BY MEDICAL OUTREACHERS

VOICES

OF RARE DISEASE PATIENTS & THEIR STORIES

To access the *Voices* website and read the full versions of the life stories excerpted in this book, please scan the QR code found below.

We would like to thank Rare Disease Hong Kong for supporting our first rare disease life project. We would also like to extend our gratitiude to all rare disease patients, patient group leaders, medical students, and doctors for taking part and sharing their most personal, inspiring, and touching stories.

- 11th Medical Outreachers, Local Team

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FOREWORD: WORDS FROM MR. K.P. TSANG

Mr. K. P. Tsang,
Chairman of Rare Disease
Hong Kong

During the first few years of growth of Rare Disease Hong Kong (RDHK), students from the two medical faculties have been our inseparable fellow partners.

There are new students joining our team batch after batch.

Amid the hectic study schedule, they squeeze in some time to make friends with the rare disease patients.

Not only do they try to understand the medical needs of the rare disease patients, but they also strive to know the patients as a person who has other human needs.

Every year they report their work progress and brilliant achievements to us, unknowingly becoming the ambassadors building bridges between the future doctors and patients.

Fellow students, having you by my side makes me feel grateful and inspired.

Shoulder to shoulder, let's keep going on our journey together!

FOREWORD: WORDS FROM PROFESSOR EDWIN CHAN

Professor Edwin Chan,
School of Life Sciences, Chinese University of Hong Kong

Rare diseases are the concern of scientists, of doctors, of caregivers, of patients... Nonetheless, rare diseases are also the concern of all of us, for it is a matter that touches upon every one of our lives.

To most people, rare diseases might seem like a distant and abstractconcept, irrelevant to them and their families. However, do you know that among every 67 Hong Kongers, one of them is diagnosed with a rare disease? Once you start understanding the causes of rare diseases and the variety of syndromes and illnesses that fall under this umbrella, you will then realize rare diseases are never far from your life.

We are grateful to have a group of medical students who, under their own initiative, compiled a book consisting of the life stories of over a dozen rare disease patients, bringing to light the different issues faced by rare disease patients in Hong Kong.

I hope that through reading this book, you will have a more thorough and vivid understanding of rare diseases and the lives of the patients, caregivers, warriors, and leaders that they affect.

WORDS FROM RARE DISEASES HONG KONG

Established in December 2014, Rare Disease Hong Kong (formerly known as Hong Kong Alliance for Rare Diseases; HKARD) is the first patients' group in Hong Kong comprising of cross-rare-disease patients and their families with the support of experts and academics in the field. Its objectives are to rally supporters to spearhead and improve related policies and services, promote public education on rare diseases, and strengthen the community's support for patients in order to ensure respect and protection for patients in terms of such fundamental rights as healthcare, social support, education, and daily needs equal to other citizens.


“Rare is many,
rare is strong
and rare is proud”
- Motto of 2020 World Rare Disease Day
香港罕見疾病聯盟
RARE DISEASE HONG KONG

WORDS FROM MEDICAL OUTREACHERS

Medical Outreachers is a certified charitable organization comprising of medical students from CUHK and HKU. Entering our 11th anniversary this year, Medical Outreachers has within this short decade expanded from local services for asylum seekers, homeless people, terminally ill patients, and rare disease patients to overseas operations in remote suburban China and Nepal communities.

Rare disease patients are one of the most neglected demographics in Hong Kong. Not only do they face issues such as incorrect diagnosis by physicians, lack of subsidy for medication and treatment, lack of clear policy by government of HK, their inspiring yet tear-jerking life stories are often obscured from public view.

Through this Rare Disease Project, we have enabled medical students to interview the life stories of rare disease patients in HK and share these stories to the public. We hope to be able to shine a light on the hidden plights of rare disease patients, and at the same time build up the empathy and communication skills of medical students, enabling them to become better doctors in the future.

Focusing on long-term projects designed to make a lasting, sustainable impact, Medical Outreachers helps volunteers reflect on the human side of medicine while empowering them to take action to alleviate the suffering of fellow human beings.

“We will always strive to reach out, create and inspire”

Rebecca Yuen

Mother of TSC patient

Tuberous Sclerosis Complex (TSC)

Cause: Mutation in TSC1 or TSC2 gene

Inheritance: Autosomal dominant genetic disease; in some patients it is caused by genetic mutations

Prevalence: Affects about 300 people in Hong Kong.

Symptoms: Seizures, tumours on vital organs, cognitive and behavioral disabilities. Severity varies with location and size of tumor

Effects: Affects functions of vital organs as well as cognitive and mental health

Treatment: Antiepileptic drugs to control seizures, medication needed for tumours and behavioural problems as well

"I wish rare diseases would become a common term. Just like the flu."

– Rebecca Yuen, Chairwoman of Tuberous Sclerosis Complex Association of Hong Kong

“I am always willing to try. Because not trying will get you nothing.”

JUST ANOTHER FILE

As Yiu Yiu’s conditions worsen, with multiple seizures occurring daily, Rebecca felt like her daughter’s time was almost up. However, her persistent inquiry for her daughter’s condition was met with a cold response- “Your daughter is just another file stacked up in my office”. This emphasises the harsh reality of our healthcare system. It may be easily forgotten amidst the long and busy hours, but patients are more than just a diagnosis written on paper. They are “files” with feelings and most importantly, they need empathy from our healthcare workers. Despite these heartless responses, Rebecca never gave up. She said, “I realised my tears will never wash away my haunting reality. And these doctors were still my only hope. So I had to face it, I just had to keep trying”. That mentality was what got her through.

Rebecca and her daughter Yiu Yiu, a TSC patient

Sandy Chan

Mother of HAE patient

Hereditary Angioedema (HAE)

Cause: C1 esterase inhibitor deficiencies

Inheritance: Autosomal dominant genetic disease

Prevalence: 1 in 50,000

Symptoms: Sudden swelling attacks in any body part that lasts a few days

Effects: Unpredictable swelling affecting all kinds of normal daily activities including breathing

Treatment: Available treatments for HAE such as C1 Esterase Inhibitor has yet to be included in the Hong Kong drug registry for subsidy; in comparison, these medications have already been registered and widely used overseas

"I don't want the disease to control me or my life. I want to be able to control the disease instead. so that I can also live a normal life without interference."

– Sandy Chan, Founder of the Hong Kong HAE patient group

Sandy and her daughter both diagnosed with HAE

"We really need the drugs to save our lives. They are already in the market. I can't see why the government won't register the drugs."

A STRUGGLE FOR ALL

When her daughter was first diagnosed, Sandy's family felt completely alone. At that time, there was no HAE patient group in Hong Kong, nor were there any registered HAE treatments. The unpredictable nature of HAE attacks resulted in severe disruptions in daily lives of patients— the frequency of attacks varies, and could render patients unable to work or go to school. Even if modern treatments were accessible, it would still be impossible for a normal household to afford these expensive treatments without any subsidies in the long run.

