She wasn't expected to be alive at 12 but girl with rare Pompe disease thrives on parents' love

Raj Nadarajan/TODAY

Chloe Mah, 12, at home with her mother Patricia Ng and father Kenneth Mah.

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Individuals with the genetic disease lack an enzyme to break down glycogen in the body

Its excessive build-up affects the heart and skeletal muscles

Most infants diagnosed in the first few months of life do not live past one or two years without treatment

One Singaporean couple tell how they have been raising a child with this rare condition who is now aged 12

SINGAPORE — At 12 years old, Chloe Mah cannot do basic activities such as eating, breathing properly and sitting up on her own due to muscle weakness.

Yet, she loves watching K-pop music videos, having her nails painted and playing games on her iPad. Her face lights up whenever she sees people grooving to her favourite music videos.

Her family counts every day with her a blessing. This is because Chloe has a rare fatal condition called Pompe (pronounced Pohm-peh) disease.

She was diagnosed with the condition when she was seven months old and doctors did not see her living past 12 months.

She has now outlived their expectations.

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Affecting one in every 40,000 births, the genetic disorder is so rare that KK Women's and Children's Hospital (KKH), where Chloe undergoes treatment, has seen just four cases in the past 15 years.

Pompe is caused by a mutation in a gene that results in a deficiency in an enzyme called alpha-glucosidase (GAA).

Associate Professor Tan Ee Shien, senior consultant and head of genetics service in the department of paediatrics at KKH, said: "Normally, the body uses this enzyme to break down glycogen, a stored form of sugar used for energy. In Pompe, mutations in the GAA genes reduce or completely eliminate this essential enzyme."

Excessive build-up of glycogen accumulates in the body, seriously affecting the heart and skeletal muscles.

Besides having low muscle tone, Chloe also had an enlarged heart and its function was failing at the time of her diagnosis.

Her father Kenneth Mah, 51, recalled that his daughter was not expected to survive past her first birthday. Yet, she has thrived and reached milestones not commonly seen in other infants diagnosed with Pompe's, he added.

In an interview to raise awareness of the rare disorder in conjunction with International Pompe Day this month, Mr Mah talked about his firstborn's near-death experiences and the struggles his family has faced, including negative comments from people who say Chloe has "no hope" and criticise them for trying to keep their child alive.

"We went through some really difficult times but we are always thankful and grateful to be able to spend so much time with Chloe. Every year is a bonus to us," he said.

On the naysayers, Mr Mah said that the family have encountered their fair share of people and "keyboard warriors" who make hurtful comments.

"Chloe needs to go to the hospital for her infusions. It's a 'need' for us but some people define it as a 'want' to keep our child alive," Mr Mah said.

"They ask why we prolong her suffering; why not let her go? Why spend so much on her? No one sees (treatment) as a future for her," he said.

He was upset by the hurtful comments at first but has since learnt to take them in his stride.

"My wife is calm and asked why I had to be so bothered about what they say because they are not in our position. Furthermore, we are not seeking sympathy, only empathy. If they had any empathy, I'm sure they would not say things like that," he said.

BREATHING DIFFICULTIES, HEART FAILURE

KKH sees mainly cases of infantile-onset Pompe disease, which means symptoms begin in the first few months of birth. Assoc Prof Tan said that patients with this form of the disease are usually most severely affected.

Besides low muscle tone and delays in meeting motor milestones such as sitting, rolling over and standing, these babies also have weak heart muscles. Heart failure can occur in the first year of life, she said.

There is another form called late-onset Pompe disease, where symptoms manifest later from childhood to adulthood. Assoc Prof Tan said that individuals experience progressive muscle weakness, often starting with difficulty in climbing up stairs.

As the disease progresses, it can affect their lung muscles and machines are needed to help with breathing.

While the effects may vary from patient to patient, Assoc Prof Tan said that skeletal and heart weakness is a characteristic of the disease and causes mobility and respiratory issues for patients, regardless of age.

However, their cognitive abilities remain good, she added.

Diagnosing the disease can be challenging. To confirm the diagnosis, a blood sample is taken to measure the level of enzymes in the patient or a genetic test is performed.

"As Pompe disease is rare, it is not often thought of early in the diagnostic process," Assoc Prof Tan said.

