An initiative by:

Rare Disorders Society (Singapore)

Mailing address:
43 Hindhede Walk #07-08, Singapore 587973

contact@rdss.org.sg
www.rdss.org.sg

Content partner:

Costello Medical Singapore Pte Ltd

101B Telok Ayer Street, #03-00
Singapore 068574

www.costellomedical.com

Prepared on a pro bono basis by Costello Medical, an independent market access and medical communications agency.

#We do not represent any medical professionals or institutions. The author(s) of this handbook may not be certified medical professionals and hence the information found herein does not constitute formal advice in any manner whatsoever. The handbook is purely a collection of the authors' personal experience and, where references are used, the relevant sources of information are cited accordingly. You are advised to consult your physicians or other healthcare professionals for a formal and accurate diagnosis or assessment of your particular medical, emotional or psychological needs (if any).

Rare Disorders

Patient Handbook

About This Handbook

This handbook is written for the parents of children living with rare diseases in Singapore. In this handbook, you will find the following:

Rare Disease Overview

This section introduces you to facts about rare diseases, including some of the possible causes of rare diseases, and treatment options that may be available for some rare diseases.

Patient Stories*

Read the inspiring stories of our beneficiaries: Pei Shan, Vera, Issac, and Emily.

Caregiver Toolkit

Some useful tips relating to the following areas are shared here:

- Caring for your child with a rare disease as an empowered caregiver
- Parenting your child with a rare disease
- Personal well-being
- Your partner's well-being
- Your other child's well-being
- Planning for your next child

Useful Resources

Find the latest public and private resources in Singapore that offer emotional, medical, financial and educational support to rare disease patients and their families.

*The stories of Pei Shan, Vera and Issac were written based on previously published newspaper articles as well as other online sources, such as blog posts and patient stories available on the Rare Disorders Society (Singapore) website. Emily's story was written in her own words. All stories were reviewed and approved by the patients and/or their parents prior to the publication of this handbook.

Rare Disease Overview

What is a Rare Disease?

There are thought to be between

5,000 and 8,000

rare diseases worldwide.[1]

The definition of a rare disease differs from region to region

In the EU,

a disease is considered rare if it affects fewer than

1 in 2,000

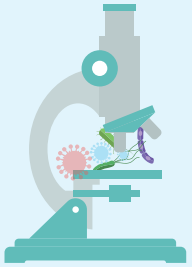
individuals.[2]

In Japan,

a disease is considered rare if it affects fewer than

50,000

individuals.[2]



While the number of people living with rare diseases in each country is low, there are approximately

400 million people in the world currently living with rare diseases.[1]

Rare disease patients often experience delays in receiving the correct diagnosis. According to a national survey in the UK, a rare disease patient typically consults **5 doctors**, receives **3 misdiagnoses**, and waits for **4 years** before being correctly diagnosed.[4]

In Singapore,

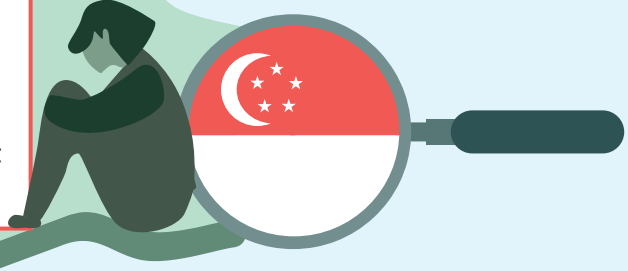
a rare disease is defined as a life-threatening and severely debilitating illness affecting fewer than

20,000 patients

in its population of 5.5 million.[3]

60.7%

of rare disease patients face issues before getting the right diagnosis.[5]

