developed methaemoglobinaemia as a result. Initial methaemoglobin was 48% which subsequently improved to 3% following 3 doses of methylene blue with good clinical response. Further investigations excluded Systemic lupus erythematosus and Crohn's disease, which have been reported as associated with PD. The infant's recovery was supported with parental nutrition and discharged home with ongoing follow up with Paediatric metabolic specialists.

Management of PD is mainly supportive with consideration of use of co-factors that can improve collagen stability, suppression of collagenase and topical application of ointments containing L-proline. Enzyme replacement is also being actively researched.

This case illustrates the importance of prompt recognition and treatment of methaemoglobinaemia in infants with unexplained metabolic acidosis. It also highlights awareness of PD, which although, is extremely rare, can be diagnosed through metabolic screening tests and genetic testing, thereby leading to earlier supportive management.

167 DEVELOPMENT OF AN ACUTE GENERAL PAEDIATRICS SERVICE SPECIFICATION AT A MAJOR LONDON TEACHING HOSPITAL

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10.1136/bmjpo-2021-RCPCH.91

Background Acute general paediatrics is responsible for the care of children (0–18) presenting acutely to hospital, usually through either the emergency department (ED) or following a GP consultation. In the acute setting patients may be looked after in the emergency department, paediatric assessment units, ambulatory or day care facilities, inpatient wards, operating theatres, intensive care units and outpatient clinics. In a teaching hospital setting, general paediatricians may also have a role co-ordinating care for complex children.

In the last five-ten years, standards from national and regional bodies govern the care provided to patients by general paediatricians. Services and individual practitioners can be called upon to demonstrate how they meet these standards and evidence the quality of care provided, such as at CQC inspection. Relevant documents, however, may be located in multiple places and are not always easily accessible to staff or adequately monitored as part of service governance.

Objectives

- To derive a practitioner focused service specification based on relevant acute paediatric care standards
- To determine local compliance against standards and identify measures needed to achieve compliance
- To plan a robust, practitioner-led methodology for ensuring ongoing compliance and monitoring

Methods Relevant guidance was identified through a variety of search methods: national bodies' websites, expert opinion and liaison with Trust managers to identify standards reported to regulators.

Standards were collated and reviewed amalgamating any areas of duplication and removing those not relevant. The final list was thematically analysed into key areas of practice. An expert group consensus methodology was then used to rate the service performance against these standards.

Results 65 standards of care were identified from six key documents. Thematic analysis identified nine main themes and 16 subthemes.

Using a Red, Amber, Green (RAG) rating system to identify the services' performance against each theme, an action plan was then created, using a plan, do, study, act (PDSA) process to achieve these standards that were then linked to the service governance strategy and to consultant action plans.

It was decided to audit one of the standards with an amber status to assess if it were being met and, if not, why not. There was a 66.6% compliance, monitored across three paediatric wards, and five themes were identified as reasons for it not being met.

Following on from the data collected, further standards not being met will be explored and their Action Plans will continue to be put into place with their progress monitored regularly within departmental governance meetings.

Conclusions The new service specification is used by clinical staff to reflect on practice and outcomes, allowing easy access to relevant standards to guide clinical practice and service development and quality improvement in a strategic and co-ordinated way. The document is a 'living' document, accessible to all clinical team members and external stakeholders.

It will aid planning, commissioning and provision of acute paediatric services and provide a framework against which to audit provision and demonstrate improvement. A similar methodology could be used across the UK.

169 USING NETWORK ANALYSIS TO FOLLOW MENINGOCOCCAL SEPTIC SHOCK PATHOGENESIS IN INFANTS

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10.1136/bmjpo-2021-RCPCH.92

Background Previously we reported the complex nature of the transcriptome in Meningococcal sepsis (MenS) limiting the detection of a representative biomarker from a single time point for a dynamic disease process [1]. In this study, alleviating that limitation, we utilised network methodology to the transcriptome of MenS patients obtained from multiple time points during disease progression.

Objectives

Methods We applied weighted gene co expression network analysis (WGCNA) [2] on normalized expression data of 5 children (P1-P5) with MenS (4 confirmed and 1 clinically assumed) who had blood samples taken at admission (designated 0 hours), 4, 8, 12 and 48 hours (T1-T5). The patients had no previous, comorbidities. We extracted RNA from blood and then the samples were checked by capillary electrophoresis and spectrophotometry for quality control purposes. We conducted microarray (Human Gene 1.0 ST Arrays with 33,297 probes) gene expression experiments to capture the transcriptome profile at each time point.