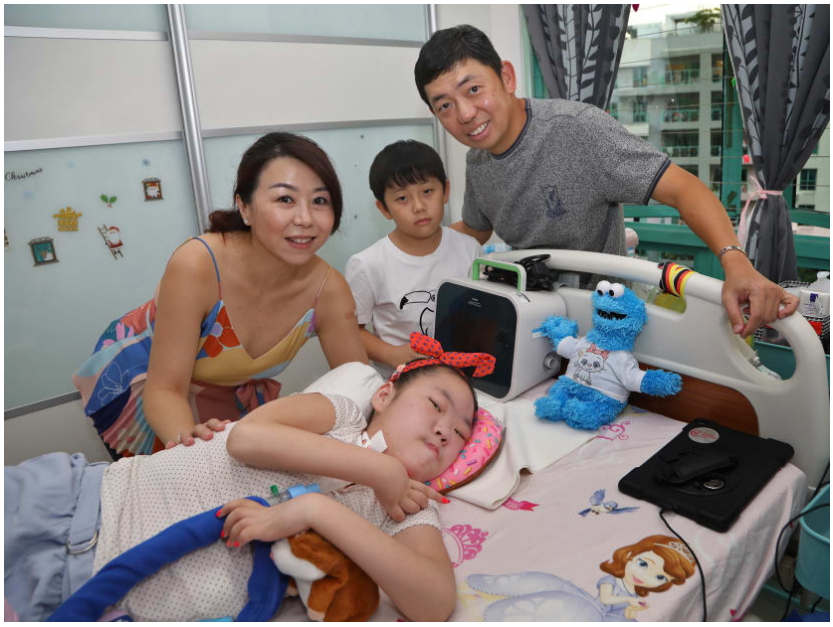
"The symptoms can be rather vague and there are other more common neurological diseases that can give rise to similar symptoms."

A paediatrician initially thought that Chloe, who was six months old at the time, had a bout of lung infection and she was referred to KKH.

X-ray images later showed an enlarged heart, and she was later confirmed to have Pompe's after her blood sample was sent overseas for diagnosis.

Recalling the hopelessness he and his wife had felt at the time, Mr Mah said: "Chloe is our first baby. It was very difficult for us to accept it."

They now have a younger son aged nine.



Chloe Mah (in bed) is the first child of Mr Kenneth Mah and Ms Patricia Ng. She has a younger brother Cayden. Photo: Raj Nadarajan/TODAY

COSTLY TREATMENT

At the time, the Mahs were given an option of an enzyme replacement therapy, which is an approved treatment for all patients with Pompe disease. Without treatment, Chloe would die.

Assoc Prof Tan said that the treatment involves administering — intravenously — a special drug containing recombinant human acid alpha-glucosidase (rhGAA) that helps break down excess glycogen not required by the body.

"Timely diagnosis is essential to ensure that the patient gets appropriate care. The lifelong treatment involves replacing the missing enzyme. If started early in the disease, it is able to preserve muscle function and prevent deterioration," she said.

"Although patients are not fully cured, the enzyme replacement therapy has been shown to extend the life expectancy of patients."

However, it came at a cost of around S$90,000 for the first year of treatment, Mr Mah said. They decided to give

it a shot and have not looked back since Chloe responded well to the treatment.

In 2015, she met a major milestone: Her cardiologist stopped giving her medication for her heart because she no longer had a risk of heart failure.

While the treatment has prolonged Chloe's life, there are other challenges.

In 2012, she underwent surgery to pass a tube directly into her stomach to help with her food intake.

She went under the knife again in 2019 for a tracheostomy to support her breathing issues. This involves creating an opening in her neck to place a tube into her windpipe so that air may enter the lungs.

There is often no one-size-fits-all treatment for rare disorders such as Pompe. Assoc Prof Tan said that treatment is patient-centred and requires coordinated efforts of a multidisciplinary team.

Another treatment looks at relieving symptoms and supporting patients. For example, respiratory support helps patients whose breathing is compromised.

At the same time, the patient may also be supported with physiotherapy to improve strength and physical ability, occupational therapy, speech therapy and other measures to ensure proper nutrition and weight gain.

Assoc Prof Tan said that there are many clinical trials looking into improving the treatment for Pompe disease as well as ongoing research that looks into the use of gene therapy.