Hence, with the support of HAE international, Sandy along with a group of dedicated patients and doctors founded HAE HK in 2019. Their aim is to improve the quality of life of HAE patients in Hong Kong by raising awareness about HAE in the community, providing a platform to share information and experiences, as well as advocating for access to modern treatments.

Even though the road ahead is long and hard, Sandy is determined to create a better future for HAE patients in Hong Kong. She will continue to do her part in raising public awareness about HAE, and she will not cease to fight for access to modern and subsidised HAE treatments.

Terry Lai

Pompe disease patient

Pompe Disease

Cause: Accumulation of glycogen in lysosome due to deficiency of acid alpha-glucosidase enzyme, leading to damage to nerve and muscle cells across the body

Inheritance: Autosomal recessive inheritance disease

Prevalence: About 10 Pompe Disease patients in Hong Kong

Symptoms: Muscle weakness, decreased lung function, scoliosis

Effects: Movement and respiratory difficulties, might require wheelchair and ventilator

Treatment: Enzyme replacement therapy

"I want to support those rare disease patients searching for the light at the end of the tunnel, to help them shorten the days they have to live in darkness and suffering."

- Terry Lai, Advocacy and Communications Officer in RDHK

“In rare disease families, the burden shouldered by parents cannot be described by words. They are indeed the greatest and the most selfless.”

LIGHT AT THE END OF THE TUNNEL

Back when his brother was first diagnosed with Pompe disease, 9-year old Terry was already warned he might share the same fate. Even so, Terry at the time remained in denial he might also be a Pompe Disease patient.

The turning point for Terry and his brother came in 2009. That year, as his brother’s condition rapidly deteriorated, doctors told Terry’s family to prepare for the worst. Yet even as the rest of his family gave into despair, Terry refused to give up. After exhausting all available resources, Terry at last caught wind of a new enzyme replacement therapy developed overseas that was about to launch in the commercial market soon. To save his brother’s life, Terry decided to bring the issue of rare disease under media spotlight, so as to raise public awareness and persuade the Hospital Authority to allocate funding for the new treatment.

The success in saving his brother’s life made Terry believe in the power of advocacy and gave him the motivation to serve in patient organizations. Terry described his experience as being stuck in a sea fog, where his life is full of unknowns, and slowly the fog parted to reveal light at the end of the tunnel.


37TH HONG KONG FILM AWARDS
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HONG KONG ALLIANCE FOR RARE DISEASES
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Kelly Lee

Mother of 1p36 deletion syndrome patient

1p36 Deletion Syndrome

Cause: Genetic deletion of the distal light band on the short arm (p) of chromosome 1

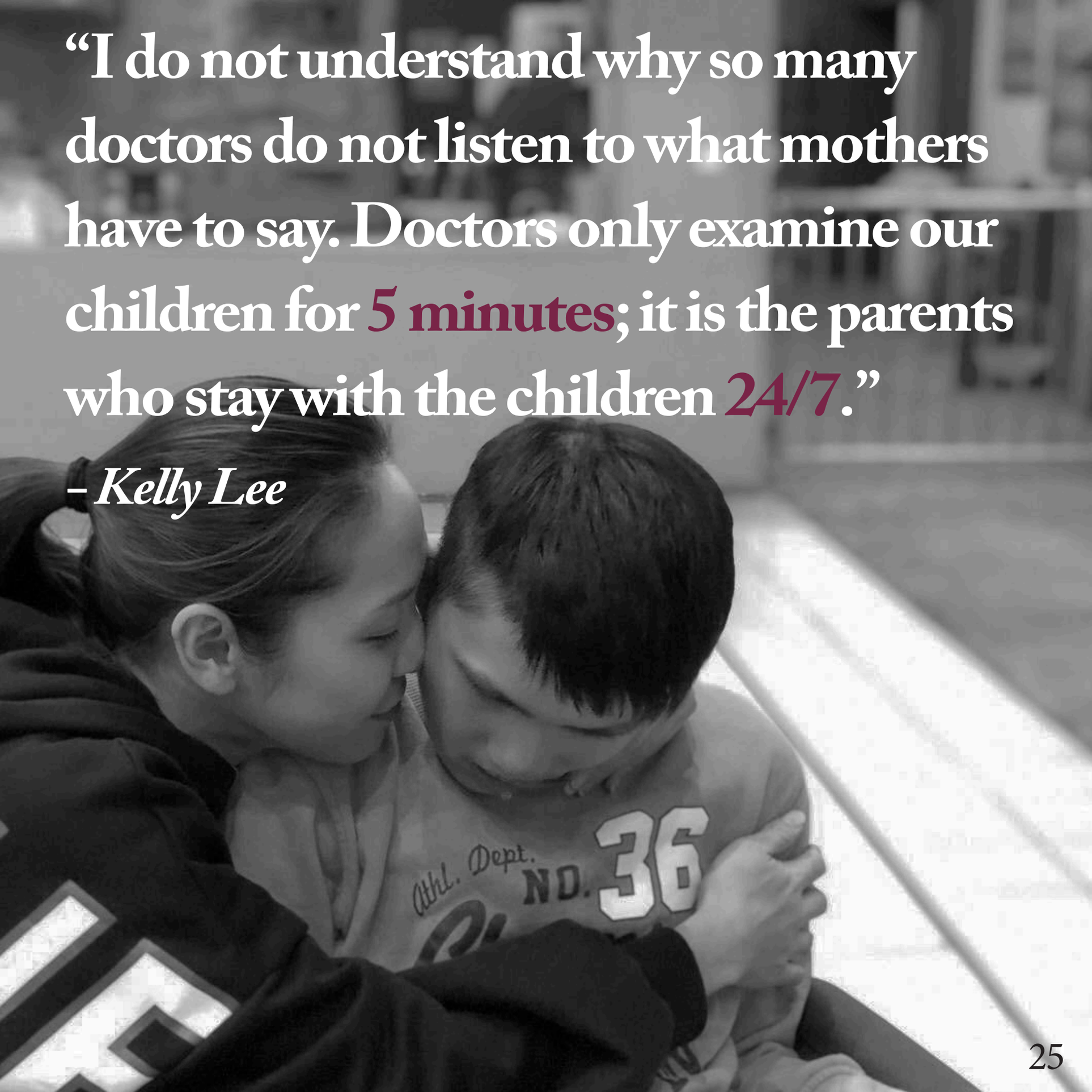
Inheritance: 95% of disease is caused a random chromosomal mutation during meiosis or in early fetal development

Prevalence: 1 in 5000 to 1 in 10,000 (0.01% to 0.02%) of live births

Symptoms: Severe intellectual disability and delayed development during childhood, such as speaking difficulties, behavioural problems and self-harm behaviour; physical symptoms include small head, distinct facial features and visual problems

Effects: 1p36 children are not able to develop normally and live a normal life

Treatment: No known cure for 1p36 syndrome; treatment used to control symptoms of the disease such as seizures



"I do not understand why so many doctors do not listen to what mothers have to say. Doctors only examine our children for 5 minutes; it is the parents who stay with the children 24/7."