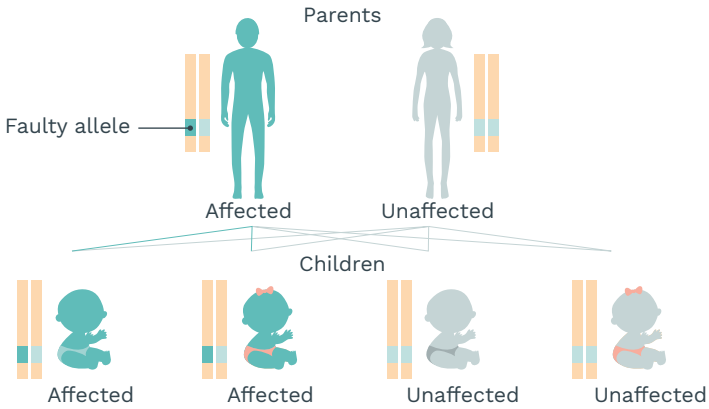


What Can Cause a Rare Disease?

80%

of rare diseases are caused by faulty genes. The rest can result from bacterial or viral infections, allergies or environmental causes.[1]

Inheritance of the faulty gene(s) follows one of these patterns:

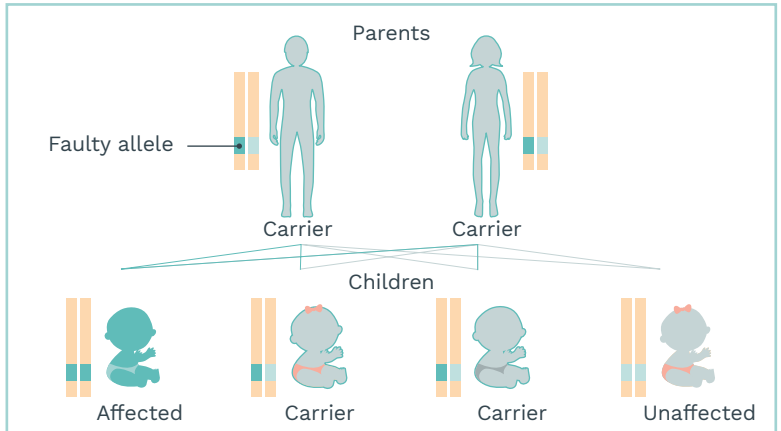


Autosomal Dominant: If one parent carries a dominant copy of the faulty gene, the child will have a 50% chance of developing the disease.

Examples: Huntington's disease, autosomal dominant hereditary ataxias

Genes carry the information that determines your traits. Humans have two copies, or alleles, of each gene. These alleles can either be dominant or recessive.

- **Dominant alleles** show their effect even when there is only one copy of the faulty allele.
- **Recessive alleles** only show their effect when both alleles are faulty.



Autosomal Recessive: If both parents carry a recessive copy of the faulty gene, the child will have a 25% chance of inheriting both copies and developing the disease.

Examples: Cystic fibrosis, Pompe disease

Sex-linked Inheritance

Some rare diseases are sex-linked, meaning that your chances of inheriting these conditions depend on whether you are a boy or a girl.

Examples: Hunter syndrome, Duchenne muscular dystrophy (DMD)

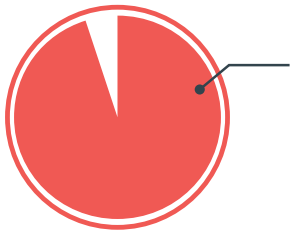
Chromosome Abnormalities

Most chromosome abnormalities are **not inherited** from the parents. Changes in the number of chromosomes occur as random events during the formation of eggs and sperm, and as a result the child may have an extra or missing chromosome.

Examples: Trisomy 18 (a.k.a. Edwards syndrome), Trisomy 21 (a.k.a. Down syndrome)

Can Rare Diseases be Treated? What are the Treatment Options?

Most rare diseases can have their symptoms managed in some way, but the vast majority do not have disease-modifying treatments available.