A NEAR-DEATH ENCOUNTER

The Mahs have spent the past decade troubleshooting problems as they arise.

Mr Mah closed his mobile phone business to be Chloe's full-time caregiver while his wife, Ms Patricia Ng, 47, is the

breadwinner of the household. She works as a compliance officer.

Although extensive insurance coverage, which they had gotten for Chloe when she was two months old, eases the financial impact, Chloe's living expenses still add up substantially.

A monitor is used to measure her heart rate and oxygen level, while a cough assist machine helps clear secretions from the lungs. She also requires support to breathe.

When TODAY visited their home in the central region of Singapore, Chloe was nursing a bedsore that had trouble healing and has not gone to school since the start of the year. She attends classes at the Rainbow Centre, which caters to people with disabilities.

One of the most frightening times the family went through was in 2018, when Chloe had a cardiac arrest.

Mr Mah said: "To see your child's heartbeat slowly going down and her turning purple in front of you, it was scary for me. She went ice-cold. I went crazy."

When her heartbeat and oxygen levels flatlined on the oximeter, he made a desperate attempt to revive her by performing cardiopulmonary resuscitation.

He lost track of time and could not tell how long he kept at it. Although he managed to revive his child, Mr Mah said that he experienced conflicting emotions of relief, self-doubt and guilt afterwards.

He has seen similar incidents whereby children who were revived too late suffered permanent brain damage and had to be hooked on life support.

"There was a fleeting moment when I had asked myself if it was time to let my child go. But as a father, how can I not do anything to save my daughter?"

Miraculously, Chloe survived the incident without apparent

damage.

Mr Mah said: "The first thing she did when she came round was ask for her iPad and started playing with her game, Temple Run. The same old Chloe came back to us."

The incident taught him something important: "I learnt that in times like these, I just have to do what I need to do (as a father) and leave the rest to God, and to tell myself to live without regrets."

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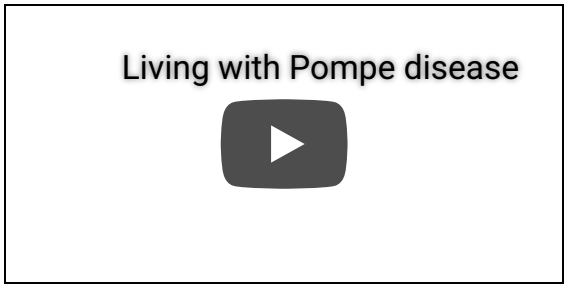

BANDING TOGETHER FOR STRENGTH

Chloe's condition spurred her parents to raise awareness of rare diseases and champion for the rights of children with rare diseases to be given the opportunity to grow and develop their abilities.

Being a minority among the population, many patients and their families receive little to no attention while coping with the devastating impact of rare diseases and the required caregiving needs.

Together with two other families battling other forms of rare disorders, the Mahs co-founded the non-profit organisation Rare Disorders Society Singapore, which offers financial support through various support

programmes. It now supports close to 150 families.

As co-founder and vice-president of the society, Mr Mah contributes on a voluntary basis. He does not receive renumeration for this.

"It's all about coming together, supporting one another. Love is not a rare thing."

KEEN EAR FOR MUSIC

To her parents, Chloe is living proof that when given the opportunity, support and love, children with rare disorders can still grow and live meaningful lives.

She can now make sounds and uses a communicator application to express her needs and emotions.

"Everyone told us she probably can't talk but when she was eight years old, she started imitating sounds that we made. That was the first time I heard her call me 'papa'," Mr Mah said.

"The joy is really — I can't describe it, (I can) only cry over it."

In recent years, Chloe has shown an impeccable sense of rhythm and a keen ear for music. For example, she taught herself to play songs such as Amazing Grace using a music app.

Like any parent, the Mahs have a simple hope for their daughter. They want her to lead a happy and fulfilling life while achieving her own potential.

Ms Ng said: "We always emphasise that we are not just keeping Chloe alive. We want her to enjoy her own abilities rather than look at what she cannot do."

They live "day by day" and try not to think too far ahead.

"Her life expectancy is not within our control. But for as long as she is still around, we just want our daughter to be happy," Ms Ng said.

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