– Kelly Lee

 Kelly Lee's oldest son Edwin, a 1p36 deletion syndrome patient

“Once, a social welfare officer told me I should send my son to a long-term hostel instead of wasting my efforts taking care of him. Those words were enough to cause me to break down in tears.”

KELLY’S WISHES

Kelly is a single mother of a 14 year old 1p36 deletion syndrome patient called Edwin. To Kelly and Edwin, who are frequent visitors to hospitals, having to wait to see a doctor often proves to be a huge challenge. As a 1p36 patient, Edwin’s patience is extremely limited-- he throws tantrums and self harms when forced to wait. That is why after each visit to the doctor, Edwin and Kelly often go home covered in bruises and tears.

Kelly does not wish for much for the future—she does not expect Edwin to live a long life, instead only wishing for her son to be happy. Regarding COVID, Kellys says, “I am not worried about Edwin losing his life due to COVID, since that means he will be able to go to heaven sooner. The only thing that distresses me is the thought of Edwin being restrained in the hospital and suffering physically and mentally as a result.”

Tara Sam

Mother of aHUS patient

Atypical Haemolytic Uremic Syndrome (aHUS)

Cause: Formation of blood clots in vessels and in the kidneys

Inheritance: Most cases of aHUS are genetic; however, some may be acquired due to autoantibodies, whilst others are idiopathic

Prevalence: Affects 2 individuals per 1 million

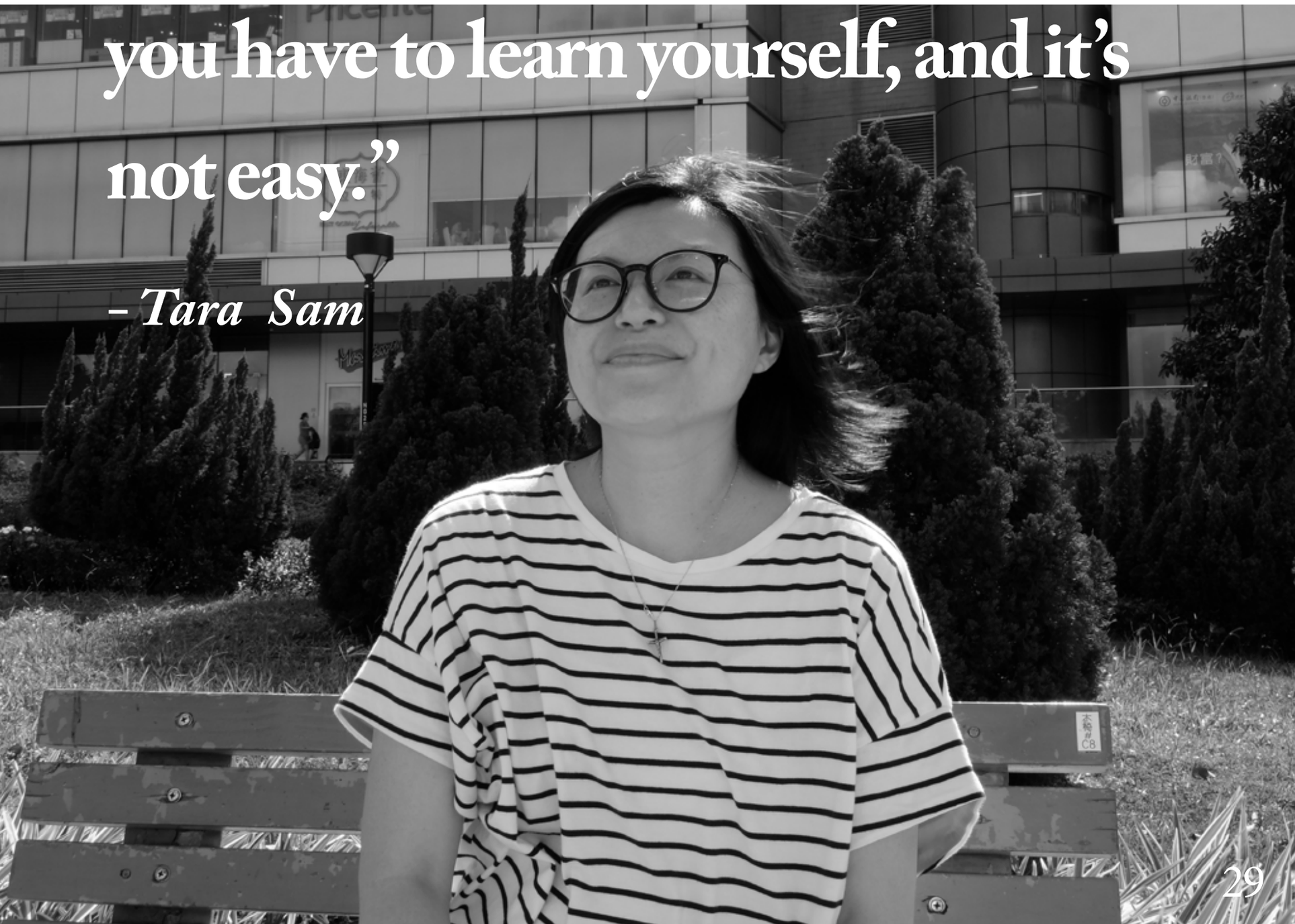
Symptoms: Disease that causes blood clots of form in various vessels of the body, primarily in the kidneys; most individuals will develop hemolytic anemia, thrombocytopenia, and acute kidney failure

Effects: Patient suffer from kidney damage, and may need dialysis

Treatment: There are only two drugs, eculizumab and ravulizumab-cwvz, which are used to treat aHUS, kidney transplants may also be needed

“It all happened really quickly, I didn’t have time to react apart from crying. There are no instructions – you have to learn yourself, and it’s not easy.”

– Tara Sam



“When I found out my child was diagnosed with a rare disease, all my expectations and goals went down the drain.”

A MOTHER’S DEDICATION

Tara is the mother of Kyle and Cyrus, who are both diagnosed with atypical haemolytic uremic syndrome (aHUS). Currently, Kyle, Tara’s second son, also suffers from end-stage kidney failure due to aHUS at the mere age of 9.

Tara reveals the drastic changes in her and her husband’s lives that had to be made in order to accommodate the needs of their children; she had to learn how to operate a dialysis machine and has to be extra wary of personal hygiene to protect her immunocompromised children.