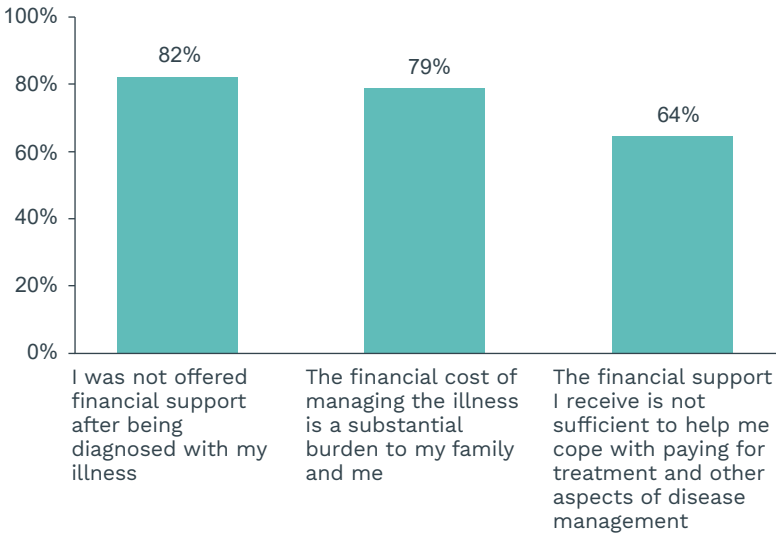


95%

of rare diseases currently have no treatments approved by the Food and Drug Administration (FDA) in the US.[1]

In Singapore, patients, including rare disease patients, have access to many FDA-approved drugs. Treatments for rare diseases, however, are typically more expensive than treatments for more common conditions.[6] For instance, Myozyme® (alglucosidase alfa), an enzyme replacement therapy, is available for patients with Pompe disease, but comes at a cost of up to USD 300,000* a year.[7]

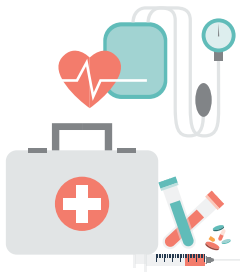
Financial Burden of Rare Disease Patients and Their Caregivers in Singapore: The Singapore Rare Disease Impact Study (n=152)[8,9]



Treating the Symptoms

- Although there is a limited number of medications targeting the cause(s) of rare conditions, there are various medical therapies to relieve symptoms.
- For example, if your child has a metabolic disease that makes them unable to digest certain types of nutrients, a dietician in a hospital may help design a special diet suitable for their condition.

Clinical Trials



- Clinical trials test the safety and efficacy of new treatments in patients with specific conditions.
- If you would like your child to receive treatment through a clinical trial, you should first consult their attending doctor(s). The doctor(s) may discuss with you the effectiveness of currently available treatments, the duration of the trial, the accessibility of the trial location, and the potential risks involved.[10]

*The cost of treatment was calculated based on the recommended dosage of Myozyme® in 20 mg/kg body weight, administered every 2 weeks as an intravenous infusion as per the product information, using the list price of Myozyme® 50 mg/vial in Singapore.[11]

Patient Stories

Living Life to the Fullest

Teo Pei Shan

A teenage girl in a toddler's body, Pei Shan had symptoms that were too rare to be classified as any previously identified rare diseases, though her official medical record reads Mucopolysaccharide disease (a.k.a. Sanfilippo syndrome).[12]

Throughout the years Pei Shan was affected by various medical conditions, such as restrictive lung disease (restricted lung expansion which results in extra force required to breathe), low potassium levels, and glaucoma (damage to the optic nerve caused by increased fluid pressure in the eye).

Despite her conditions, Pei Shan never stopped pursuing her artistic interests. She found joy in drawing, spreading love and smiles to others through her art. In 2012, her first canvas piece was featured on the front cover of Club Rainbow (Singapore)'s commemorative hardcover book "Colour of Life, Celebrating 20 Years of Club Rainbow".

A talented pianist, Pei Shan enrolled in the Beautiful Mind Music Academy, a free music programme offered by Beautiful Mind Charity for potential artists with disabilities to pursue a musical career. In 2014, Pei Shan realised her dream of playing a grand piano for a large audience when she performed in front of more than 500 people at the Beautiful Mind Music Academy Annual Concert at School of the Arts Singapore (SOTA). Subsequently, she performed in their annual concerts in 2015 and 2016. Pei Shan also performed live on TV during the Serving People with Disabilities (SPD) Charity Show in 2015.

