of overcoming challenges into something positive for those who might share similar circumstances and stressful times.

The mission of the foundation is to advance the awareness, diagnosis, treatment and research of orphan diseases, and to improve the welfare of children with orphan diseases and their families in Hong Kong. It focuses on raising awareness and providing support for the diagnosis of orphan diseases among patients of age 18 and below, who have been recommended by licensed pediatricians based in Hong Kong. I also hope to provide a source of information and resources related to the diagnosis, care and treatment for patients and their families afflicted with orphan diseases.

Early diagnosis is another key focus area of the foundation, as it is often the area that presents a bottleneck to treatment. We ultimately hope to make equal access to accurate diagnosis to all children in Hong Kong who are suspected of suffering from orphan diseases. Through my own experience, the long wait and blind guesses were very painful and not helpful at all. I hope that no family will need to go through the pain like ours.

Orphan disease patients often face enormous difficulties in obtaining an accurate and timely diagnosis or even basic information about their conditions; this is because their physicians are often unfamiliar with their symptoms, which are usually vague and confusing. Since most orphan diseases are heavily under-researched, patients and medical professionals often lack the information they need to develop appropriate options and leverage the resources that are available.

Diagnosis and possible treatment for a significant proportion of orphan diseases are not routinely available and covered by the Hospital Authority. Therefore, in order to have a professional diagnosis, funding is required. The cost of an individual diagnostic test can run into the thousands; this is simply too much for many families to afford. Some tests are not even available in Hong Kong, and so families who know about these tests often need to go overseas or send their samples overseas, and wait for results.

Not an orphan

I am deeply grateful and most fortunate to have gathered a team at the foundation, which shares a genuine interest in continuing my son's vision of hope. This team includes some of the leading medical

specialists in Hong Kong and abroad. Through our shared knowledge and experience, and with the help of our supporters, I aspire to improve the quality of life of all children who suffer from orphan diseases in Hong Kong.

I am also grateful to my friends and family who gave me support along the way. Through the many supporters and people who care and lend a helping hand, with remembrance for my dear son, I feel that I am not walking the path alone, and I have confidence that his courage and perseverance will live on in us.

Mother **Christina Hellmann-Ipsaryaris**
Son **Joshua Hellmann**, patient with MELAS Syndrome, passed away in 2007 at the age of 15

Hitting the jackpot twice

It has been many years since I talked about my story. I wonder how many others have gone through what I have been through. There is a story to every family. What is my story? I sincerely hope that each parent can understand and be aware that when a new life is brought to this world, there is a chance for the baby to have genetic disease.

I felt like I had won the lottery twice, which brought me unforgettable experiences for the rest of my life. The second wave hit just when I had recovered from my first fall. Why did it have to be me?

The little treasured one

I come from a big family of eight brothers and sisters. All their children grow up healthily. My daughter also grew up well and has completed her university degree. I also had two sons, who gave me the impression of tumblers that quickly righted themselves. They were always cheerful and happy, and they would eat anything, their favourite being barbequed pork buns.

My eldest son Man-hon was born in 1976. Soon after his birth, we found that he would get sick easily with breathing problems, and he often had seizures. He had not yet learned to speak when other kids his age would already be nagging their parents. Although our family was well off enough to find help from specialist doctors in Hong Kong, they were at a loss as to how to treat him.

Broken dreams of emigration

I believed that Canada's environment and healthcare system would be good for my son, so we decided to jump on the bandwagon back in 1982 and arranged for investment emigration to Canada.

The steps to migrate to Canada were to first obtain immigration approval

before getting physical exams. We passed each and every immigration requirement except for Man-hon's physical exam. At that time, we told the immigration officer that we planned to pay for all his medical bills in the future, so the Canadian government would not need to pay a penny. But his application was still rejected. In order to make our case a success, our immigration lawyer even suggested letting someone else adopt our son, so that our family could move to Canada! How could there be a lawyer as heartless as this?

That marked the end of our emigration plan.

Sadly, Man-hon left us when he was 13 years old.

My younger son Ying-yu was born in 1985. Because of his arrival, my broken heart glowed in warmth and I saw hope in life again. Ying-yu was a very cute boy even as a baby. Unfortunately, soon after, I realised that his arms, legs, joints and tummy looked swollen. Also, whenever the weather changed, he would have breathing problem like his brother did, and it would take him a long time to recover.

In his early years, when his symptoms have not yet shown, I prayed everyday for a miracle that he would grow up healthily. I thought that with no findings, with no evidence of illness, that meant everything would be OK for me. I soon realised that I was lying to myself.

When Ying-yu was ten years old, the doctor gave us the bad news. He did have the same rare genetic disease as his brother, and it was called Mucopolysaccharidosis (MPS). It was still in the eighties to nineties back then. The diagnosis did not make much difference to us, because the disease was not treatable.