Tara hopes that in the future, the Hong Kong government will pay more attention to the rare disease community and allocate more resources for information provision so that rare disease patients and their caretakers will know where they can find help.

Tara Sam's second son Kyle who is diagnosed with aHUS

Jessie Yu

Mother of anti-NMDA receptor encephalitis patient

Anti-NMDA Receptor Encephalitis

Cause: Autoimmune disease caused by antibody attacks against NMDA receptors

Inheritance: No evidence shown that it is hereditary

Prevalence: Affects 5 patients in Hong Kong

Symptoms: Convulsion, hallucination, fever and lost of memory may occur. Thus, the clinical manifestations resembles that of psychosis and schizophrenia

Effects: Many patients can recover, but may suffer from memory loss, change in personality, etc.; it may take a long time to train lost skills again

Treatment: Drugs such as rituximab and cyclophosphamide can be used. If the disease is caused by an ovarian tumour which produces the antibodies, surgical removal could be done

"Fighting against a rare disease is like practicing high jump—one must put their focus on the progress made, be it 1cm or 10cm, and do not focus on past failures." – *Jessie Yu*

"Joint clinics by a team of doctors from different specialties is multitudes better than each specialist seeing a patient on their own."

A NEW BEGINNING

Jessie Yu is the primary caregiver of her daughter, a girl suffering from anti-NMDA receptor encephalitis since 2018. Despite undergoing a plethora of diagnostic tests such blood tests and bone marrow biopsies, doctors failed to diagnose her daughter's illness. It was not until her daughter fell into a coma and had to be transferred into the ICU that doctors finally managed to find the correct prescription to relive her symptoms. Even so, the damage was done, and due to an injured hypothalamus, Jessie's daughter lost all short-term memories as well as the ability to sit, speak, walk, and write. Nonetheless, this did not deter Jessie from accompanying her daughter in every step of the recovery process.

Jessie Yu's daughter, diagnosed with anti-NMDAR encephalitis

Eric Ma

MPS Type VI patient

Mucopolysaccharidosis Type VI (MPS)

Cause: Patients lack the enzymes that digest glycosaminoglycans, which would accumulate in their body gradually, and destroy body cells

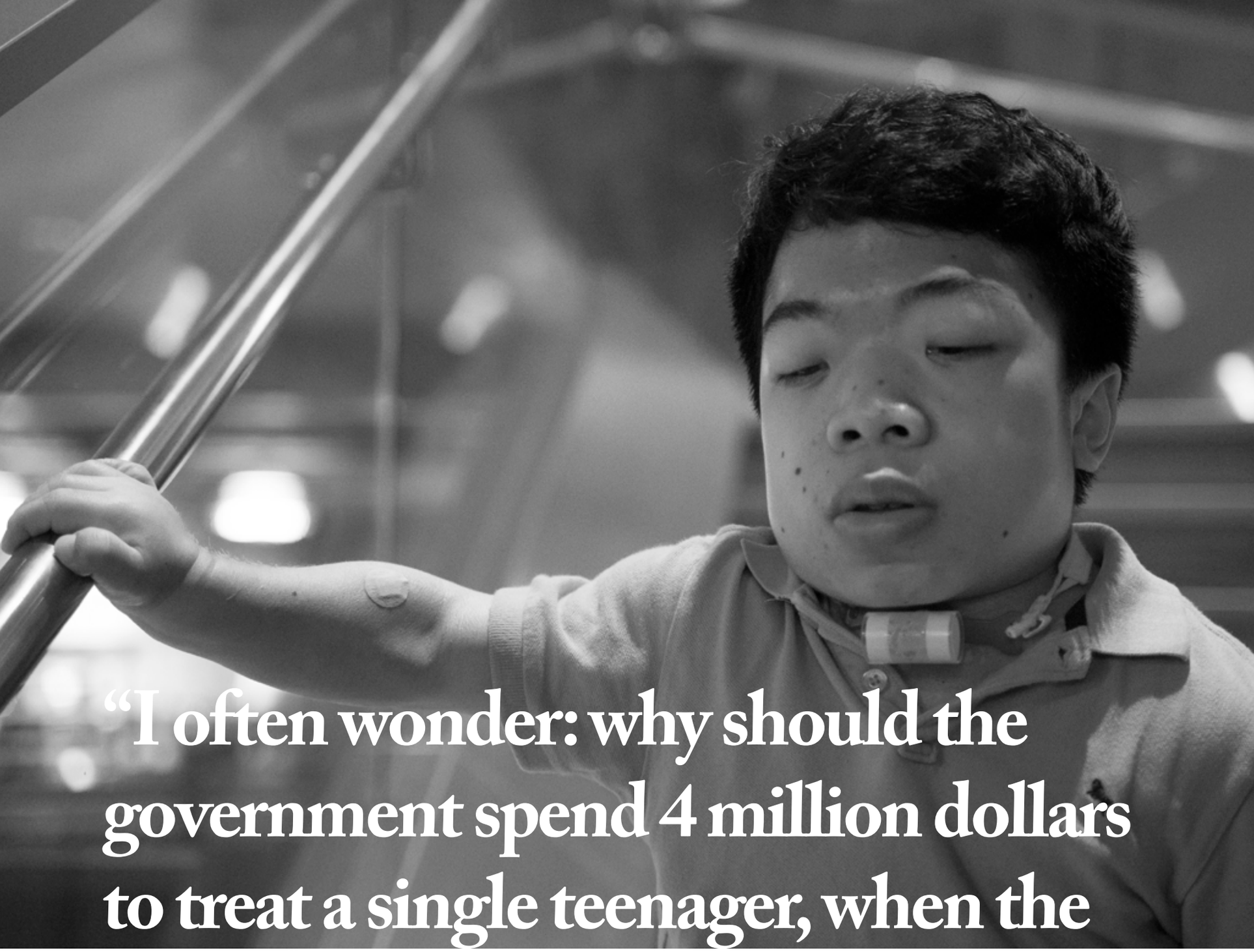
Inheritance: Autosomal recessive disorder

