KK Women's and Children's Hospital
SingHealth

IN THIS ISSUE

RAPID GENOMIC SEQUENCING FOR CRITICALLY-ILL CHILDREN

At KKH, rapid genomic sequencing is accelerating genetic testing for critically-ill children who are suspected to have a genetic disorder - enabling timely clinical care and management, and providing families the opportunity to find answers.

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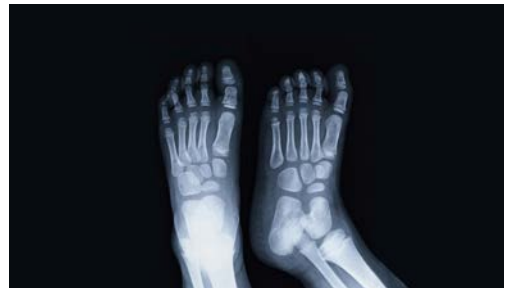
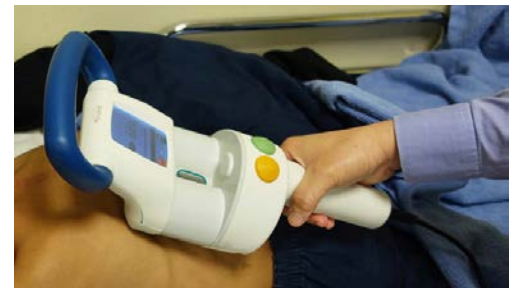
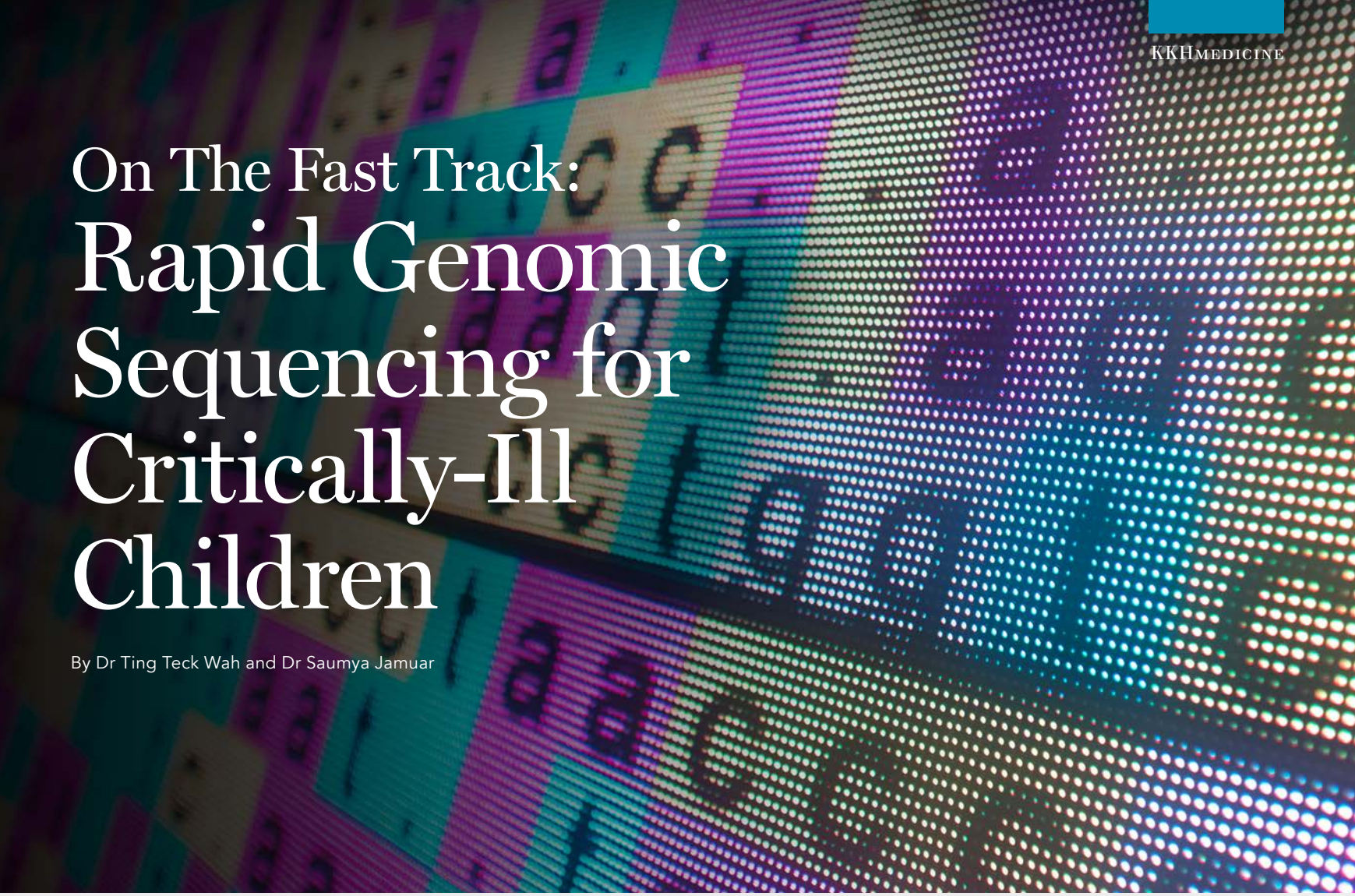
CONTENTS

News from Singapore's Academic Tertiary Hospital For Women And Children







On The Fast Track: Rapid Genomic Sequencing for Critically-Ill Children

By Dr Ting Teck Wah and Dr Saumya Jamuar

Globally, there are more than 7,000 known rare diseases, of which 80 per cent have an underlying genetic aetiology[1]. Although individually rare, affecting fewer than one in 2,000 individuals, birth defects – a subset of rare diseases – are reported in approximately 2.4 per cent of live births in Singapore[2].

In addition, about 50 per cent of rare diseases present in childhood, and up to one in three children admitted to the paediatric intensive care unit (ICU) is suspected to have an underlying genetic disease[3].

The diagnostic odyssey for most patients with rare diseases can be lengthy, and often include multiple evaluations by different healthcare providers, without a definitive diagnosis.

On average, it is estimated that a patient with a rare disease will see up to eight different specialists, and wait about 7.6 years to achieve a diagnosis. About 30 per cent of children with rare diseases die by the age of five years, some without ever receiving a correct diagnosis.

In recent years, the increasing application of clinical exome sequencing in the paediatric ICU setting has been gaining traction, where a short turnaround time (usually within two weeks) coupled with a relatively high diagnostic yield (30% to 40%) allows for significant changes in the medical management of patients with rare diseases[4,5], and, at the same time, provides patients and their families an opportunity to get answers.

A GENETIC DIAGNOSIS IN 40 PER CENT OF RAPIDSEQ CASES

RapidSeq (Rapid Genomic Sequencing test) was launched at KK Women's and Children's Hospital (KKH) in April 2018, with the aim to provide a genetic diagnosis within two weeks for critically-ill patients in the Neonatal Intensive Care Unit (NICU) and Children's Intensive Care Unit (CICU) who are suspected to have a genetic disorder. In comparison, a waiting period of three months is usual using traditional methods of genetic testing.

From April 2018 to January 2019, 10 cases were enrolled into RapidSeq, with the youngest patient being just three days old. Seven were patients from NICU, two were patients from CICU, and one was a stored fetal DNA sample.

In four of the 10 cases (40%) a genetic diagnosis was achieved. For three of these, the diagnosis led to changes being implemented in the patient's clinical management plan. The median turnaround time for results was nine working days, with six days being the shortest, and 12 days the longest.

We highlight two cases where the RapidSeq diagnosis directly impacted the management of the patient, and aided counselling for the family.