Pei Shan passed away in her parents' arms on 19th July 2016, two weeks before her 18th birthday. While her life may have been short, Pei Shan lived it to the fullest, and her positive outlook on life continues to inspire others today.

Help your children live a fulfilling life by encouraging them to pursue their passion.

Family, Where Life Begins and Love Never Ends

Vera Lau

Shortly after birth, Vera was diagnosed with Trisomy 18 (Edwards syndrome), a rare genetic disorder where an extra 18th chromosome causes severe mental, physical, and developmental delays, as well as multiple medical issues.[13]

Despite the fact that only 1 out of 10 infants with this condition survives more than a year, Vera has thrived with the full support of her family, caregivers, teachers and doctors.

Throughout the years, Vera's parents have always shared the responsibilities of caregiving. In Vera's first year, her father took 9 months of no-pay leave to be a full-time caregiver, so that her mother could return to work at the end of maternity leave. Later, in 2016, due to increased secretions caused by decreased lung function and swallowing ability, Vera started to require suctioning 2–3 times a day. During these suctioning sessions, her father would hold her still while her mother performed the procedure.

Vera's parents make a point for little brother Daen to learn to take care of her—by giving him more love so he can pass it on to his sister. Daen has now learned to tube feed Vera and push her around in her wheelchair.

The outpouring love and support from their extended family - looking after Daen, bringing food and nourishment, and staying through the night during Vera's darkest days - has been crucial in getting them through times of difficulty.

Vera is turning 10 years old in 2018. A medical miracle, she continues to persevere in her battle. While unable to speak, Vera shows abundant and unconditional love: she responds happily to requests for hugs and can identify loved ones by their names. Vera's parents see her as a blessing in life, and treasure every moment they spend together.

Successfully looking after a child with multiple medical needs requires the support of committed parents, caregivers, as well as a support network for caregivers.

We Rise by Lifting Others

Issac Tan

Issac was diagnosed with Rubinstein–Taybi syndrome (RTS) at birth, which affects the critical stages of early development. As a result, he suffers from gastrointestinal issues, heart and facial defects, as well as delays in speech and language development.[14]

Patients with RTS require an interdisciplinary treatment approach to improve their physical and mental well-being. Issac's family ran into financial difficulties as his mother had to give up her job to become a full-time caregiver.

Issac was referred to Rare Disorders Society (Singapore) and, through the society, he has received financial support for his treatments, including various forms of therapies. Over the years, his communication skills have improved the most and he now socialises freely with other children during the society's outings.

Issac's family is grateful that he has entered a programme at SPD's Building Bridges Early Intervention Programme for Infants and Children (EIPIC) centre, which is designed to maximise the developmental growth potential of children with special needs. A sponsorship from a private organisation has also allowed Issac to attend therapy sessions at the Young Women's Christian Association (YWCA) of Singapore for 2 years.

Slowly but surely, Issac's condition is improving, and he now enjoys exploring his surroundings and playing with his peers like any other boy. At the age of 7, Issac is currently studying at the Assocation for Persons with Special Needs (ASPN) Chaoyang School and continues to make progress in developing his social and life skills.

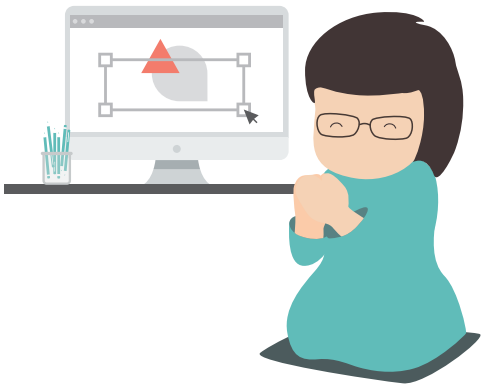
Recently, the family welcomed a new member and Issac is now a proud brother! As Isaac has learned to be more independent, his mother has also been able to resume working.

If you are experiencing financial difficulties because of your child's condition, you may seek assistance from various public and private schemes.