Prevalence: Estimated to occur in 1 in 250,000 to 600,000 newborns

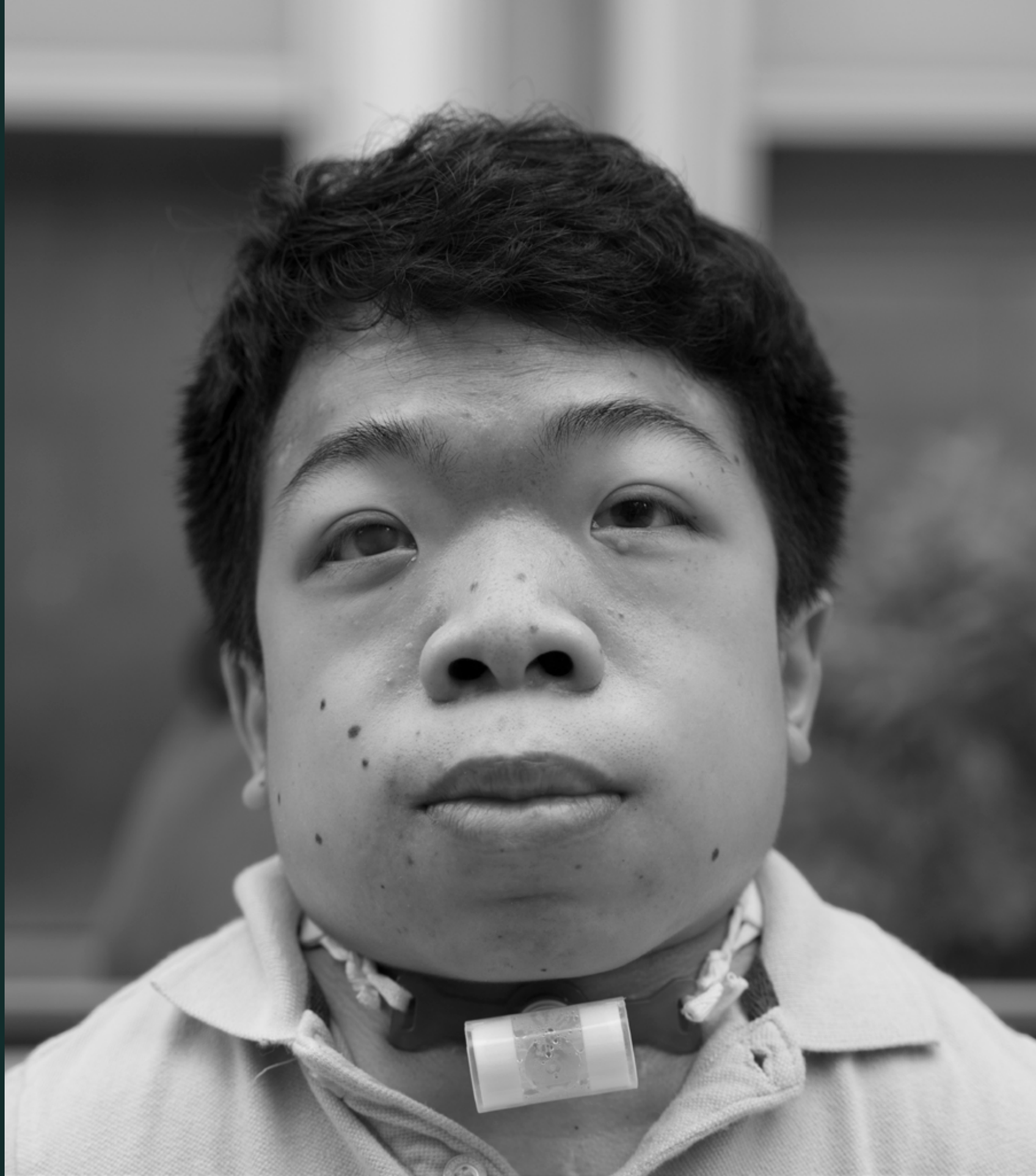
Symptoms: Short stature, rough facial features, recurring respiratory infections

Effects: Heart-related diseases, abnormal growth of bones affects motor ability, different senses like eyesight and hearing will deteriorate gradually

Treatment: Type VI patients can receive enzyme replacement therapies to improve their symptoms, and bone marrow transplantation has proven effective



"I often wonder: why should the government spend 4 million dollars to treat a single teenager, when the same amount of money could be used to give 100,000 people the flu vaccination instead?" *-Eric Ma*



"The government has the ability to develop policies for rare diseases, but they never act until the society pushes them to do it."

THE GENEROSITY OF HONG KONG

Among all the things he experienced since diagnosed with MPS VI, Eric is most grateful for the love and generosity he found in the midst of Hong Kong. Indeed, as Eric often asks himself— why should the government spend four million dollars to save a NEET, when it could be used to give 100,000 people the flu vaccine? What makes him thankful is that even though Hong Kong people are profit-minded, people still understand rare disease patients are not unwilling to pay for medicines, but simply cannot afford them. They also strongly support the cause of rare disease groups, such as donating and volunteering often. This made Eric deeply feel the love of Hong Kong people. Thanks to his medications, Eric has already retired from the front line of advocacy, but he hopes that his story can help patients stand up and rely on their own strength to strive for policy progress. Eric also referred to himself as an ant; although small and inconspicuous, ants can be powerful when gathered together, just like rare disease patients. As long as they work together, they can finally bring about change.

Clayton Lo

Undiagnosed rare disease patient

Undiagonosed Disease

Cause: Undetermined, but suspected neurological issues involving skeletal and tendon development

Prevalence: About 1 in 13 people suffer from a rare, undiagnosed condition

Symptoms: neurological issues such as light and noise sensitivity, hyper-active immune system disorder, joints inflammation, sore limbs especially during mornings

Treatment: No known treatment, though Clayton have been attempting alternative medicine such as chiropractors and chinese medicine

“Not only was my experience in the public healthcare system time-consuming and humiliating, it also made me recognize my marginal position in society.”

– *Clayton Lo*

“Some doctors are willing to acknowledge my symptoms cannot be explained by our current understanding of medicine; however, this is not accepted within the government’s framework of healthcare.”

A HISTORY OF DISAPPOINTMENTS

Clayton Lo, a 42-year-old literary creator and editor, has suffered from an undiagnosed rare disease since he was 12 years old. Over the years Clayton has visited countless physicians yet the result is always the same-- after several tests with no diagnoses, he will be referred to a psychiatrist. In the end, either Clayton gives up out of frusturation; or the doctor believes that there are no more medicines to prescribe and gives up. Because mainstream medicine fails to treat his disease, Clayton has turned to alternative medicine such as bone-setting and chiropractic.

The undiagnosed status of Clayton's disease prevents him from getting the social support other disabled people usually enjoy. For instance, family and friends are sometimes skeptical of his claims and think his symptoms are subjective instead of objective problems. Even so, Clayton strives to remain honest to himself and hopes one day he can find the root cause of his disease.

Bithiah Nip

Myasthenia gravis patient

Myasthenia Gravis

Cause: Autoimmune disorder onreceptors that receive nervous signals on muscles.

Inheritance: Non-inheritable

Prevalence : Around 1000-2000 patients in HK

Symptoms: Drooping of the eyelids, double vision, weakness of muscles in the face, causing chewing and swallowing problems; and weakness of muscles in the limbs

Effects: Symptoms listed above usually appear after prolonged activity and hard work; the condition improves with rest

Treatment: Managed with various medications, and removal of the thymus gland may alleviate symptoms


“I only have
one wish now:
to live a good
day, every day.”
– Bithiah Nip

“I know more from patient organizations than from doctors.”