Continued on page 4...

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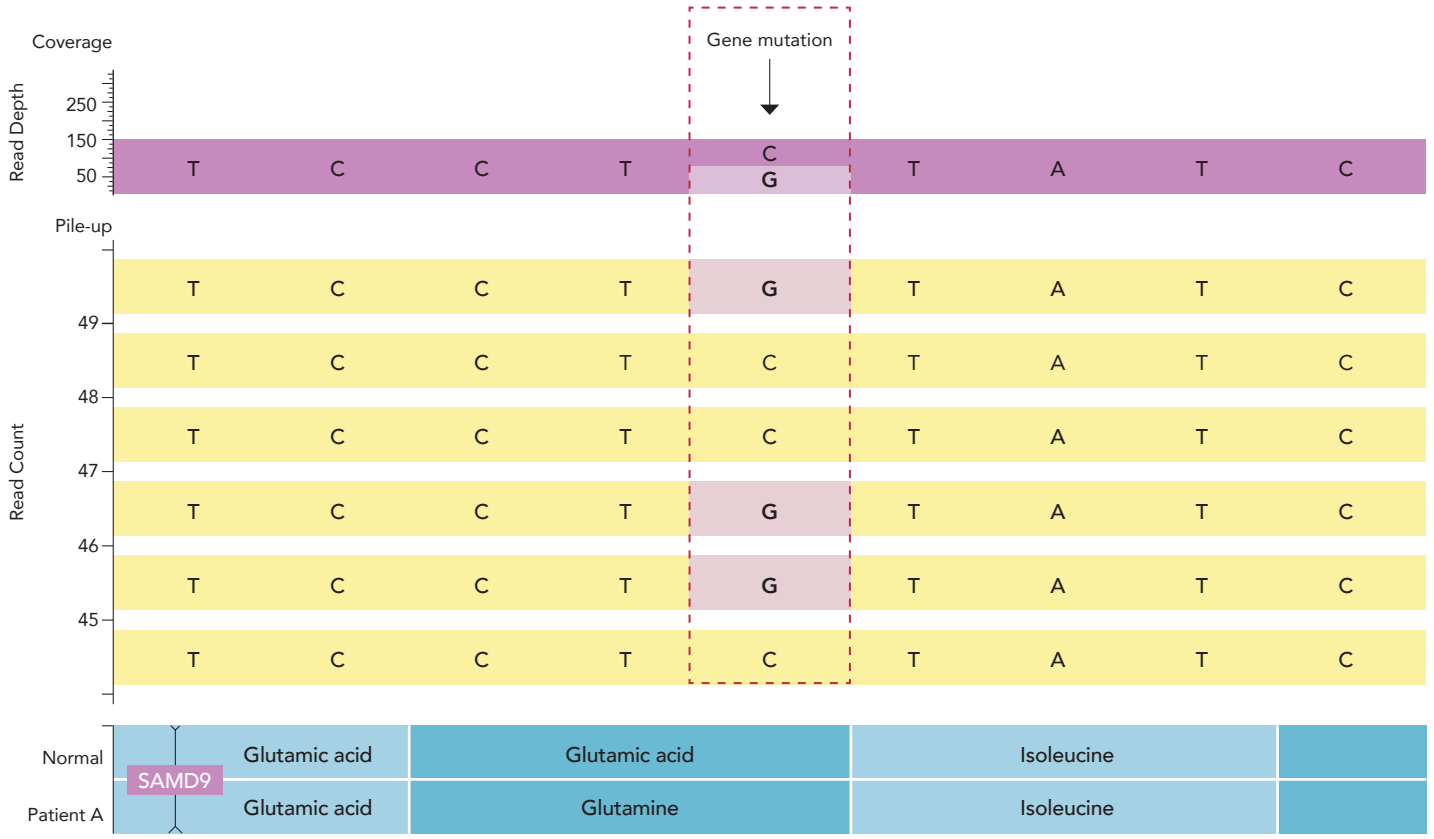
CASE 1: MIRAGE SYNDROME

Patient A was born premature and presented with hyperpigmentation of the skin. She was later found to have congenital adrenal insufficiency, with hypoplastic adrenal gland as well as severe persistent thrombocytopenia (low platelet count).

Enrolled in RapidSeq, the patient was found to have a *de novo* known pathogenic mutation C>G in the *SAMD9* gene (indicated by the red box in Figure 1). Mutations in the *SAMD9* gene are associated with MIRAGE (Myelodysplasia, Infection, Restriction of growth, Adrenal hypoplasia, Genital phenotypes and Enteropathy) syndrome, which is consistent with the patient's clinical presentation. This disorder was first discovered in 2016[6] and there are fewer than 30 known cases worldwide.

This unifying diagnosis provided more clarity to the patient's care team, and surveillance was instituted to observe for associated complications such as haematological malignancy and immunodeficiency. The diagnosis also ended the diagnostic odyssey for the patient and family, providing clarity – through genetic counselling – on the condition and her expected developmental prognosis. The patient is currently doing well and receiving follow up care at KKH.

Figure 1. Whole exome sequencing result, identifying a known pathogenic mutation C>G (indicated by the red box) in the *SAMD9* gene of the patient, which corresponded to a change in the amino acid from glutamic acid to glutamine. Mutations in the *SAMD9* gene are associated with MIRAGE syndrome.



CASE 2: CRANIOECTODERMAL DYSPLASIA

Patient B presented for prenatal genetic counselling while pregnant. She had had a previous pregnancy which had been terminated as the fetus showed signs of skeletal dysplasia. No genetic diagnosis was made at that time, but the couple had been counselled that the risk of recurrence in subsequent pregnancies was low. However, an antenatal scan in the second pregnancy showed fetal anomalies similar to the first pregnancy.

Using a stored fetal DNA sample from the first pregnancy, the family was enrolled in RapidSeq. The team discovered pathogenic variants detected in *IFT122* gene, which is consistent with the diagnosis of cranioectodermal dysplasia. Cranioectodermal dysplasia is a multisystemic disorder causing skeletal dysplasia, ectodermal defects and joint laxity. Fewer than 60 cases have been reported in literature[7].

Based on these results, the patient underwent amniocentesis to obtain a definitive diagnosis – which confirmed the same pathogenic variants in the *IFT122* gene. As the condition is inherited recessively, the risk of recurrence is 25 per cent, and the couple was counselled accordingly and provided options regarding future pregnancies.

HIGH DIAGNOSTIC YIELD WITH A SHORT TURNAROUND TIME

The first of its kind in the local setting, RapidSeq is made possible through a collaborative effort between the Genetics Service, DNA Diagnostic and Research laboratory and Translational laboratory, with specialists from NICU and CICU.

Eligible patients are first assessed by the clinical geneticist, and enrolled patients undergo sequencing targeting exonic regions of 4,800 genes associated with human diseases (also referred to as the clinical exome). The variants generated are filtered and prioritised using in-house computational algorithms.

Shortlisted variants are then correlated with clinical phenotype and classified based on established guidelines. Lastly, a clinical report is generated, which is shared with the primary care team – to inform the management of the patient's condition – as well as the patient's family, to provide answers and a measure of closure.

A positive genetic diagnosis in critically-ill patients can play a valuable role in guiding clinical care management and therapeutics. This can result in benefits such as treatment modification, initiating a new treatment, or surfacing the need to involve other specialists in the clinical care of the child.

It can also guide the care team in potentially reducing unnecessary investigations such as stopping a treatment that is not useful, or precluding a potentially invasive measure.

In some cases, a diagnosis with a dire prognosis can help to bring closure, and enable the care team and family to discuss treatment limitations and palliative care options. Additionally, the family will benefit from genetic counselling about the risk of recurrence in future pregnancies, for the same genetic disorder.