Joining a patient support group could be beneficial for you and your child.

Faith, My Pillar of Strength

Emily Ho



Emily Ho shares her ongoing journey of living with Pompe Disease.

I was diagnosed with juvenile onset of Pompe Disease when I was 14. It's a rare disease which affects and weakens the muscles in my entire body, including the respiratory system. There is no cure for this condition, and I can only receive a lifelong treatment to slow down the deterioration.

Prior to the diagnosis, I used to fall down quite often and could not catch up with my peers in physical fitness. I slowly began to struggle with my mobility, and eventually had to start using an elbow crutch to get around. I also get breathless from time to time, so I use a bi-pap machine to keep my airway open when I sleep at night.

Living with these conditions is not easy, but I try not to let these circumstances define who I am. It was my faith in God and His love that helped me pull through the initial years of struggle.

With the love and support of my family and friends, I graduated with a diploma in 2013 and pursued my dream of becoming a full-time graphic designer in my church. It gives me a huge sense of fulfilment as I get to contribute to my church and serve God with the talent that He has blessed me with. My bosses and colleagues are very supportive and understanding, which I am also very thankful for.

By God's grace and with the help of my doctors and social worker, I was finally able to receive treatment for my condition. I believe this entire journey is helping me to grow. People often comment on my cheerfulness and positive attitude, and I would like to say the joy of the Lord is my strength!

If you are going through an emotional upheaval but do not know how to deal with it, you may find comfort in spiritual or religious support.

Caregiver Toolkit

Learning that your child has a rare disease can be devastating, and you may feel overwhelmed by the medical, financial and emotional stress of taking care of your child.

This toolkit shares tips on how to maintain the well-being of your child with a rare disease, yourself and other family members, including your partner and other child(ren).

Caring for Your Child with a Rare Disease as an Empowered Caregiver

Being prepared and informed can empower you as a caregiver. The following suggestions aim to help you feel more confident and less stressed as a caregiver.[15]

- **List the questions you would like to ask relevant medical professionals before a consultation.** Be as specific as possible when describing your child's medical condition. Express your doubts honestly.
- **Prepare a complete record of all your child's medications for the doctor's reference.** Knowing the mechanisms and side-effects of your child's medications is critical in emergency situations.
- **Make your own notes during medical consultations** either in writing or through voice recordings.
- **Be your own medical research advocate.** If your child's condition is rare, the doctor may not have answers to all your questions. You can look up the latest findings about your child's condition on rare disease-specific databases such as Raremark and RareOmics.
- **Consolidate your child's medical records, dietary information, and your own notes in a care handbook.** This helps to provide you with a good overview of your child's condition and development.
- **Join a patient support group.** There are patient support groups in Singapore that offer various means of caregiver support, including subsidised online training courses for caregivers.
- **Prepare for unexpected medical emergencies.** Having at hand a portable medical kit with emergency medications, a visible identification card with information on key medical conditions and emergency contacts, special foods or formulas and extra diapers will help you and your child stay calm. Come up with an emergency plan with the whole family, so that everyone knows what to do. Learn life-saving skills such as basic cardiac life support (BCLS) and ensure life-saving equipment is within your reach when you need it.
- **Ensure good maintenance of your child's medical equipment.** If your child requires medical or mobility equipment at home, periodic maintenance of all the equipment, ideally once or twice a year, is highly recommended.
- **Be your child's therapist.** Regular physio-, speech and language, and occupational therapy sessions will improve your child's overall condition. Learn various essential exercises from the therapists during these sessions and come up with a routine for your child to follow. Consistent and regular therapy interventions can prevent deterioration or even improve your child's condition(s).
- **Manage your expectations.** Be realistic about your child's development. Discuss and set milestones together with your child's doctor(s), therapist(s), teacher(s), and anyone who has a part to play in the caring, development, and learning of your child. Try not to compare your child with others. Focus on your child's achievements and the present challenge(s). Do not be disheartened by what they cannot do and extend such pessimism into the future.
- **Engage home-care services if you need to manage your child's medical needs from home.** Palliative care services can also help to improve the quality of life of your child and yourself. Home-care services can be arranged via hospitals or home care support. These services also reduce the burden of frequent hospital visits.