THE PROBLEMATIC HEALTHCARE SYSTEM

For Bithiah, the Hong Kong healthcare system has failed to give her appropriate treatment. After being diagnosed with Myasthenia Gravis, she realized how both the government and doctors have insufficient knowledge regarding rare diseases. She was particularly troubled during the start of her diagnosis as she could not obtain any information about the disease and medications from her doctors. She said frankly, "I learnt more from patient organizations than from doctors." During the first two years after her diagnosis, she was admitted to the hospital for nearly ten times. When she needed more medication, the process took another six months and only a small dosage was approved. Bithiah emphasizes her disappointment and how there is a lack of research, data, and experience with rare disease patients in HK.

Shirley Choi

Scleroderma Patient

Scleroderma

Cause: Autoimmune disease (immune system attacking connective tissue under the skin, around internal organs and blood vessels)

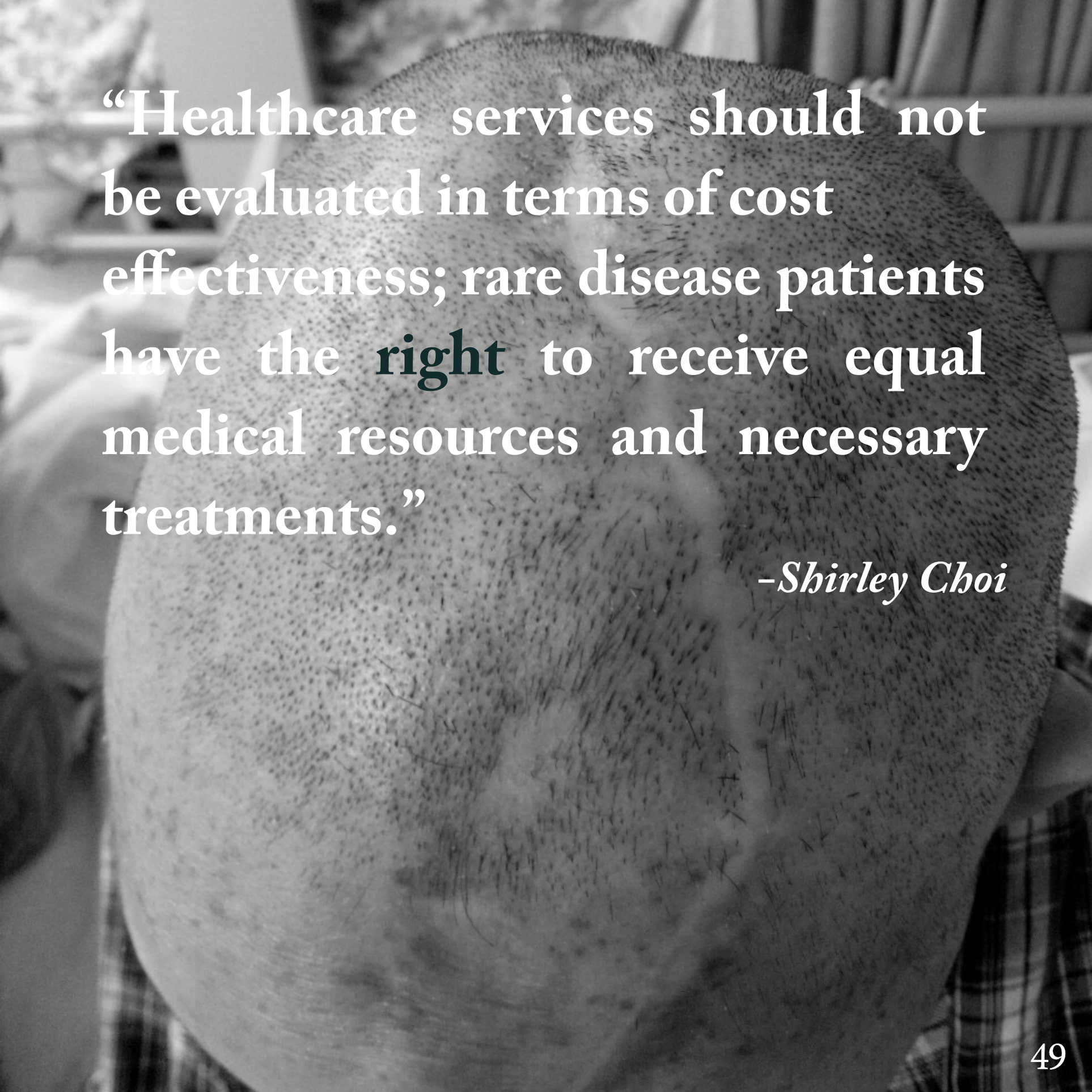
Inheritance: Possible but rare

Prevalence: 3-5 individuals per 100,000

Symptoms: Hardening and tightening of patches of skin, Raynaud's disease (blue and numb fingers or toes in response to cold temperatures)

Effects: Affects their physical appearance, and damages internal organ functions, such as lungs, intestines and esophagus

Treatment: Chronic disease that lasts for a lifetime; supportive treatment: immunosuppressing drugs, corticosteroids



“Healthcare services should not be evaluated in terms of cost effectiveness; rare disease patients have the right to receive equal medical resources and necessary treatments.”

-Shirley Choi

“I made it through again and again; God’s grace is indeed sufficient. I hope someday I can find another scleroderma patient, so that we can fight hand in hand against it.”

OVERCOMING HARDSHIPS

Having to bear with this condition for more than twenty years, Shirley still has not managed to find another person that shares the same disease as hers. This disease has been negatively affecting her daily life in terms of job seeking, social interaction and even romantic relationships. Due to the fact that scleroderma is incurable and the affected skin and follicles cannot turn back to normal, she felt as if she suffered from an incurable deadly disease. In an attempt to keep an appearance that looks ordinary, she has undergone quite a lot of plastic surgeries, but at the same time, she has experienced countless discrimination and taunts. Apart from that, she received chemotherapy because of a kind of rare cancer of the lymphatic system – Burkitt’s lymphoma, which is again a deadly autoimmune disease. One morning in 2018, she had a haemorrhagic stroke and nearly lost her life because of the uncommon cavernous malformation in her brainstem. When all of you readers are wondering how she managed to go through all these misfortunes, she has made it, again and again.