Where a patient's condition remains undiagnosed, other forms of tests, including whole genome sequencing, can be offered as per their clinical indication, to better understand their conditions.

Research efforts are ongoing at KKH to ascertain the diagnosis by using newer tools as they become available. Regardless, the patient will continue to be managed symptomatically albeit without a diagnosis.

Regardless of the diagnosis, RapidSeq enables the critically-ill patient and their family to receive information and, in some cases answers, in a timely manner. This is in comparison to a longer wait time for serial genetic testing in a stepwise manner with more traditional technologies.

With the promising results of these first 10 patients, the team continues to explore the impact and benefits of RapidSeq on optimising clinical care for paediatric patients in the ICU.

The co-authors of this article gratefully acknowledge the support and involvement of their colleagues in Rapid Genomic Sequencing at KKH. Special thanks to Dr Tan Ene Choo, Principal Investigator, KK Research Centre; and Associate Professor Law Hai Yang, Chief Scientific Officer, Genetics Service, KKH and their teams, as well as the multidisciplinary teams involved in the care of these critically ill patients.

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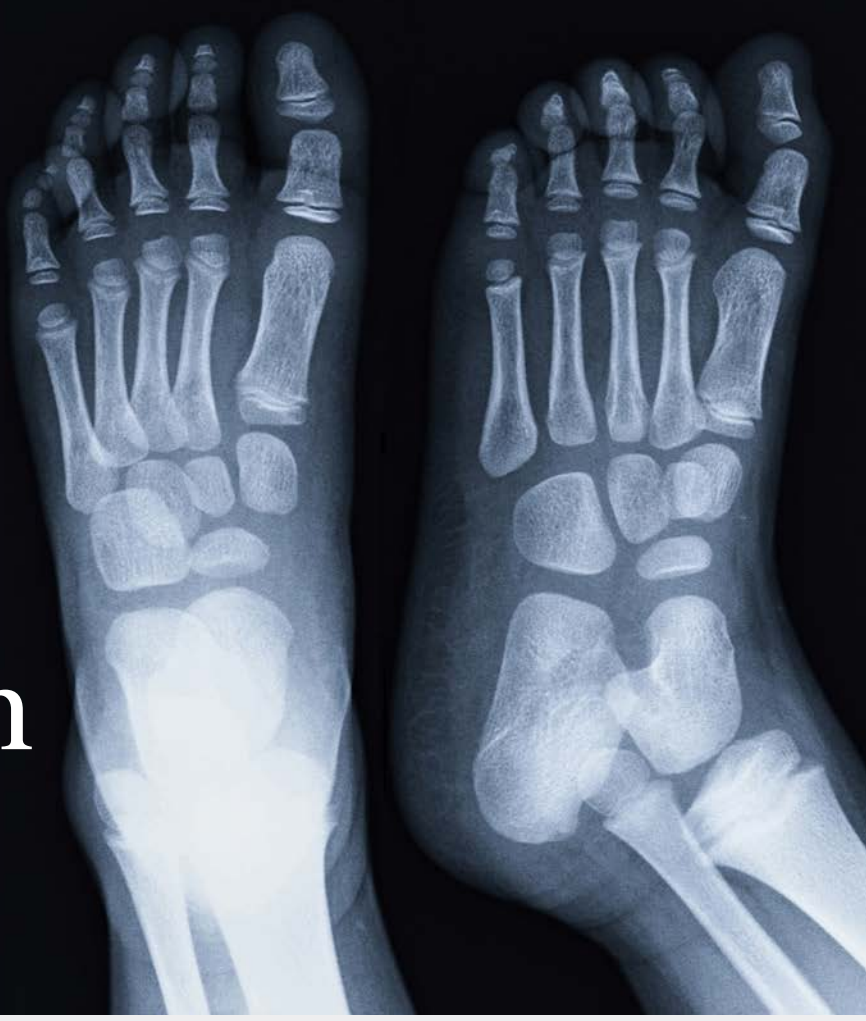
Dr Ting Teck Wah graduated from the National University of Singapore in 2006 and completed his paediatric medicine specialist training in 2014. His research interests are in the area of inborn errors of metabolism and genetics of neurodevelopmental delay. In 2017, Dr Ting completed his fellowship in the metabolic department of The Royal Children's Hospital Melbourne, Australia.

Dr Saumya Jamuar, Senior Consultant, Genetics Service, KK Women's and Children's Hospital

Trained in clinical genetics at the Harvard Medical School in Boston, United States, Dr Saumya Jamuar is a clinical geneticist at KKH, and serves as the lead primary investigator of the Singapore Childhood Undiagnosed Disease Programme. Dr Jamuar is also the clinical director of the SingHealth Duke-NUS Institute of Precision Medicine. Actively involved in translational research, Dr Jamuar has published in top tier journals and has won multiple awards for his research, including the SingHealth Outstanding Young Researcher Award in 2015.

3D-Printing Tech Helps Children With Limb Deformities To Walk Again



By Dr Lam Kai Yet

The etymology of the word "orthopaedic" comes from the Greek words "orthos" – meaning "straight or correct" and "paedieia" – meaning "rearing of children".

Limb discrepancies and deformities can occur in children as the result of a range of causes such as congenital, trauma, infection, growth disturbance and tumours. Limb deformities can present as bowed legs, knock-knees or a difference in limb lengths. Secondary symptoms include an awkward walking gait, short stature and, in rare instances, pain.

KK Women's and Children's Hospital (KKH) sees about 50 new cases of limb discrepancies and deformities each year. Of these, about 10 per cent are complex deformities, and 10 per cent as part of very rare conditions. Utilising 3D-printing technology, the paediatric Lower Limb Discrepancy (LLD) and Deformity Clinic at KKH is able to carry out a range of intervention and reconstruction options for a wide spectrum of complex and rare lower limb discrepancies and deformities in children.

Based on the patient's actual limb, anatomically accurate 3D-printed limb models are reconstructed – enabling patients and their families to better visualise the discrepancy or deformity. The patient's limb model is subsequently utilised in the assessment, planning, surgical simulation, family counselling and eventual surgical intervention for the patient. At KKH, this method has been successfully used in the planning and simulation of corrective surgeries, including:

- Fixation of a comminuted calcaneus fracture
- Hip arthroscopy in the resection of a cam lesion
- Valgus proximal femur osteotomy for a malunited femur neck fracture
- Arthroscopic resection of a physeal bar

Studies have shown that the use of 3D-printed limb models is helpful in improving patients' appreciation and understanding of their anatomy[1,2]. Additionally, being able to view a simulation of the entire corrective process and likely end result has greatly enhanced patient engagement and feedback. It also manages more accurately patient and parent expectations. This has led to greater patient satisfaction with the correction.

CASE STUDY: 3D-PRINT-ENABLED INTERVENTION FOR AN ADOLESCENT WITH COMPLEX FOOT DEFORMITY

An adolescent boy was diagnosed at birth with bilateral clubfoot deformities (Figure 1A) due to neuromuscular imbalances from an underlying spina bifida. The spinal cord condition also resulted in weak lower limb muscles.

Due to the boy's feet deformities, he was unable to wear normal shoes or stand properly, and often developed painful calluses and ulcers on his feet.

The boy underwent several operations to correct the deformities when he was younger in an attempt of an acute correction to allow for shoeing. However, the deformity recurred after each surgery since the primary source of the deformity in the spinal cord remained. The condition progressively worsened.

Due to the severity and long-standing nature of the boy's feet deformities in adolescence, surgical correction was not amenable using standard means. Correcting the deformities acutely could result in damage to the blood and nerve supply in the feet, possibly causing necrosis of the skin.