Parenting Your Child with a Rare Disease

This section provides parenting dos and don'ts and how to offer support and encouragement to your child without being overly protective or permissive.[16]

Do view your child's behaviours as signals of needs. If you are unable to interpret certain behaviours, record them and consult your childs' doctor(s) or therapist(s).

Do communicate with your child. If your child struggles to express themselves verbally, encourage them to communicate their thoughts and feelings through pictures, music, and assistive devices such as a picture board with pictures or symbols of items and activities that make up your child's daily life, or a computer programme that synthesises speech from simple text.

Do encourage your child to interact with others. Early intervention programmes or special education schools are good avenues of therapy and education, and they also create opportunities for your child to socialise with people beyond the family nucleus.

Do help your child find and pursue feasible interests such as sports programmes for children with special needs, drawing, or music. This can help improve their motor skills, mental and emotional well-being, as well as create a sense of achievement.

Do celebrate every little milestone your child achieves and set goals that are realistic. Focus on your child's achievements rather than their disabilities.

Don't compare your child to other children. Every child is special in their own way. Accept your child's physical and mental disabilities.

Don't put your child's health at risk. As much as you hope to see improvement(s) in your child's condition or achievements, you need to consider their health first. Activities that might cause health issues should be avoided.

Don't fall into the "different = fragile" trap. It's important to teach your child (at an early stage) what behaviour is appropriate or acceptable. You can start with teaching your child simple routines. This will help your child develop a sense of responsibility and discipline, making it easier for them to assimilate into society.

Don't get too emotional and pessimistic in front of your child (regularly). You want your child to be cheerful and have a positive outlook on life. Being emotional in their presence may make them feel like they are a burden to the family.

Personal Well-Being

While taking care of a child with a rare disease presents a unique set of challenges, it is also important to look after your own well-being.[17] Those who take care of themselves have the energy to take care of others joyfully because caregiving doesn't come at their own expense.

Get support from your spouse, relatives, friends, and patient support groups. Support from family and friends makes caregiving less stressful and more manageable. There are several patient support groups in Singapore that provide emotional support and an excellent opportunity to network with parents who are on the caregiving journey.

Take care of your physical health. Caregivers are often sleep-deprived, which can cause illness and stress. Make sure you have routine health check-ups, exercise regularly, and eat a balanced diet.

Take care of your mental health. If you feel trapped and helpless about your situation but do not know how to deal with your emotions, seek professional help immediately. You may also consider seeking spiritual or religious support.

Spend some time on personal grooming. When one looks good, one feels good. It need not be an extensive make-over; it can simply be a hair wash, a perm or a facial, depending on how much time you have. When you are in a good mood, you can care for your child better. Remember, your emotions rub off on your child.

Learn to identify, accept, and manage your stress. Perceive situations with optimism: there are often more solutions than problems. Don't neglect any unresolved issues, as they will simply add to your stress.

Set up simple self-care goals to accomplish in the next 3 to 6 months. These goals may be meeting friends for dinner or as simple as going for a short walk 3 times a week.

Take a break from caregiving. There are respite programmes that provide short-term caregiving services or organise outings for caregivers and their children. A break does not need to be extensive in terms of duration or intensity. It can be as simple as reading your favourite book, listening to your favourite song or sipping your favourite drink. If there is sufficient or appropriate support, you can consider stepping out for a gathering or going for a holiday. In some cases, meditation helps calm your emotions and gives you mental strength to resume your duties.

Your Partner's Well-Being

While living with a child with a rare disease poses challenges to your family, the shared experience can also bring the family closer.[18] This section shares tips on preserving family relationships in the journey of raising a child with a rare disease.[19,20]

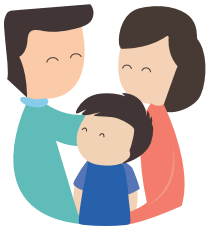
Acknowledge that you and your partner may face different challenges. For example, mothers often struggle with being the primary caregiver, while fathers tend to feel isolated and bear the financial burden. You and your partner may also experience different emotions in the same situation. After all, "women are from Venus and men are from Mars"!