Shirley
Choi

Josy Chow

SMA Patient

Spinal Muscular Atrophy (SMA)

Cause: Mutation of the gene SMN1

Inheritance: Autosomal recessive condition

Prevalence: Occurs 1 in 6000 to one in 10000 children; it is the second most common autosomal recessive disease in humans following cystic fibrosis

Symptoms: Muscle weakness due to death of motor neurons; patients experience difficulty in swallowing and breathing

Effects: Might require the use of wheelchair and respirator in severe case

Treatment: In Hong Kong, the Hospital Authority provides Nusinersen treatment to certain SMA patients; furthermore, new drugs to treat SMA are developed overseas

“If I need to choose one thing to describe myself, I am a cat with nine lives, nothing can kill me or put me down.” – Josy Chow

"Small improvements in their conditions might mean nothing in normal people's eyes, but it might be one that can greatly improve their quality of life."

YOU NEVER KNOW UNTIL YOU TRY

Josy's goal is to become a writer. Since young, Josy has spent a great deal of time in and out of the hospital receiving different kinds of treatment. Nonetheless, her passion in writing is not something that can be taken away easily, and despite not being able to type and write like all of us do, she still believes in the power of education and insisted on studying. She proved to all of us that nothing is impossible when she managed to type out a 22-page proposal to the government calling for life-saving muscle medication using only two fingers. She said frankly that it took her a lot of effort and time, but she did not give up and persevered throughout. Good news came and the proposal was approved by the government and she was able to fight for the providence of the drug for SMA patients. With the drug, her quality of life is improved in terms of being able to swallow some food orally, being able to type on her own and decreased its reliance on ventilators, and the drug also benefited those who are suffering from the same condition as her.

Kwan Tsz Ki

Wilson's Disease Patient

Wilson's Disease

Cause: Mutation on chromosome 13 causes change in copper metabolism, resulting in accumulation of copper in different organs

Inheritance: Autosomal recessive disorder

Prevalence: About 211 patients in Hong Kong

Symptoms: Chronic hepatitis, cirrhosis or even liver failure may occur; neurologically, patients suffer from slurred speech, dystonia, etc.; psychiatrically, mood swings or bipolar disorder may occur; there would also be a greenish brown ring around the cornea

Effects: Slow movement, difficulty in swallowing, learning and speaking abilities

Treatment: Copper chelating drugs taken lifelong, and control of copper ingestion from diet

"I hope that doctors will remember their original intentions and practice medicine with the mentality of treating people instead of treating the disease." – Tsz Ki

“What patients need most is not informative information, but **emotional support**.”

ESTABLISHING A PATIENT ORGANIZATION

Looking back at his early stage of diagnosis, Tsz Ki recalled that he felt very helpless, mainly because of the lack of psychological support. “I was mentally tortured at that time. I couldn’t face myself, my family and friends.” After personally overcoming the helplessness, hesitation and despair, he founded the “Hong Kong Wilson’s Disease Association” with the help of his academic background in biochemistry. Tsz Ki wishes to provide emotional support to all patients by sharing his own experiences, “I hope I can encourage other patients and bring out their confidence, ensuring that they will get better, just like I did.”

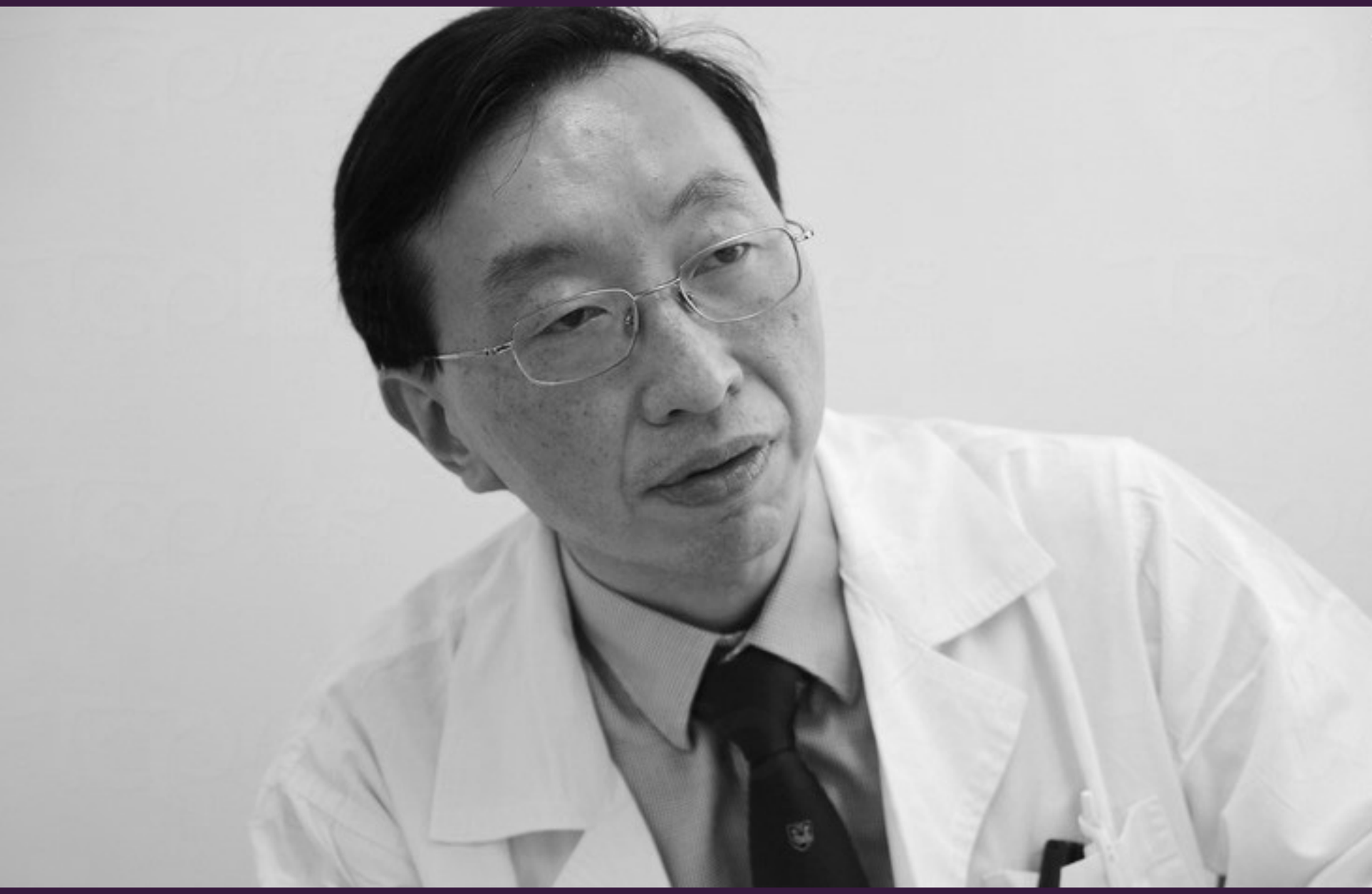
William Chu

6th year HKU Medical Student

“As medical students, we should regard ourselves as **companions** of rare disease patients - we are not here to help them, but to accompany them.”

