GOSH Conference 2023

Oral Presentations

13 SUCCESSFUL SEQUENTIAL HAPLOIDENTICAL MATERNAL HAEMATOPOIETIC STEM CELL AND KIDNEY TRANSPLANTATION FOR SCHIMKE IMMUNO-OSSEOUS DYSPLASIA

[1]Camille Laroche, [2]Giovanna Lucchini, [2]Wesley Hayes, [2]Iona Madden, [1]Stephen D Marks. *[1]Great Ormond Street Hospital for Children NHS Foundation Trust and University College London, UK; [2]Great Ormond Street Hospital for Children NHS Foundation Trust, UK*

10.1136/bmjpo-2023-GOSH.1

Schimke immuno-osseous dysplasia (SIOD) is a rare autosomal recessive disease caused by homozygous SMARCAL1 gene mutation leading to focal segmental glomerulosclerosis (FSGS), immunodeficiency and disproportionate short stature. The quality and quantity of life improvement relies on management of end-stage kidney disease (ESKD) and prevention of possibly fatal opportunistic infections caused by T-cell deficiency.

A five-year-old girl presented with steroid resistant nephrotic syndrome, dysmorphic features and short stature and had genetic confirmation of SIOD. In the year following her diagnosis, she reached ESKD and commenced haemodialysis (HD). The possibility of proceeding with maternal stem cell then kidney transplant from the same haploidentical donor was accepted by the hospital's ethic committee. She underwent conditioning treatment with pharmacokinetic adjusted chemotherapy followed by successful engraftment of the αβ T-cell– and CD19 B-cell–depleted haematopoietic stem cell transplantation (HSCT) at seven-years-old. She was switched from HD to continuous veno-venous haemofiltration (CVVH) for 24 days to sustain strict fluid control as chemotherapy and nutrition were given intravenously. She developed mucositis and mild skin graft versus host disease which resolved at discharge 40 days after conditioning had begun.

The 175-day period between HSCT and living-related kidney transplantation (LRKT) was uneventful. She underwent LRKT with no human leukocyte antigen (HLA) mismatch, no induction or anti-proliferative agents and low trough tacrolimus levels. She recovered normal kidney function five days post-transplant and remains stable with a median eGFR of 125 ml/min/1.73m^2 with discontinuation of all immunosuppressive therapy since then. Her infectious prophylaxis regimen included oral letermovir, aciclovir, co-trimoxazole and itraconazole.

Haploidentical HSCT prior to LRKT is a high-risk intervention which could improve life expectancy in patients with SIOD. To our knowledge, we are the first team to demonstrate the success of the Stanford protocol in a paediatric recipient receiving CVVH during HSCT conditioning treatment. This intervention led to a successful LRKT with minimal immunosuppression, which was the first case performed in our centre.

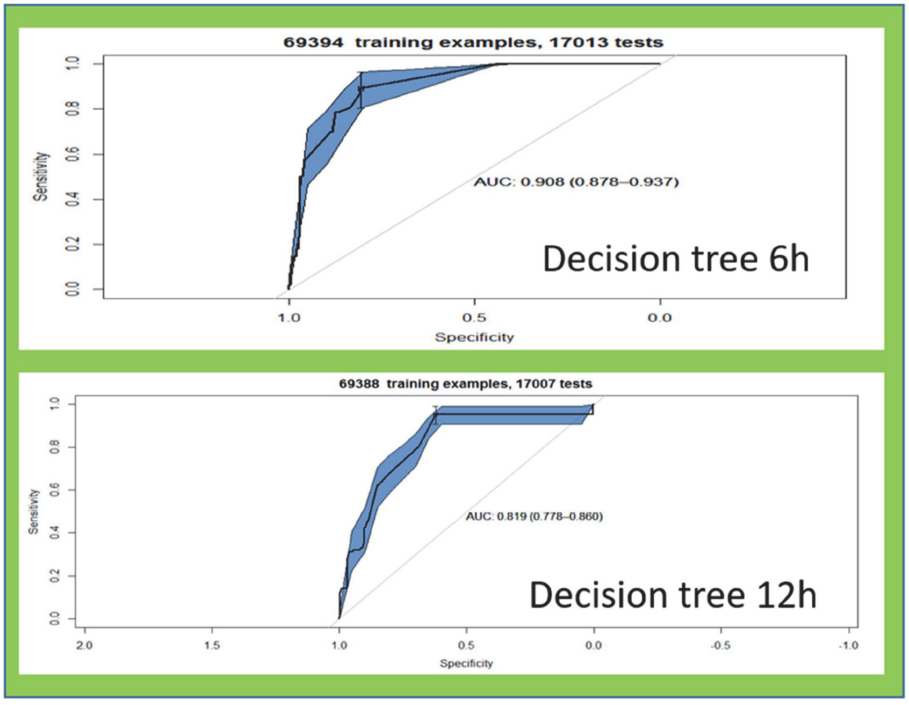
Acknowledgements for funding or support C. Laroche received funds from Sainte-Justine University Hospital, Montreal, to carry out her work at GOSH

56 PREDICTING DETERIORATION IN PICU PATIENTS USING ARTIFICIAL INTELLIGENCE

[1]Jack Pickard, [1]Samiran Ray, [2]Nazima Pathan. *[1]Great Ormond Street Hospital for Children NHS Foundation Trust, UK; [2]Cambridge University Hospitals NHS Foundation Trust, UK*

10.1136/bmjpo-2023-GOSH.2

How likely is a Paediatric Intensive Care Unit (PICU) patient to survive the next six hours, or the next 12 hours? Six years



Abstract 56 Figure 1

of EHR big data (2120 patients) from Cambridge University Hospitals PICU will soon be validated using GOSH PICU data (2686 patients). A decision tree algorithm combined with proportional-integral-derivative analysis and polynomial modelling. 100% sensitivity and 50% specificity (AUC 0.93) for predicting death within the following six-hour time period, with 95% sensitivity and 70% specificity for a 12-hour time period (AUC 0.82) applying a gradient boosted decision tree to the Cambridge dataset. A potential means to intervene, manage or communicate regarding patient acuity on PICU.