Just like his brother, Ying-yu had the severe type of MPS type II. His condition deteriorated quickly and he was suffering his whole life. When he was around six to eight years old, he could still go to special education school everyday at the Caritas Lok Yi School. However, he was crippled by the time he was

fourteen. His basic necessities of life had to be taken care of around the clock. In his last few years in life, he was bedridden and had to stay in the hospital.

As a parent of children living with MPS, I often hurt my wrists and shoulders because of the countless liftings and continuous care-taking for my child. But, what little pain that was, compared to what my child was going through!

Keep on trying

Seeing Ying-yu slowly deteriorate from an active, fun-loving child to a boy who could only express himself through his glance, it really broke my heart.

We tried everything for our son, anything that could possibly help him: Western medicine, Chinese medicine, *Qigong*, massage, etc. We even travelled all over mainland China in hopes of finding him a treatment or cure. We did not have the fantasy of him being totally recovered from his disease. We simply hoped to lessen his pain and suffering.

Although we had tried many ways and none could cure him, I met several children with MPS in Beijing and Qongqing, and realised that my child was not the only one with this disease. Unfortunately, we did not speak the same dialect, so I could not communicate with them to exchange information.

Ying-yu was able to live a few years longer than his brother. Everyone at home still missed him so much. His nineteen short years was a bitter life to him. The most important thing was for us as family members and myself as the mother to provide him with the best possible, to comfort him when he was sad or hurt, and to make him feel that there is love and care in this world, so that he can experience the bright and happy side of things.

What comforted me most was that he loved to smile way more than others. His happy little face never showed a frown. Life was a joyful ride; perhaps that was his blessing! Our whole family loved him, cared for him and treasured our memory of him from the bottom of our hearts.

If there were no SARS

In his final years, Ying-yu could only smile and barely open his eyes. He could only be a listener. Later on, all he was able to do was to blink his eyes. Whenever he saw a member of the family, especially his daddy, he would blink so hard and so much, as if he was afraid that we would miss him out. Also, whenever we touched him, he would try to push back at us as much as he could. That was his way of responding to us.

I stayed at the hospital with him everyday. I massaged his arms and legs with sandalwood oil to help improve his circulation. I told him every little thing that happened to us in our daily lives. Whenever his daddy had to go on overseas trips, father and son would have regular telephone calls. Ying-yu would listen patiently to what his daddy was saying at the other end of the phone and responded with the rhythm of his breathing.

His IQ was higher than other people, so I knew that he understood everything that he had seen and heard. I told him frankly that he did not need to miss us at all if he no longer wanted to suffer and live on like that. He could go peacefully. When he heard me say that, his tears started to roll down, as if he was responding to me that he did not want to walk away like that.

At that time, he required frequent suction at his airway so that he could breathe, as well as tubes for breathing and feeding. He had already lived in the hospital for two years, and his condition was regarded as stable.

If it were not for SARS, he would not have left us so soon.

SARS hit Hong Kong in May 2003. Our little world was not peaceful anymore. The hospital where he was staying had implemented quarantine measures because of SARS. Ying-yu was sent into private wards and I was only allowed to visit him for one hour everyday. That was not enough at all for me and Ying-yu. During the day there were four nurses on the floor, while only two to three nursing aids at night. No one had time to care for him. To Ying-

yu, that was as if he was totally isolated from the outside world!

Although the chief nurse had made a request to hospital management on our behalf so that we could stay with him longer or pay him another visit in the morning, the hospital did not allow us to visit him outside of the limited visitation hours.

That afternoon, my husband who was then in the United Kingdom, gave me an anxious call immediately after his phone chat with Ying-yu. He said that he could not hear our son's breathing. I tried to stay calm and told him that things were fine and he should not worry while overseas. However, I had a bad feeling that our son may not live long.

Sure enough, at around five in the morning the following day, the hospital called me, saying they were trying to resuscitate Ying-yu. I reached the hospital at six, but by that time Ying-yu had already gone….

He was gone. All I could do then was to stay by the side of his tiny body. The hospital was considerate for us then, and let us stay in the room for almost four hours. My maid and I cleansed his body, talked to him, recited Buddhist prayers with him, telling him that he did not need to miss mommy and daddy, telling him to go peacefully to heaven.

After Ying-yu had gone, there was one miracle after another. I believed he was trying to tell us that he had reached the yonder, as Buddhist believers would put it.

Life goes on

Although both my sons were now gone, my thoughts of them will long remain in my heart. All I could do was to do something in their legacy.

In the names of my two sons, I sponsored the founding of a school in Hengshan province in mainland China. We named it the "Hon Yu Hope School", using

the names of my two sons. Every year, we donated several scholarships to children with financial difficulties there, to lay the path for them, hoping that one day they could attend university.