To assess the complexity of the deformities, the care team at the KKH LLD and Deformity Clinic created anatomically-correct, 3D-printed manipulable models of the boy's feet (Figure 1B). The 3D-printed feet models were then used to simulate surgical correction via different techniques. Clubfoot can be corrected gradually via Ilizarov frames, or a computer-based hexapod frame.

The completed simulated frame and model was shown and explained to the patient and his parents, enabling them to better understand the limb correction process, visualise the likely end result, and better prepare themselves for the surgery, as well as manage expectations.

During the actual surgery, the 3D-printed limb models were brought into the operating theatre to guide the positioning of the Taylor spatial frames (Figure 1C). Prior surgical simulations aided the team in achieving a satisfactory shape, alignment and size for the boy's feet, as planned pre surgery (Figure 1D).

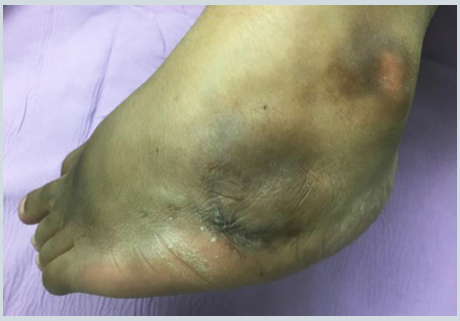


Figure 1A. Clubfoot deformity in an adolescent boy

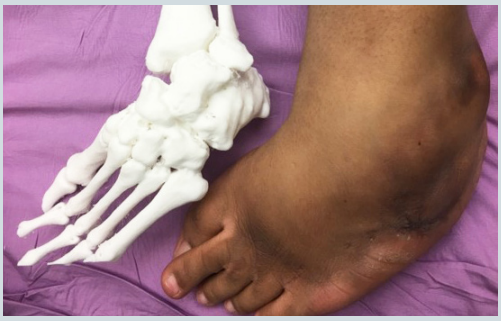


Figure 1B. Anatomically-correct 3D-printed model of the boy's foot

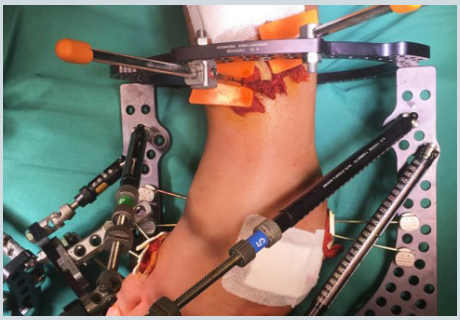


Figure 1C. Taylor spatial frame fitted to the boy's foot

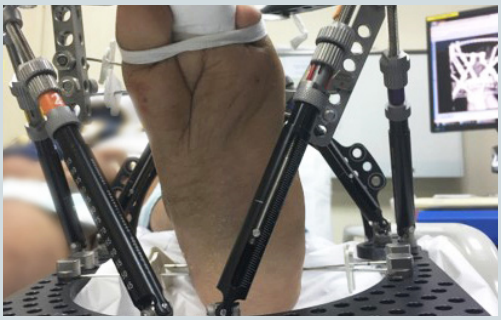


Figure 1D. Foot alignment using the Taylor spatial frame to achieve a satisfactory shape and size

After being fitted with the Taylor spatial frames for close to five months, the frames were removed and the boy was fitted with ankle-foot orthoses, enabling him to wear normal sports shoes which he had not been able to do throughout his childhood. He is currently undergoing physiotherapy and is able to stand with a walking frame. With continued follow up care at the LLD and Deformity Clinic, he is likely to progress to ambulatory exercises.

BENEFITS OF 3D-MODEL-GUIDED SIMULATION AND SURGERY

The use of anatomically-correct, 3D-printed manipulable feet models enabled the team to better appreciate the limb deformities on all three planes, which can be more accurate in comparison to traditional modes of diagnostic and interventional imaging which are viewed on a two-dimensional screen.

Additionally, by simulating the feet models undergoing surgery with each of the frame techniques, the team was able to assess the feasibility of each construct, the ease of correction, and more importantly, the precision of the final correction. This aided the team in deciding on the hexapod Taylor spatial frame as the better option as well as examining the ideal level at which to perform the osteotomy (bone cuts).

The surgical simulation further enabled the team to identify, pre-empt and overcome technical challenges which, previously, would have been discovered intra-operatively. This included the issue of the distal ring abutting a proximal U-ring due to the severity of the deformity, which would have obstructed the correction of the connecting struts. In this instance, a ¾ distal ring was chosen for its decreased rigidity, allowing just enough pliability to avoid abutment of both rings.

Continued on page 8...

Continued from page 7...

MULTIDISCIPLINARY CARE FOR LIMB DEFORMITIES IN CHILDREN

To help each child develop to their fullest potential, it is crucial for limb discrepancies and deformities to be assessed and corrected before secondary complications such as contractures, arthritis or scoliosis develop.

Through the sharing of knowledge and best practices amongst larger centres around the world, the repertoire of skills and surgical options is increasing. Today, most limbs can be reconstructed and deformities that were once thought to be too severe, or trivial can be addressed. Patients can be offered a wider range of options in limb reconstruction.

For the child with a limb discrepancy or deformity, the aim of treatment is to have a limb with aligned mechanical axes, functional, pain-free and equal in limb length. Most patients will need a detailed surgical life-plan. Appropriate surgical procedures are planned at certain stages in their life such that they can achieve the above aims by the time they reach skeletal maturity.

If the deformities are part of a physiological spectrum (e.g. physiological bow-leggedness in children less than two years old), reassurance is often all that is needed. However, any child with unequal limb length, or with obvious limb deformities (e.g. a varus or valgus deformity, flexion or extension deformity or even a rotational deformity) can be referred for assessment by the KKH LLD and Deformity Clinic.

ABOUT THE KKH LLD AND DEFORMITY CLINIC

The LLD and Deformity Clinic at KKH sees and manages a wide range of paediatric conditions, which can include rare and complex conditions such as fibula and tibia hemimelias, congenital femoral deficiencies, osteogenesis imperfecta (brittle-bone disease), congenital pseudarthrosis of tibia, arthrogryposis, Blount's disease, rickets, skeletal dysplasias, resistant clubfeet and multiple exostoses.

Upper limb conditions such as unusually short arm lengths, or deformed elbows and forearms due to previous infection, trauma or growth disturbance and tumours are also seen at the clinic.

Due to the complex nature of limb deformities, the clinic is staffed by a multidisciplinary care team, and designed to provide patients and families time and information to understand and appreciate the problem and treatment options available.

Following a consultation with the orthopaedic surgeon, a physiotherapist and an occupational therapist are also in a room adjacent to the consultation room to advise patients and their families on rehabilitation requirements and equipment that they would require post-surgery. Patients who need to be put in external fixation frames for a period of time would also be counselled by specialty care nurses who would review pin site wounds and offer advice such as special clothing and home modifications.

The clinic has also successfully incorporated the use of growth modulation, Ilizarov frames, advanced hexapod frame technology and internal lengthening rods to surgically correct these deformities. These advanced techniques make surgical correction safer and more precise, and in some cases, provide a better cosmetic result.

In conclusion, almost all limb deformities can now be managed and reconstructed. Any limb deformity should not be trivialised. With currently available technology and expertise, there is no reason for patients to live with a limb discrepancy or deformity if they choose not to.

REFER A PATIENT

Healthcare professionals can refer paediatric patients to the Department of Orthopaedic Surgery at KKH for tertiary assessment of their limb deformities by contacting the hospital at **+65 6294 4050.**

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Dr Lam Kai Yet obtained his basic medical and postgraduate qualifications from the National University of Singapore and the Royal College of Surgeons of Edinburgh, United Kingdom. He also has a Diploma in Children's Orthopaedics from the European Paediatric Orthopaedic Society. Upon completion of his specialist training in orthopaedic surgery, Dr Lam joined KKH in 2014, and is actively involved in research in 3D-printing, paediatric trauma, and sports injuries.