Acknowledgements for funding or support This work was made possible through the support of the Biomedical Research Centre at the University of Cambridge/Cambridge University Hospitals NHS Foundation Trust

68 NON-INVASIVE VENTILATION FOR SLEEP DISORDERED BREATHING: PARENT RECOMMENDATIONS FOR FUTURE CARE

[1]Jessica Russell, [1]Elaine Chan, [1]Garry Rendle, [2]Jo Wray. *[1]Great Ormond Street Hospital for Children NHS Foundation Trust, UK; [2]Great Ormond Street Hospital for Children NHS Foundation Trust and University College London, UK*

10.1136/bmjpo-2023-GOSH.3

Background The COVID-19 pandemic, the GOSH hospital construction programme and industrial action in 2023 have changed aspects of care for children treated for sleep disordered breathing (SDB) using non-invasive ventilation (NIV). This patient population continues to increase, supported by a hospital-based specialist NIV team and has a wide range of clinical conditions, high levels of co-morbidities and medical complexity. This paper presents the findings from two studies asking parents about their child's NIV care: a survey from the NIV Adherence Study, conducted in 2020 and preliminary results from interviews with parents and children and young people (CYP) during the ongoing SPIRITUS study.

Method All eligible parents were invited to complete a survey; data were analysed using descriptive statistics and non-parametric tests. A sample of parents and children were invited to participate in semi-structured interviews, using a sampling matrix. Verbatim transcripts were analysed using framework analysis. Parents and CYP were asked to identify potential interventions that could help their child use NIV, changes to their medical device and NIV service.

Results 125 parents (49% response rate) completed a survey and n=4 parents and n=3 parents and children completed a semi-structured interview. Parents' recommendations focused on improving their ability to care for their child at home, rather than increasing hospital based care or bringing hospital staff into the home. Parents suggested bespoke masks, remote monitoring, troubleshooting ventilator issues and ventilator settings changes, as well as device improvements and automated machine parts delivery. Parents would also benefit from sharing and hearing other families' experiences with NIV.

Conclusion Parents' recommendations focused on reducing some of the known side effects of NIV, keeping their child out of hospital, reducing human error and streamlining medical information from their child to the hospital's NIV team. This is in line with the virtual wards concept.

Acknowledgements for funding or support The NIV Adherence Study was funded by the GOSH Children's Charity. The SPIRITUS study is funded by the National Institute for Health and Care Research (NIHR) under its Research for Patient Benefit (RfPB) Programme (Grant Reference Number NIHR204106). The views expressed are those of the author(s) and not necessarily those of the NIHR or the Department of Health and Social Care. This work is supported by the NIHR GOSH BRC. The views expressed are those of the author(s) and not necessarily those of the NHS, the NIHR or the Department of Health.

76 DEVELOPING TRANSFER OF CARE PATHWAYS FOR HEALTHCARE TRANSITION IN ENDOCRINOLOGY

Alexander Chesover, Catherine Peters. *DRIVE, Great Ormond Street Hospital for Children NHS Foundation Trust, UK*

10.1136/bmjpo-2023-GOSH.4

'The Inbetweeners', a National Confidential Enquiry into Patient Outcome and Death (NCEPOD) review, concluded that the transition and transfer of care for young people with complex care needs from paediatric to adult health services is unclear and fragmented. Transition is a priority for the endocrine department at GOSH. We initiated a quality improvement project to address young people's needs with complex endocrine health conditions in healthcare transition.

Our weekly 'Big Room' service development time is a forum for team working and to engage stakeholders and collaborators. First, we agreed a shared vision: 'safe, effective, and coordinated healthcare transition for endocrine patients that is patient-centred and meets young people's individual needs'. Next, four objectives aligning to our vision and the NCEPOD review recommendations.

One objective, to 'define and monitor transfer of care pathways', was our first focus. We assessed (1) current practice – what we do well and the challenges; (2) endocrine patient cohorts requiring pathways; and (3) with which adolescent and adult services we should partner.

Developing each pathway was an iterative process. We collaborated and 'tested' the pathway's feasibility with sub-specialists, clinical nurse specialists, and external adolescent and adult health teams. We created 19 transfer of care pathways for endocrine conditions. These show the recommended referral age; transition multidisciplinary teams (MDT), adolescent service, and adult service; and hospital and named consultant or clinic.

Outcome measures will include monitoring patient numbers above a certain age in our endocrine clinic. We created a report in Epic (our electronic patient record) to extract this data. Other outcome measures will be feedback from partners in adolescent and adult health services and feedback from young people. We are currently surveying young people at GOSH and after their transfer of care, which will also inform our next healthcare transition focus: the creation of endocrine-specific and patient-centred resources.