DEEPLY TOUCHED

William is a Year 6 medical student studying at the University of Hong Kong. He has been involved in rare disease-related work and contributing to the rare disease community for the past 3 years. He and his classmates set out to compile a ‘Patient Registry’, which aims to record different information about rare disease patients, such as their quality of life, the medical expenses, and social resources needed. In addition, they also participated in different rare disease summit forums and international conferences to firstly, broaden their horizons and enhance their understanding of rare diseases; secondly, establish connections between Hong Kong rare diseases experts all over the world so as to strengthen international relations; and thirdly, simplify complicated academic information into digestible notes to enhance patients’ understandings.



Dr. Sheng Bun

Consultant physician of Hospital Authority and specialist in neurology, geriatrics, and internal medicine

“As a doctor, you need to be where your patients are.”

A DIFFERENT APPROACH

Although Hong Kong’s medical system is divided into a lot of distinct specialties, this might not be to the benefit of rare disease patients and their doctors. Since rare disease patients are dispersed throughout the healthcare system, doctors seldom have the chance to meet them and hence lack understanding of these disorders. Dr. Sheng discovered the importance of a centralized approach towards treating rare diseases in one of his trials when he organized a gathering for familial amyloidotic polyneuropathy patients referred to his clinic. From that gathering, he learnt what rare disease patients sought the most is support from peers with the same experience.

Dr. Sheng believes centralizing rare disease treatment benefits both patients and doctors. While patients can receive more support from peers and learn more about their disease, centralized treatment also enables doctors to enrich their knowledge and understanding on rare diseases. Having a team specializing in rare diseases allows accumulation of knowledge, experience and training, while benefiting clinical research into developing new treatments as well.

Dr. Brian Chung

Clinical Associate Professor,
Department of Paediatrics and Adolescent Medicine,
LKS Faculty of Medicine, The University of Hong Kong

“Finally after 10 years, I was able to diagnose one of my rare disease patients. I no longer owe that patient an answer. That was my happiest and most satisfying moment as a doctor.”

HIS PROUDEST MOMENT

Dr. Brian Chung is currently an associate professor in HKU and one of the first fellows in clinical genetics in Hong Kong. However, it was only after completing his medical degree did Dr. Chung’s passion for rare diseases flourished. In particular, Dr Chung says he was touched by the gratefulness and support this special patient group always shows. The proudest moment for Dr. Chung was when his genomics team led the discovery of a new rare disease — MN1 C-terminal Truncation Syndrome. Not only was it the first time an Asian genomics team led the discovery of a new rare disease; it was also a deeply emotional moment for Dr. Chung, since he could finally give his patient the answer to the question he could not explain for the past 10 years.

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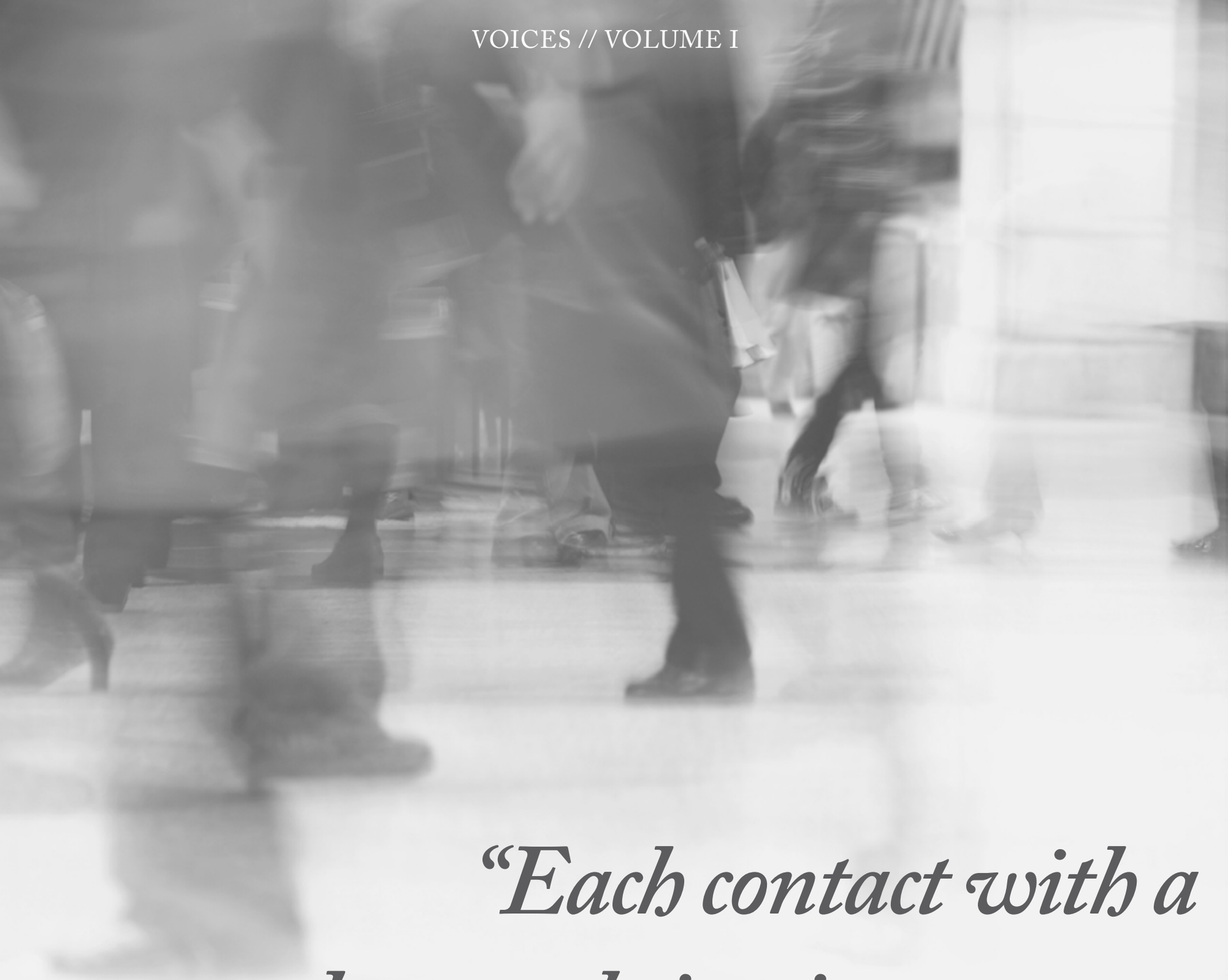
Social Media
Instagram/ @medicaloutreachers
@humansofmedicaloutreachers
Facebook/ @medicaloutreachers

Drawn by Summer Sze, 12th Excecutive Committee of Medical Outreachers



"Each contact with a human being is so rare, so precious, one should preserve it."