Dr Lam's areas of interest include children and adolescent sports injuries, patella dislocations, hip dysplasia, osteogenesis imperfecta, and limb deformity correction and limb lengthening. He has since completed an arthroscopy and knee fellowship at the Centre of Albert Trillat, Hospital de la Croix-Rousse, France, and a paediatric orthopaedics fellowship at the Great Ormond Street Hospital, London, and University College Hospital London.

Dr Lam is currently an Adjunct Assistant Professor at the Lee Kong Chian School of Medicine, and a Senior Clinical Lecturer at the Yong Loo Lin School of Medicine. A member of the Singapore Orthopaedic Association Humanitarian and Outreach Subcommittee, Dr Lam compassionately cares for the underprivileged in Singapore and regional countries.

Beating Scoliosis With MAGEC Growing Implants

By Ms Lee Li Wen, Ms Christina Ong and A/Prof Kevin Lim

At KK Women's and Children's Hospital (KKH), MAGEC (MAGnetic Expansion Control) rod implants are helping children with severe early-onset scoliosis to achieve spinal correction with less pain and fewer surgical procedures.

Comprising magnetically-controlled, implantable rods and an external remote control, the MAGEC growing rods are a new surgical treatment used to guide spinal growth in young children with scoliosis.

Scoliosis is characterised by an 'S' or 'C'-shaped lateral curvature of the spine, commonly accompanied by a degree of rotation leading to a three-dimensional deformity. There may be a curving of the body to one side when viewed from the back, and asymmetry of the shoulder or scapulae height and waistline. The condition is seven times more common in females than in males.

While the incidence of scoliosis is highest amongst adolescent children (aged 10 to 16 years) – comprising 70 to 80 per cent of newly diagnosed cases at KKH – idiopathic scoliosis can also present in younger age groups: infantile (<3 years old) and juvenile (4-9 years old). These subtypes of scoliosis are considered to be 'early onset' due to the child's young age, and where the curvature of the spine is severe (exceeding a Cobb angle of 40˚), special considerations for surgical treatment are required, as the child has many years of growth remaining.

NON-INVASIVE MAGNETIC SPINAL DISTRACTION

When a child undergoes spinal correction via MAGEC rod implantation, first a single or dual MAGEC rod is implanted into the spine. Three months after implantation, spinal distractions will commence in the outpatient clinic setting. First, the magnet in the MAGEC rod implant is located (Figure 1A). Then, an external remote control device is lined up with the magnet, and used to magnetically "grow" or lengthen the implant to align the spine and reduce its curvature (Figure 1B). Some discomfort or a stretching sensation is expected during the process, which is generally well tolerated.

FIGURE 1. DISTRACTION (LENGTHENING) OF A MAGEC ROD

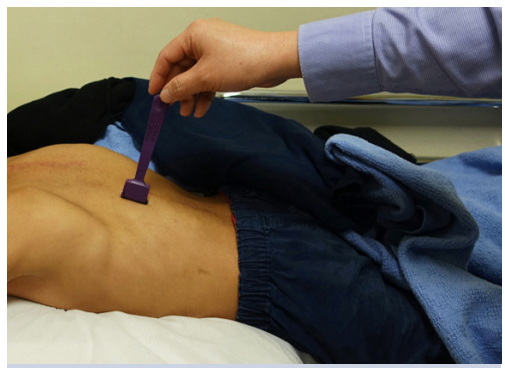

Figure 1A. The magnet in the middle part of a MAGEC rod implant is identified.

Figure 1B. An external remote control is lined up with the magnet, and the MAGEC rod is lengthened to align the patient's spine by pushing a button on the remote control.

The novel MAGEC rod implant method is a boon for the young child with severe early-onset scoliosis, as up to recent years, the traditional surgical option and standard of care utilised stainless steel rod implants which required lengthening every six to nine months, in tandem with the growth of the child's spine. Each lengthening procedure required the child to undergo an invasive surgery under general anaesthesia.

In contrast, MAGEC rod implantation allows for non-invasive gradual distraction of the spine (from three to five millimetres at a time) at shorter time intervals of up to three minutes per session, without a need for general anaesthesia and hospitalisation. To keep up with the growth of their spine, younger patients can expect to return to the outpatient clinic for spinal distraction as frequently as once a month.

Following each distraction, the patient can immediately resume their daily activities, such as attending school. They are advised against strenuous activities and contact sports, and should not undergo magnetic resonance imaging.

Children who are eligible for intervention via MAGEC rod implantation include those who are diagnosed with severe early-onset scoliosis (Cobb angle of 50˚ or more) and are at risk of further curve progression.

At present, two paediatric patients in KKH have undergone MAGEC rod implantation over the last two years; both have made good progress and remain pain-free. Their treatment processes are described as follows:

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Continued from page 9...

CASE STUDY 1: SINGLE MAGEC ROD IMPLANT

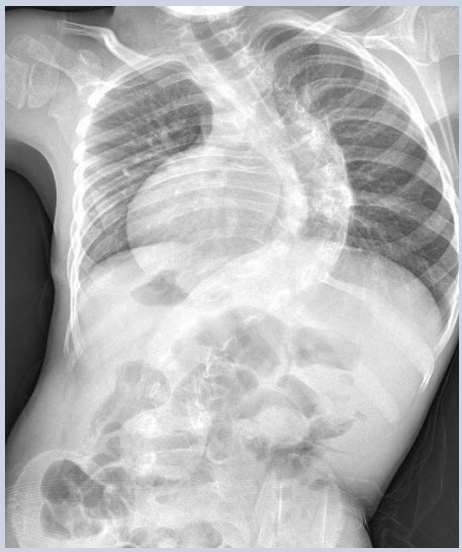

Figure 2A. X-ray scan showing a spinal curvature with a Cobb angle of more than 100˚ before commencing MAGEC rod implantation.

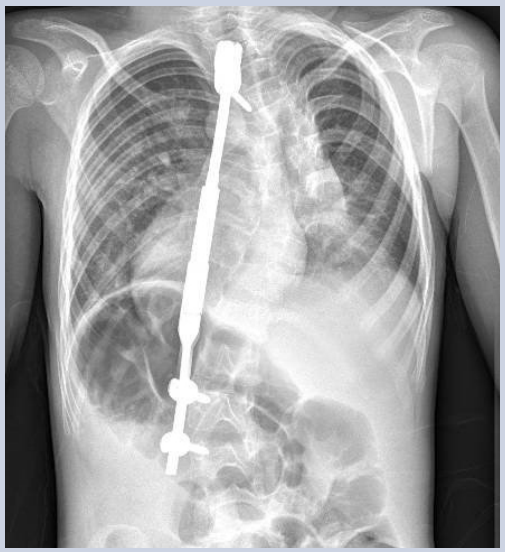

Figure 2B. X-ray scan indicating a reduced spinal curvature three months after the implantation of a MAGEC rod.

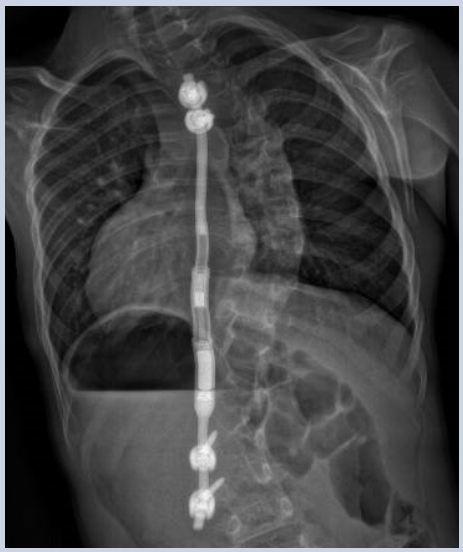

Figure 2C. X-ray scan showing a reduction in spinal curvature of more than 50 per cent, six months after the implantation of a MAGEC rod.