Results Our initial differential gene analysis between all-time points against T1 showed significant ($p < 1.0 \times 10^{-7}$) up regulation of protein binding and immune response activities. Cluster map was compiled (figure 1) and then this was matched to trait data (figure 2). (Next, applying WGCNA analysis we identified 18 cluster (or modules) that have distinct topological characteristics (sizes ranging from

34–14941 to genes). For each module, we defined a quantitative measure of module membership as the correlation of the module eigengene with the gene expression profile. Modules were phenotypically stratified based on weight, age, mortality, and organ dysfunction. A heatmap plot was generated of the adjacencies in the eigengene network (figure 3). Genes from the significant modules showing high module membership were filtered and selected ($p_{MM} \leq 0.05$). Number of Genes related to trait included 740 (mortality) 2151 (weight) and 1616 (age). Weight was correlated high (r 0.53 and p value 0.003) with the purple module (MEpurple) to weight (figure 4).

Conclusions Our analysis shows that using the time series transcriptome in MenS, novel associations can be identified that could influence future treatment options for improved outcomes.

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EXPANDED CARRIER SCREENING: PRIMARY PREVENTION OF RECESSIVE MONOGENIC DISEASES EVALUATED USING 1,909 CHINESE GENOME AND EXOME SEQUENCING DATA

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10.1136/bmjpo-2021-RCPC.93

Background Expanded carrier screening (ECS) is a genetic test that investigates the genetic composition of a couple and determines whether their offspring has an elevated risk of inherited disorders. Comparisons between commercially available ECS has, however, only shown small overlaps in common genes offered for screening.

Objectives Compiled with the inadequate information surrounding carrier frequencies in the Chinese population, secondary usage of next generation sequencing could be used in the optimization of ECS panels surrounding clinical utility, public health benefits, and reducing unnecessary socio-psychological stress.

Methods In this study, a total of 1543 Southern Chinese and 366 Northern Chinese genome and exome sequencing were screened for carrier status over 315 genes. The gene list curated for this study was compiled from three ECS panels offered by frequently used commercial companies and literature reporting high carrier frequencies for treatable inherited disorder in South East Asia population genomics.

Results 180 unique disease-causing variants were identified and 47.8% ($n = 738$) of Southern Chinese individuals screened in this study harboured at least 1 disease-causing variant. CNV calling determined 4 unique pathogenic or likely pathogenic copy number variants. A total of 285 unique carrier variants were classified as pathogenic or likely pathogenic. Results have also identified 12 genes with a carrier frequency over 1% including GJB2, HBA1/HBA2, SMN1, SLC22A5, SLC25A13, ATP7B, SLC26A4, GALC, POLG, USH2A, and HBB.

Conclusions This study shows that secondary analysis of NGS data can illustrate the carrier frequencies in the Southern Chinese population. Through the comparison of different commercially available ECS panels, we identified potential for improvement in the optimization of commercially available ECS panels for the future of precision medicine.

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DISCHARGE MANAGEMENT OF CHILDREN UNDER 5 WITH SUSPECTED ASTHMA IN A UK HOSPITAL

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10.1136/bmjpo-2021-RCPC.94

Background The British Thoracic Society (BTS) recommends that children under 5 years of age who are admitted with suspected asthma should be discharged as per the BTS Asthma Discharge Care Bundle since this improves patient outcomes¹. Many children with suspected asthma are prescribed inhaled corticosteroids (ICS) to reduce airway inflammation². We suspected substandard discharge care of children under 5 hence decided to audit our practice.

Objectives

1. To assess compliance with the BTS asthma discharge bundle, for children under 5 admitted with suspected asthma, who were discharged on inhaled corticosteroids.
2. To improve and standardise the discharge planning of this group of children.

Methods Children with a discharge diagnosis of viral induced wheeze (VIW) or asthma, between 01/12/2018 and 31/11/2019, were provided by the hospital information department. Patient notes on iSOFT electronic patient record system were reviewed and those discharged on ICS were identified. From these, every other case in 'date of admission order' was selected and audited against the BTS bundle. This retrospective audit was approved by the trust.

Results Out of a total of 447 children discharged with VIW or asthma under the age of five, 123 patients were discharged on ICS and of these 62 were randomly selected for audit.

Only 9/62 (15%) patient records were 100% compliant with the BTS bundle. 22/62 (35%) were less than 50% compliant. 17/62 (27%) did not have any follow up (neither hospital nor community).

Abstract 172 Table 1

BTS discharge care standards	Compliance (n=62)
Community follow up within two days	13 (21%)
Importance of adherence to preventer medication discussed.	19 (31%)
Triggers considered- including smoke exposure.	24 (39%)
Specialist care follow up in a month.	27 (44%)
Inhaler technique assessed.	32 (52%)
Consultant appointment in 2–3 months.	41 (66%)
Inhaler use instruction provided.	50 (81%)
Medications reviewed prior to discharge.	55 (89%)
Written action plan provided.	59 (95%)

Conclusions The BTS discharge standards for children under 5 years with suspected asthma were not followed in the majority of children. The lack of follow up of many children who were discharged on ICS is particularly concerning, potentially sentencing them to long term ICS and consequent risk of side effects.

We recommend that shortfalls identified by this audit be addressed followed by re-audit after a year.