

Giving hope to persons with rare diseases

Christopher, a two-year old boy, has an extremely rare condition called bile acid synthesis disorder. There are fewer than 50 reported cases around the world.

His condition was discovered when he was four months old when his parents noticed that a scratch on his nose and a prick from a blood test left his nose and finger bleeding for more than 30 hours. Christopher's eyes and skin were also jaundiced, and he was slow in gaining weight.

For his parents, the pain was immense as they watched Christopher struggle and being bruised from blood tests. They were told that his liver was enlarged and had some scarring.

The rare disease that Christopher has interferes with the production of bile acids such as cholic acid, which help the flow of excretion of bile from the liver and assist in the absorption of fat and fat-soluble vitamins from food. Without early intervention, the toxic bile acids will build up and damage the liver, ultimately causing liver failure and eventually death.

The medicine that Christopher needs costs at least \$7,000 a month. As he will require a higher dose of medicine as he grows older and gains weight, the family worries each day about their ability to continue to afford Christopher's treatment, as he has to be on medication for life.

Much like Christopher, 11-year old Zecia also has a rare disease. She suffers

from Gaucher disease, a rare neurodegenerative condition where her body does not produce an enzyme to get rid of a specific type of fat. This fatty substance can then build up in various organs causing damage.

When Zecia was first diagnosed, her liver and spleen were extremely enlarged and hence her abdomen was severely distended. This affected her ability to eat and her growth was poor. Her father recalled how the disease caused her stomach to become so bloated as a toddler

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that she could not balance well and she kept falling down.

As Gaucher disease was very rare, it took more time to arrive at a definitive diagnosis. The family also had to contend with the high cost of the treatment, more than \$24,000 a month.

Without treatment, her liver and spleen will swell progressively and affect her health and life. Further accumulation of the fatty



Mount Alvernia Hospital, in partnership with Community Chest, will be organising an awareness cum fund raising event regarding rare diseases. Photo: MOUNT ALVERNIA HOSPITAL

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mission to help the marginalised as Singapore's only Catholic and not-for-profit general hospital," said Ms Anthea Neo, Assistant Director, Community Outreach, Mount Alvernia Hospital.

The event will be held on Nov 3 at Punggol Town Square from 8.30am-2pm. Members of the public are welcome.

At the event, participants will have a chance to listen to a sharing on rare diseases. There will be interactive games and booths, performances including a magic show as well as a fitness session.

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The hospital's community outreach medical and dental clinics, located at the Enabling Village at Redhill and Agape Village at Toa Payoh, have also benefitted many underserved communities and marginalised individuals who do not have easy access to regular healthcare services. □

About rare diseases

Rare diseases can affect people of all ages but they are often diagnosed at birth or during early childhood. Without treatment, most conditions can be life-threatening. About one-third of patients with rare diseases globally do not live beyond five years old.

There are no treatments available for most rare diseases. But for a few, effective medicines are available which can replace the missing enzymes or biochemicals. In such cases, with early treatment, patients' lifespans and quality of life will be greatly improved, allowing them to live relatively normal lives.

The medicines however usually have to be taken for the patients' entire lives and can be very expensive (up to hundreds of thousands of dollars a year).

The Rare Disease Fund (RDF) is a charity fund that seeks to provide long-term financial support for patients with rare diseases requiring treatment with high cost medicines. The RDF is managed by KK Women's and Children's Hospital (KKH) Health Fund.

For more information on the event and on rare diseases, visit www.kkh.com.sg/rarediseasefund

Submitted by Mount Alvernia Hospital