Patient A presented to KKH in early childhood with a spinal curvature Cobb angle of more than 100˚, and was diagnosed with severe early-onset scoliosis (Figure 2A). Due to the severity of the spinal curve and the patient's young age, he was recommended to undergo implantation of a MAGEC rod. After a discussion of the potential risks and benefits with the patient and his family, a single MAGEC rod was implanted to correct the patient's scoliosis.

Post-operatively, the patient has since undergone five non-invasive distraction procedures at the KKH Orthopaedic Outpatient Clinic to progressively lengthen and adjust the MAGEC rod, with each procedure taking place every four to six weeks (Figure 2B). At the patient's most recent hospital visit, his spinal Cobb angle has improved to 46˚, indicating a reduction of his spinal curvature by more than 50 per cent (Figure 2C).

THE FUTURE OF SCOLIOSIS MANAGEMENT

Since the use of MAGEC rod implantation was approved by the United States Food and Drug Administration in 2014, there have been multiple publications regarding the efficacy of the method in comparison to its traditional counterpart[1]. Preliminary short-term results have showed promise and has potential in routine clinical practice. Additionally, in spite of the larger expense at the start of therapy, the cost of magnetically-controlled growing rods becomes comparable to traditional systems by a duration of four years[2]. More is to be learned about the merits and safety considerations of the MAGEC rod as patients are followed up for longer periods of time.

REFER A PATIENT

Healthcare professionals can refer paediatric patients to the Department of Orthopaedic Surgery at KKH for tertiary assessment of their scoliosis and management options, by contacting the hospital at **+65 6294 4050.**

References:

1. *Johari A and Nemade A. Growing spine deformities: Are magnetic rods the final answer? World Journal of Orthopedics 2017; 8(4): 295-300.*
2. *Charroin C, Abelin-Genevois K, Cunin V, Berthiller J, Constant H, Kohler R, Aulagner G, Serrier H, Armoiry X. Direct costs associated with the management of progressive early onset scoliosis: estimations based on gold standard technique or with magnetically controlled growing rods. Orthopedics & Traumatology: Surgery and Research 2014; 100: 469-474*

CASE STUDY 2: DUAL MAGEC ROD IMPLANT

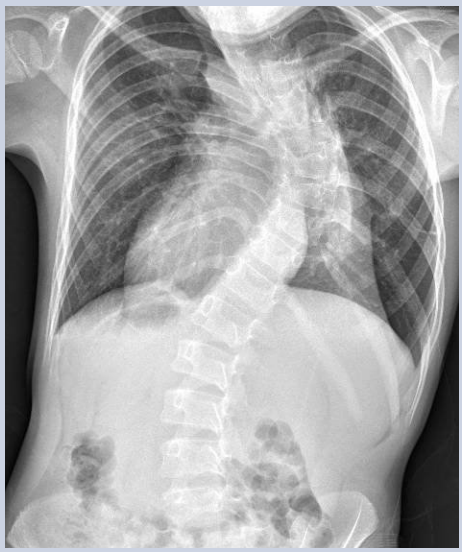

Figure 3A. X-ray scan showing a spinal curvature with a Cobb angle of 68˚ before commencing MAGEC rod implantation.

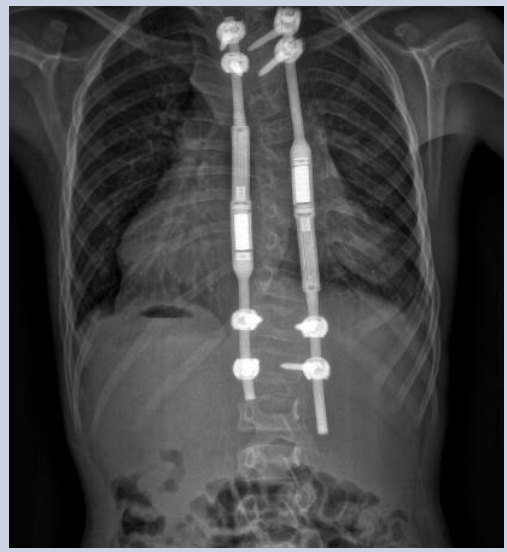

Figure 3B. X-ray scan indicating a reduction of spinal curvature immediately after the implantation of dual MAGEC rods.

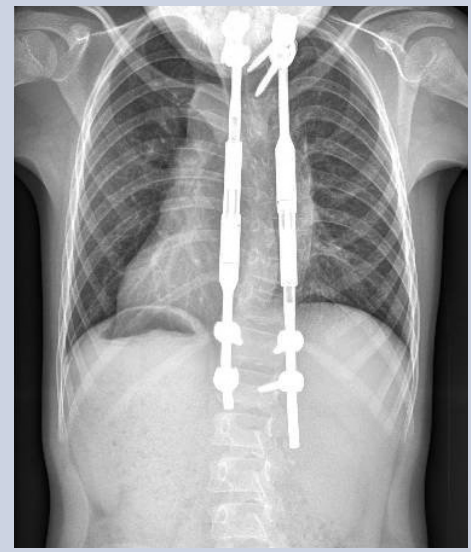

Figure 3C. X-ray scan showing a reduction of spinal curvature to a Cobb angle of about 25˚ eight months after the implantation of the MAGEC rods.

Patient B presented to KKH in his early pre-teens with a spinal curvature Cobb angle of 68˚ (Figure 3A). Two MAGEC rods were successfully surgically implanted for the correction of his scoliosis (Figure 3B). Post-operatively, the patient has undergone four non-invasive distraction procedures at the KKH Orthopaedic Outpatient Clinic to progressively lengthen and adjust the MAGEC rods.

At the patient's most recent visit, his spinal Cobb angle was measured at 25˚ (Figure 3C), indicating a reduction in his spinal curvature of more than 50 per cent compared to preoperatively.

Patient A and B will continue to undergo regular distractions of their MAGEC rods implants until their early teens, when they reach skeletal maturity. At which time the rods will be removed, and spinal fusion surgery – which permanently fuses the vertebral column of the spine – will be performed to manage the progression of the spinal curvature as they grow to adulthood.

Ms Lee Li Wen, Medical Student, Duke-NUS Medical School

Ms Lee Li Wen is a third-year medical student at Duke-NUS Medical School. Ms Lee underwent an attachment with the Division of Surgery, KKH, in February 2019.

Ms Christina Ong Mei Zhen, Intern, Division of Surgery, KK Women's and Children's Hospital

Ms Christina Ong Mei Zhen is a final-year student at Nanyang Technological University, pursuing a Bachelor of Science (Honours) in Biological Sciences, with a Minor in Business. Ms Ong is currently undergoing an internship with the Division of Surgery, KKH.

Associate Professor Kevin Lim, Chairman, Division of Surgery, KK Women's and Children's Hospital and Academic Deputy Chair, SingHealth Duke-NUS Surgery Academic Clinical Program

Associate Professor Kevin Lim underwent subspecialty training in paediatric orthopaedics at The Hospital for Sick Children in Toronto, Canada and at The Starship Children's Hospital in Auckland, New Zealand. His subspecialty interests include scoliosis, clubfoot, cerebral palsy, and fractures in children.

In addition to his roles at KKH and SingHealth, A/Prof Lim is Board Chairman and volunteer doctor at the Cerebral Palsy Alliance Singapore. He is also an elected board member of the National Council of Social Service.