One day, I read from the newspaper about a girl called Chan Man-fong, who also had MPS. It was then that I knew about other patients with that disease in Hong Kong. Through the help of the reporter, I was in touch with several patients such as Wong Wing-kan and Eric Ma. With the help of the social worker from the Social Welfare Department, we were introduced to other families who were living with MPS. I felt so touched to know them. Since then, we had gatherings every two to three months. This really helped us in supporting and sharing what we had learned with each other.

At the same time, Ying-yu's red pocket savings of ten thousand dollars was used as the operating fund for a new patient support group. Founding the patient group was to provide support to families who are living with rare genetic diseases, as well as to lift the misconception that parents have nothing to do with the illness of our children. The defective gene comes from us, so we must be responsible for our next generation. We should take care of them and share our experience with others.

Today, this support group has become widely accepted by patients and the public. I am sure that Man-hon and Ying-yu will both feel consoled.

Mother **Ching Ma Oi Lee, Ellie**

Son **Ching Man-hon**, patient with MPS II, passed away in 1989 at the age of 13

Son **Ching Ying-yu**, also patient with MPS II, passed away in 2003 at the age of 19


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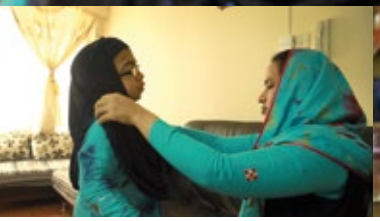

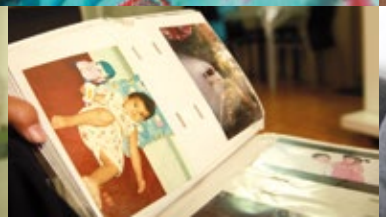

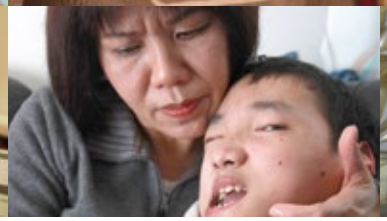

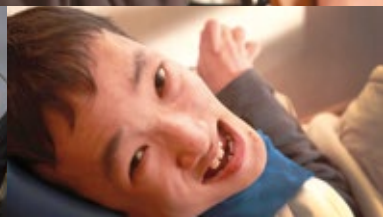



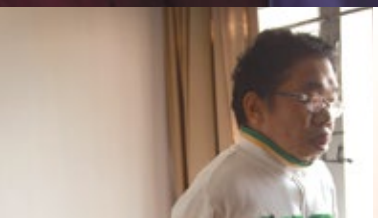




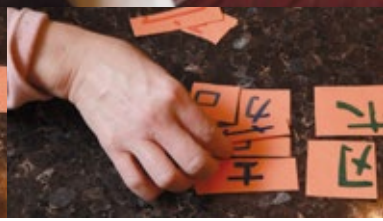

Thank you !

HK Society for Rehabilitation
Community Rehabilitation Network
Rbluesky
Maggie loh
Ducky Tse Chi Tak
City University of Hong Kong,
Department of English and Communication
Ron Wong
Noel Chan
Joonie Lai
Pearl Leung
Wimmy Lam
Derek Kwan
Dr JE Wraith (UK)
Dr Gregory Pastores (USA)
Dr Paul Harmartz (USA)
All kindergartens,
primary and secondary schools,
and universities that support us
Kid Says
Annie Lau Sum-Yau
Disney VoluntEARS
Jaime Fong Ka-yau
Ronald Leung Vik-lun
Jason Chan Pak-yu
Fiona Fung Hoi-yu
Queen's Scout (HK) and
Queen's Guide Club
HK Medical Association Orchestra
Hong Kong Bank Foundation
BioMarin Pharmaceutical Inc
Genzyme Corporation (Hong Kong)
Mannings
Motta Communications

Thank you!!

Equal Opportunities Commission & Joshua Hellmann Foundation

About HKMPS

Hong Kong Mucopolysaccharidoses & Rare Genetic Diseases Mutual Aid Group

HKMPS was formed by patients and families living with rare genetic diseases for mutual support and encouragement. With the assistance of The Hong Kong Society for Rehabilitation's Community Rehabilitation Network, the Group later registered as a charitable organisation on 23 March 2005.

Apart from Mucopolysaccharidoses (MPS), HKMPS also have members who are diagnosed with Mucolipidosis, Glycogen Storage Disease, Multiple Sclerosis, Gaucher's Disease, Pompe Disease, Hereditary epidermolysis bullosa, Phenylketouria (PKU), Glutaric aciduria Type I (GA I) and Sotos Syndrome. All patients with rare genetic diseases are welcome to join as well.