Maintaining good communication is vital to a strong relationship. As a starting point, you may consider writing a shared journal or attending couples therapy. Speak freely and discuss concerns amicably, bearing in mind that it is for a common objective and for the greater good of the family.

Get help from your family, friends and patient support groups. Networking with parents of children living with rare diseases can help relieve the stress of caregiving. It helps to open your mind and heart to various possibilities and look at issues or challenges from a different perspective.

Your Other Child's Well-Being

Siblings of children with rare diseases often experience mixed feelings of guilt, grief, confusion, anger, and jealousy. It is essential to detect and manage such emotions early before they develop into bigger issues.



Communicate with your other child regularly and help your child build a network of support. This ensures that your child's needs are met and that their emotions are kept in check.

Make your child feel special and let them know that you care for them:[21]

- Physical affection confers your child emotional assurance. Simple gestures like holding their hand, hugging or giving them a goodnight or goodbye kiss are ways to show your love for your child.
- Shower them with praises for their accomplishments. Give your child consistent recognition by saying "well done", "good job" or "I am proud of you" for displaying the right behaviour. It not only helps reinforce the correct behaviour, but also signifies that you are paying attention to your child.
- Be in tune with your child's hobbies, education and extra-curricular activities. You may make small talk with your child on topics relating to any of the above when you are dropping them off at school or at bedtime.
- Make a point to specifically set aside dedicated time and attention to be spent with your child alone. For instance, schedule a regular "special play time" to play their favourite game. Let them take the lead and you can play along. It could also be a weekly outing (e.g. movie, swimming, etc.) where only you and your child spend some quality time with each other.
- Treat your child as an equal member of the family by keeping them well-informed about the medical conditions of their affected sibling. Explain what tasks need to be done first and how everyone can play a part. Teach them how special their sibling is and how he/she can be proud of their special family member.

Planning for Your Next Child

For parents of children with rare diseases, family planning can be complex. If you and your partner are planning to have another child, it is important to be aware of the chances that your next child will have a rare disease, so that you can make informed decisions.

- **Attend genetic counselling prior to conception**. Genetic counselling helps translate complex scientific information about the risks of having children with inherited rare diseases, and provides information about the options available.[22]

  – During counselling, the counsellor would ask you about your family health history and any harmful substances or medications you have been exposed to. You may also take this opportunity to address any doubts about family planning. At the end of counselling, the counsellor may refer you to other hospitals for appropriate testing upon your request.

  – You and your partner may wish to undergo blood or saliva testing to determine if you are carriers of any faulty genes that could be inherited by your next child (please refer to the Rare Disease Overview section for the definition of a carrier).

- **Receive prenatal screening during the early phase of pregnancy**. Prenatal screening allows the detection of genetic abnormalities as early as possible.

  – Discuss with your gynecologist or genetic counsellor the types of prenatal tests you could take at different phases of pregnancy, the kind of rare diseases that can be detected by each test, and any risk of miscarriage associated with invasive tests such as chorionic villus sampling and amniocentesis.[23]

Useful Resources

Rare Disorders Society (Singapore)

RDSS is a non-profit organisation that was first established by parents of children with Lysosomal Storage Disorder in 2011.

RDSS is currently the only organisation in Singapore that supports all patients with diagnosed or undiagnosed rare diseases and their families through various means of support: raising public awareness on rare diseases, organising support group sessions, providing financial support for sustainable treatment methods, and participating in rare diseases conferences in the APAC region.

We encourage you to visit the RDSS website (http://www.rdss.org.sg) for the latest public and private resources that provide medical, financial, and emotional support for patients living with rare diseases in Singapore, as well as their families and friends.