Life Changing Collaboration In Jakarta

KKH team pilots a ground-breaking occupational therapy education programme to benefit children with special needs

KKH occupational therapist, Ms Emily Ong, conducts a workshop for training participants at YPAC Jakarta.

In February 2019, a team of occupational therapists from KK Women's and Children's Hospital (KKH) flew to Jakarta, Indonesia, to conduct a three-and-a-half-day workshop for 36 special education (SPED) practitioners from 10 local institutions for children with special needs.

No ordinary workshop, it marked the ground-breaking inauguration of a four-year partnership between Singapore Health Services (SingHealth), Singapore International Foundation (SIF) and Foundation for the Development of Children with Special Needs (YPAC) Jakarta, aimed at enhancing educational support for children with special needs in Jakarta.

Over four years, the KKH team aims to provide occupational therapy training for 50 SPED practitioners from YPAC Jakarta and other local institutions, with the potential to benefit an estimated 1,750 children with special needs and their families. From the cohort of training participants, a core team of 20 master trainers will receive additional training to cascade their knowledge to others in the field.

STRENGTHENING SUPPORT FOR CHILDREN WITH SPECIAL NEEDS

"Currently, the majority of children with special needs in Indonesia are enrolled in special or inclusive schools," shares Ms Soh Siok Khoon, Head and Principal Occupational Therapist, Occupational Therapy Service, KKH, who also leads KKH's involvement in the SingHealth-SIF-YPAC partnership.

Based on a KKH study trip to Jakarta in 2017, visiting key institutions for children with special needs, the main clinical conditions observed in the children included cerebral palsy, intellectual disabilities and developmental disabilities.

"From an occupational therapy perspective, four areas have been identified that could benefit from improvement: staff knowledge and skills in the management of children with special needs; frequency and access to intervention by children with special needs; availability of training opportunities for staff; and interdisciplinary communication and collaboration in the management of children with special needs," Ms Soh adds.

"Our aim is to begin the task of upskilling the staff at the various institutions for children with special needs, building their clinical skills and knowledge, and equipping them to deliver evidence-based therapeutic interventions, strategies and tools. To accommodate resource and manpower limitations, developing a trans-disciplinary mindset and approach is also vital to optimising the children's access to and frequency of intervention."

EMPOWERING EDUCATORS WITH EVIDENCE-BASED SKILLS

The inaugural workshop in February was attended by a wide range of SPED practitioners, including occupational therapists, physiotherapists, speech therapists, special education teachers and institutional-based caregivers. Training was conducted along two parallel tracks for generalists and specialists.

Along the generalist track, all training participants were equipped with tools and strategies to adopt an integrated approach in the management of children with special needs – such as a common language to communicate about students' needs, goals and plans; universal intervention strategies; and tools for assessing students' performance of school-based occupations, classification of student's functioning levels, and facilitating basic activities of daily living, such as showering, feeding, grooming and toileting.

(Front row centre) KKH occupational therapists, Ms Foo Ce Yu, Ms Jo Chen and Ms Emily Ong at the launch ceremony of the Occupational Therapy for Children with Special Needs programme, together with representatives from SIF and YPAC Jakarta.

A smaller cohort of master trainers was identified to participate in the specialist track, where they received further equipping to cascade the training to other healthcare and education professionals, and caregivers.

"Special needs education is collaborative and interdisciplinary in nature. Bridging the understanding between SPED practitioners on childhood occupations, and helping them to establish a common language to communicate about the children's needs, goals, and plans, is absolutely crucial to the seamless delivery of care and intervention," says team member, Ms Jo Chen, Principal Occupational Therapist, Occupational Therapy Service, KKH.

"The hands-on training methods of workshops and case study discussions provided valuable opportunities for the participants to share cross-disciplinary perspectives, learn from one another, and establish networks of support. It is heartening to see that this first workshop has provided a common platform for the different SPED professions to come together and work in concert toward this common, shared vision," adds team member, Ms Foo Ce Yu, Principal Occupational Therapist, Occupational Therapy Service, KKH.

GEARING UP FOR LONG TERM, POSITIVE OUTCOMES

Over the next four years, the KKH team aims to conduct ongoing training, with a focus on clinical skills training to facilitate the performance of children with special needs in childhood occupations beyond the basic activities of daily living. These include facilitating handwriting performance, managing attention and behaviour issues, and facilitating play in children with special needs.

"To address key clinical conditions such as cerebral palsy, intellectual disabilities and developmental disabilities, the master trainers will also be provided more in-depth clinical skills training in sensory integration and neurodevelopmental treatment, to build their capabilities to provide more effective individual therapy to students with more significant needs," says team member, Ms Emily Ong, Occupational Therapist, Occupational Therapy Service, KKH.

Other training components will emphasise the development and enhancement of management tools and strategies; professional sharing via a symposium; and public education for patients and their caregivers to increase awareness of patient care requirements.

Seeking to benefit a wider community, the training programme will continue to work along a train-the-trainer model, with the aim of cascading skills and knowledge taught to wider groups of SPED practitioners and caregivers of children with special needs in Indonesia.

"In our role as allied health professionals, we have seen evidence that early intervention and support for children with special needs makes all the difference for their developmental growth. Empowering the children and the community enables long-term support and positive outcomes. It then becomes even more meaningful to be able to share and exchange knowledge and experience with our Indonesian friends," says Ms Soh.

"We hope that the training programme will be a key catalyst in upskilling and bridging understanding and communication between the professions and institutions involved, to enable robust and accessible interprofessional care and education to be delivered more effectively for Jakarta's children with special needs."

Caring For The Caregivers

KKH caregiver support programme helps families of chronically-ill children gain respite and resilience

Established by KK Women's and Children's Hospital (KKH) in partnership with Temasek Foundation Cares in 2016, to strengthen the psychosocial support available to caregivers of children with chronic illnesses, the Temasek Foundation Cares – Caregiver Support Programme for Families with Chronically Ill Children on Long-term Home Care is making a difference in families' lives.

"For chronically-ill children who require long-term medical home care, caregivers play a critical role in enabling the transition back into the home and community, and facilitating their daily care and activities of daily living," says Dr Cristelle Chow, Director, Caregiver Support Programme and Consultant, General Paediatrics Service, KKH.

However, the demands of long term home care for chronically-ill children can have a debilitating impact on the psychological and emotional health of caregivers. A survey of 88 caregivers of children in the KKH Paediatric Home Care Programme between March 2015 and January 2016 found that 44.3 per cent had CES-D (Center for Epidemiologic Studies Depression) scores that suggested that they were at significant risk of clinical depression. The Family Functioning Summary Scores self-reported by the caregivers of these patients were also lower compared to published literature.

"As the health of caregivers has a direct impact on the health of patients, they can be said to be our 'second patients'," says Dr Chow. "The Caregiver Support Programme was born to strengthen the psychosocial support available to these caregivers of children with chronic illnesses, and introduce screening and intervention for those identified with moderate-to-high perceived stress levels and who are at risk of clinical depression."

POSITIVE INTERIM INTERVENTION OUTCOMES

Two years on, the Caregiver Support Programme is showing positive interim intervention outcomes.

To date, the pilot programme has screened 326 caregivers from the KKH Paediatric Home Care Programme, of which 110 caregivers who were categorised as being at high or moderate risk according to their perceived stress and risk of depression, have accepted the intervention provided.

Mrs Kang (left), the main caregiver of daughter En Ning (centre), who has Antley Bixler syndrome, has benefited from the support and respite care provided through the Caregiver Support Programme.

"The interventions help to provide caregivers with skills to improve their resilience, stress management and relationships with other family members. For caregivers who are home-bound due to their child's high care needs, the medical social worker and respite nurse also provide a listening ear and a helping hand," says Ms Maryani Bte Abdul Wahab, a Nurse Clinician with the Paediatric Home Care team.