The Group receives no regular funding from the government. Our operational expenses are mainly covered by annual membership fees and donation, whereas individual activities are supported by funds from different charitable foundations. The Group does not have a permanent address. We borrow venues from the Hong Kong Society for Rehabilitation for meetings and gatherings.

Please visit our website **www.mps.org.hk/en** or contact us **info@mps.org.hk** for more information about us and about rare genetic diseases.

Support Us

If our stories have inspired you, please support us!

Your donation will help with implementation of self-help and mutual aid efforts among our patients. Your support is greatly appreciated. Hong Kong residents who donate $100 or above can apply for tax deduction with our official receipt.
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ISBN: 978-988-99703-0-7

Little Giants—Dreams of Braving MPS

Publication Date: September 2006 (First edition) |
Publisher: Bbluesky | Language: English | Pages: 80 |
ISBN: 988-98181-7-5

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Publisher: Bbluesky |
Language: Traditional Chinese, Simplified Chinese |
Pages: 108 | ISBN: 978-988-19578-4-9

罕有父母 *(Rare Parents)*

Publication Dates: February 2010 (First edition),
May 2010 (Second edition) | Publisher: Bbluesky |
Languages: Traditional Chinese, Simplified Chinese |
Pages: 144 | ISBN: 978-988-99703-9-0
(The Traditional Chinese, printed version of this book is still available in small quantities and can be purchased from us. Price: HKD 68.)

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Publication Date: July 2008 (First edition) | Publisher: Bbluesky |
Language: Traditional Chinese | Pages: 56 |
ISBN: 978-988-99703-3-8

醣豆豆　大夢想 *(Little Giants—Dreams of Braving MPS)*

Publication Date: September 2006 (First edition) |
Publisher: Bbluesky |
Language: Traditional Chinese, Simplified Chinese |
Pages: 80 | ISBN: 988-98181-6-7

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Language: Traditional Chinese
Pages: 191 | ISBN: 978-981-25914-4-9 | Price: HKD 68

大不了的3呎高人生—罕有病少年的濃縮人生自白書

Authors: Eric Ma, Stephen Ma | Publication Date: June 2010 |
Publisher: 青源出版社 | Language: Traditional Chinese | Pages: 184 |
ISBN: 978-988-80217-9-6
(The electronic version of this book can be purchased from the online bookstore of Handheld Culture Limited. Price: USD 3.99 (Approx. HKD 30):
http://store.handheldculture.com/book.php?category_id=7&book_id=606)

寫出生命的彩虹—一位黏多醣症女作家的自白

Author: 陳雯芳 | Publication Date: 1999 | Publisher: Metro Broadcast Corporation Limited | Language: Traditional Chinese | Pages: 105 |
ISBN: 978-962-85596-5-7

Rare Parents

Book Title: Rare Parents
Published by: Bbluesky Company Limited
Publisher: Leslie Ng
Editor: Bbluesky Creative Team
Translator: Inez Wu
Photographer: Ducky Tse
Graphic Designer: daydrea:ming
Published on: May 2010
ISBN: 978-988-99703-0-7
Price: HK$68

Bbluesky Company Limited

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All parents share an ordinary wish, which is for their children to live and be well. But for parents with children suffering from rare genetic diseases, this simple wish is an impossible dream. Children with rare genetic diseases cannot be healed, and their conditions cannot be diagnosed before birth. They have to face daily challenges to their body and will in their very short lives.

Rare Parents features touching, personal stories of 12 parents whose children were born with rare genetic diseases — these are stories of shouldering pain, suffering, despair for the little ones, accepting and overcoming difficulties with courage, and immense love for their children and themselves.

Rare Parents is the third book of Hong Kong Mucopolysaccharidoses and Rare Genetic Diseases Mutual Aid Group. Proceeds from the sales of this book will help toward improving patients' quality of life.

It has been said that children with rare diseases are specially arranged by heaven to be born in the most loving families — I totally agree! After getting to know them, I was shocked to find so many selfless and inspirational stories behind every child's life.

"A good sword comes from intense sharpening, while the scent of a plum flower comes from enduring severe winters."

Bless you all!

Chea Shuk Mui, Candy
Radio Host

Parents of children with rare genetic disorders experience tremendous life turmoil and challenges. Their perseverance and unconditional love and care to their children are definitely a reflection of the true meaning and value of life.

From guiltines, depression and hopelessness to acceptance with positive embracement is in no doubt a very difficult but fulfilling introspective path. These parents have absolutely exemplified how they can become the true angels of their children and live with selfless love. We should give a big applause to all these parents with great respect.

Dr Huen Kwai-fun
Consultant of Hong Kong Mucopolysaccharidoses and Rare Genetic Diseases Mutual Aid Group

HK $68

ISBN 978-988-99703-0-7
9 789889 970307