Notes to Self

1

2

3

Notes to Self

4

5

6

References

1. de Vrueh R, Baekelandt ERF, de Haan JMH. Priority Medicines for Europe and the World: Update on 2004 Background Paper, BP 6.19. Rare Diseases. World Health Organization 2013.
2. Richter T, Nestler-Parr S, Babela R, et al. Rare disease terminology and definitions–a systematic global review: report of the ISPOR rare disease special interest group. Value in Health 2015;18:906-914.
3. Medicines (Orphan Drugs) (Exemption) Order 1991. In: Edition) ER, ed. Singapore: Attorney-General's Chambers Singapore, 1991.
4. Rare Disease UK. The Rare Reality-an insight into the patient and family experience of rare disease, 2016. Available at: http://www.rarediseases.org.uk/media/2361/patient-experiences-2015.pdf
5. Loh CJW, Ong CD. ASEAN+ Rare Disease Network: Unifying Voices of Patients. Asia-Pacific Biotech News. Singapore, 2017. Available at: https://www.asiabiotech.com/21/2106/21060014x.html
6. Amiculum. World Orphan Drug Congress Asia 2014, 2014. Available at: https://www.amiculum.biz/media/11019/world_orphan_drug_congress_asia_2014_amiculum_snapshot.pdf
7. Rare Disorders Society Singapore. Patient Stories, 2014. Available at: http://www.rdss.org.sg/story-view/chloe-mah/
8. Rainbow Across Borders. Hearts, Hopes and Aims: The Singapore Rare Disease Impact Study, 2017. Available at: http://rabasia.org/publications/hearts-hopes-and-aims-the-singapore-rare-disease-impact-study
9. ASEAN+ Rare Disease Network Established. Asia-Pacific Biotech News. Singapore. 2017. Available at: https://biotechin.asia/2017/03/24/asean-rare-disease-network-established/
10. Rare Toolkits. From Molecules to Medicine: Clinical Research, 2016.
11. MYOZYME® (alglucosidase alfa). [package insert]. Genzyme., Massachusetts, United States, 2016. Available at: https://www.accessdata.fda.gov/drugsatfda_docs/label/2014/125141s219lbl.pdf
12. Fedele AO. Sanfilippo syndrome: causes, consequences, and treatments. The application of clinical genetics 2015;8:269.
13. Rosa RFM, Rosa RCM, Zen PRG, et al. Trisomy 18: review of the clinical, etiologic, prognostic, and ethical aspects. Revista paulista de pediatria 2013;31:111-120.
14. Hennekam, RCM. Rubinstein–Taybi syndrome. European Journal of Human Genetics 2006;14:981.
15. Rare Toolkits. Becoming An Empowered Patient: A Toolkit for The Undiagnosed, 2014. Available at: https://globalgenes.org/toolkits/
16. Ask Dr Sears. Disciplining the Special-Needs Child. Available at: https://www.askdrsears.com/topics/parenting/discipline-behavior/disciplining-special-needs-child
17. Family Caregiver Alliance. Taking Care of YOU: Self-Care for Family Caregivers, 2012. Available at: https://www.caregiver.org/taking-care-you-self-care-family-caregivers
18. Anderson M, Elliott EJ, Zurynski YA. Australian families living with rare disease: experiences of diagnosis, health services use and needs for psychosocial support. Orphanet J Rare Dis 2013;8:22.
19. Rare Toolkits Parenting A Child with Life-Limiting Illness, 2014. Available at: https://globalgenes.org/toolkits/
20. Raiees-Dana D. Diamonds or Dust: Keeping Your Marriage Together When Your Child Fights For Life: Arkansas Children's Hospital, 2012.
21. Siblings Support Project. Available at: https://www.siblingsupport.org/
22. Resta R, Biesecker BB, Bennett RL, et al. A New Definition of Genetic Counseling: National Society of Genetic Counselors' Task Force Report. Journal of Genetic Counseling 2006; 15:2.
23. Akolekar R, Beta J, Picciarelli G et al. Procedure-related risk of miscarriage following amniocentesis and chorionic villus sampling: a systematic review and meta-analysis. Ultrasound Obstet Gynecol 2015.45.