Seventy per cent and 86 per cent of caregivers who received medical intervention comprising psychosocial counselling showed improvements in their perceived stress scale and patient health questionnaire scores respectively. Additionally, 40 per cent of caregivers who received intervention improved their Family Functioning Summary Score.

RALLYING A NETWORK AND COMMUNITY SUPPORT

Prior to piloting the programme, a survey and focus group sessions were conducted to identify key areas where timely attention and intervention were needed – namely, respite care for the child and help in managing caregivers' physical and emotional stress.

"Based on these findings, we established a network of paediatric nurses from KKH, who are specially trained to manage complex care needs specific to children on long-term care in the home care setting," says Dr Chow. "This enables caregivers to care for themselves, including having couple-time, simply having a haircut, or spending time with their other children."

The programme has also developed a pedagogy and curriculum for a competency-based programme in respite care, training community providers to provide respite care services.

Adds Dr Chow, "For the continued and sustained expansion of caregiver support, it is vital for the model of caregiver respite to rally and include the community in supporting the child and family unit as a whole."

KKH Paediatrician Receives MOH Scholarship For Research Into Paediatric Respiratory Disease

Dr Judith Wong (left) receives her MOH Healthcare Research Scholarship – Master of Clinical Investigation Programme from Mr Chan Heng Kee, Permanent Secretary, Ministry of Health.

Dr Judith Wong, Consultant, Children's Intensive Care Unit, KK Women's and Children's Hospital (KKH), has been awarded a Ministry of Health (MOH), Healthcare Research Scholarship – Master of Clinical Investigation Programme for research into paediatric acute respiratory distress syndrome (PARDS), at the recent National Medical Research Council Awards Ceremony and Research Symposium 2019.

A significant and challenging disease entity in paediatric intensive care units globally, PARDS is associated with oxygenation failure and a high mortality rate of up to 65 per cent[1].

"Pneumonia is the most frequent cause of PARDS, and the progression of this disease from mild to severe occurs unpredictably with no current methods of prognostication," shares Dr Wong. "Other causes can include drowning, sepsis and burns. A proportion of patients with PARDS deterioriate and require extracorporeal membrane oxygenation (ECMO) life support."

Dr Wong's study team will look into differentiating the global lipid landscape of patients with PARDS to identify potential prognostic markers correlating to the clinical phenotype of mild, moderate and severe disease. The team also aims to characterise and compare the lipidomic profile of viral and viral/bacterial-coinfection-induced PARDS to better understand the pathogenic interplay between virus and bacteria, and shed more light on disease severity.

"The findings would be of great utility in informing condition diagnosis and prognosis, and the planning of intensive care therapies – which can ultimately aid us in improving the overall health outcomes in critically-ill children," adds Dr Wong.

Reference:

1. *Wong JJ, Loh TF, Testoni D, Yeo JG, Mok YH, Lee JH. Epidemiology of pediatric acute respiratory distress syndrome in singapore: risk factors and predictive respiratory indices for mortality. Frontiers in pediatrics. 2014;2:78. PubMed PMID: 25121078. Pubmed Central PMCID: PMC4110624. Epub 2014/08/15. eng.*

Strengthening Support In The Community For Children With Trauma

On 4 and 5 April 2019, international and local trauma experts and practitioners came together at Singapore's inaugural Child Trauma Conference – themed 'Prevention to Recovery' – to interact and share best practices in trauma recovery and building resilience in children.

Highlights included a panel discussion on developments in child trauma, led by eminent trauma experts, and a public forum enabling members of the community to learn more about identifying and mitigating stress, and building resilience in children.

Organised by KK Women's and Children's Hospital (KKH) with support from Temasek Foundation Cares, and graced by Madam Halimah Yacob, President of the Republic of Singapore, the conference was convened as part of the Stay Prepared – Trauma Network for Children established in 2016 to strengthen the capability of the community to support children affected by trauma, and their families.

To date, more than 2,300 children affected by various forms of trauma and parents have been provided with therapy and support through programmes under the Stay Prepared – Trauma Network for Children. Close to 1,050 therapists, school counsellors and social workers have also been trained to deliver the appropriate support and intervention for these children.

"Timely and targeted support and intervention are imperative in mitigating the adverse impact of trauma and building resilience in children following a traumatic experience," says Associate Professor Chan Yoke Hwee, Chairman, Division of Medicine, KKH and Director of the Stay Prepared – Trauma Network for Children.

"Another key aspect is the establishment of a learning network. This conference provides a platform for experts, practitioners, policy makers, and community leaders to come together with the aim to keep moving forward in community support for child trauma."

Guest-of-Honour Madam Halimah Yacob, President of the Republic of Singapore (centre), graces the symbolic launch of the opening ceremony of the Child Trauma Conference 2019, with (from left) Ms Woon Saet Nyoon, Chief Executive, Temasek Foundation Cares; Professor Alex Sia, Chief Executive Officer, KKH; Mr Richard Magnus, Chairman, Temasek Foundation Cares; and Ms Lynn Soh, Chairperson, Child Trauma Conference 2019 Organising Committee, KKH.

KK Women's and Children's Hospital
SingHealth

KK PARENTING CLUB

Research has shown that breast milk has the perfect combination of nutrients needed by newborns. There is absolutely no substitute for breast milk as it contains complete nutrition, antibodies and benefits required for optimal growth and development especially from birth to six months of age.

We also know that breastfeeding establishes strong bonds between mother and baby during the early stages of life, which is vital for the baby's emotional wellbeing and development. Studies show that babies who room-in with their mothers cry less than babies who stay in nurseries. As an accredited baby-friendly hospital under the Baby-Friendly Hospital Initiative – a global effort by the World Health Organisation, KKH supports breastfeeding and encourages all mothers to engage in skin-to-skin contact with their babies within five minutes after birth and to room-in with them.

Winners of Most Popular Baby Contest, KKH Healthy Breastfed Baby Contest 2018

Celebrates 25 Years of KKH Healthy Breastfed Baby Contest 2019

A STAR IS BORN IN KKH!

As part of KKH's efforts to promote and encourage breastfeeding in Singapore, the hospital has been organising the Healthy Breastfed Baby Contest since 1995. This year marks 25 years of healthy breastfed babies and we are proud of the more than 4,000 babies (and their mommies and daddies!) who have participated in this event over the years. In conjunction with the celebrations, KKH is also organising an inaugural talent search, 'A Star is Born in KKH!' contest. This contest calls for anyone who was born in KKH (all ages above 5 years old) to step out and showcase their special talent.

Please scan the QR code for more information on the contests

- KKH Healthy Breastfed Baby Contest 2019
- A Star is Born in KKH! Contest

ABOUT KK WOMEN'S AND CHILDREN'S HOSPITAL

KK Women's and Children's Hospital (KKH) is Singapore's largest tertiary referral centre for Obstetrics, Gynaecology, Paediatrics and Neonatology. Founded in 1858, the 160-year-old academic medical institution specialises in the management of high-risk conditions in women and children. More than 500 specialists adopt a compassionate, multi-disciplinary and holistic approach to treatment, and harness medical innovations and technology to deliver the best medical care possible.

Accredited as an Academic Medical Centre, KKH is a major teaching hospital for all three medical schools in Singapore, Duke-NUS Medical School, Yong Loo Lin School of Medicine and Lee Kong Chian School of Medicine. The 830-bed hospital also runs the largest specialist training programme for Obstetrics and Gynaecology and Paediatrics in the country. Both programmes are accredited by the Accreditation Council for Graduate Medical Education International (ACGME-I), and are highly rated for the high quality of clinical teaching and the commitment to translational research.