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A RETROSPECTIVE STUDY OF MYELOID LEUKAEMIA IN CHILDREN WITH DOWN SYNDROME IN IRELAND

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Background Haematological abnormalities are common in children with trisomy 21. These children have a remarkably high risk of acute leukaemia. The incidence of acute myeloid leukaemia is 150 fold greater in young children with DS compared to children of the same age without DS. Acute Megakaryoblastic Leukaemia (AMKL) is a subtype of myeloid leukaemia and is the most common leukaemia type in children with DS under 4 years of age. AMKL is often preceded by a transient neonatal pre-leukaemic syndrome, called Transient Myeloproliferative Disorder (TMD). Although TMD often spontaneously resolves, 20–30% of these patients subsequently develop AMKL within the first 4 years of life.

Aims To perform a retrospective consecutive national audit of all documented cases of childhood TMD and AMKL-DS from 1990–2018 at the National Paediatric Haematology/Oncology Centre, Our Lady's Children's Hospital Crumlin (OLCHC) Ireland. We also aimed to compare our demographic and outcome findings with the recent medical literature and make recommendations for future research.

Methods Charts of the patients with a diagnosis of AMKL and treated consecutively at (OLCHC) between 1990 – 2018 were sourced from medical records. Charts and the hospital database were reviewed and information including date and age of diagnosis of AML, comorbidities, previous history of TMD, treatment regime and outcome was noted. The Hospital Patient Administration System (PAS) was interrogated for additional information such as percentage blasts in the bone marrow and blood count values at time of diagnosis of AMKL. Cytogenetic information was obtained by accessing a dedicated database. Kaplan-Meier survival curves were constructed.

Results Twenty-seven patients with AMKL-DS were treated in OLCHC. A prior neonatal diagnosis of TMD was described in 10 patients (37%). Patients had a low platelet count (median $34 \times 10^9/L$) at presentation. Nineteen patients (70%) are alive and well, in complete remission, at a median follow up of 11.4 years. Overall survival (OS) of this cohort has risen from 54% from those treated between the years 1990 – 2004 ($n = 13$) to 93% for those treated between the years 2005 – 2018 ($n = 14$).

Conclusion High cure rates are observed in AMKL-DS using current polychemotherapy protocols. However, in general children with DS are more prone to the cytotoxic effects of chemotherapy, which can lead to significant treatment related mortality. The finding of a low platelet count at time of diagnosis is in keeping with the knowledge that AMKL-DS is a malignancy of platelet progenitor cells.

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114 CASES OF CHRONIC GRANULOMATOUS DISEASE IN MAINLAND CHINA

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Background Chronic granulomatous disease (CGD) is a rare inherited disorder, with phagocytes failing to produce antimicrobial superoxide due to deficient NADPH oxidase activity. Mutations in the gene encoding CYBB are responsible for the majority of the CGD cases. In this study, we described the clinical and molecular features of 114 suspected CGD patients in mainland China.

Methods During 2013 to 2018, we enrolled 114 cases diagnosed with CGD in Children's Hospital of Chongqing Medical University. Patients' clinical data were collected, neutrophil function was detected, and the genotype of patients and fetal cells in the carrier mother's amniotic fluid were detected.

Results 114 CGD patients (109 male, 5 female), from 20 provinces of mainland China. The mean age at onset was 4 months, and the mean age of diagnosis was 23.1 months. To date, 33 patients have died, 27 patients have hemopoietic stem cell transplantation, and 12 patients were lost. For all patients, the types of genetic mutations include CYBB (95, 83.3%), CYBA (8), NCF1 (3) and NCF2 (2), and 6 cases have not been found yet. The type of infection symptoms in this cohort included recurrent fever (100%), pneumonia (92.1%), tuberculosis (58.8%), BCG disease (55.1%; 54/98), repeated diarrhea (45.6%) and others. In addition, we prenatally diagnosed 8 XL-CGD, 6 healthy fetuses and 1 carrier (a XL-CGD female infant patient with X-chromosome inactivated).

Conclusions Children with recurrent bacterial and fungal infection not easy to cure, with higher inflammatory markers, with or without BCG disease and family history, should be alert to CGD. BCG disease and tuberculosis is still a very severe problem in CGD patients in China. Early diagnosis and prophylaxis treatment can prolong the survival time of CGD patients. DHR-1,2,3 test and NBT test can quickly aid in the diagnosis of CGD. Molecular analysis is an important tool for identifying diagnosis, prenatal diagnosis of CGD.

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INTRAOPERATIVE NERVE INTEGRITY MONITOR AS PART OF SAFETY DISSECTIONS OF RECURRENT LARYNGEAL NERVE IN CHILD AND ADOLESCENTS THYROID SURGERY

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Introduction In thyroid cancer (TC) operations most frequently damaged vital structures are recurrent laryngeal nerves (RLN), external branch of superior laryngeal nerves and parathyroid glands.

Material and methods To improve our knowledge of RLN surgical anatomy we investigated autopsy material (30 RLN), photo documented 1717 RLNs. There are about 30 variations of RLN and inferior thyroid artery attitude and three most common points of RLN visualization They are: 1 – subclavian point known as Lore's triangle, 2 – RLN 'cross point' with the inferior thyroid artery and 3 – RLN laryngeal entry point. After anatomical investigations we used the Intraoperative Nerve Integrity Monitor (IONIM) during thyroid operations (2001–2016) in adults (1791), adolescents and children (117 cases).

Results It was found that RLNs in 3rd point were crossed by blood vessels in 94.9% (Romanchishen F.A., 2006). Ligation

and transection of those vessels was very important, because thyroid tissue removal between the trachea and RLN became more convenient. This technique guaranteed safety of the RLN. Our technique of thyroidectomy allowed to find and save 15 (0.37%) nonrecurrent laryngeal nerves among 4070 thyroid patients. Postoperative laryngoscopic control allowed estimating the postoperative laryngeal palsy rate objectively. In 21 cases RLN was restored with the perineural suturing under the IONIM control. During 1–2 years postoperatively 15 (71.4%) demonstrated vocal cord function improvement. To protect SAN during lateral neck dissection the procedure of its surgical separation was specified. The best place for the first nerve visualization (because of safety) was the level of sternocleidomastoid muscle's upper third where it accompanied the internal jugular vein. Our technique assumes dissection of SAN from above with separating and saving of nervous branches to sternocleidomastoid muscle's, C3 and TM with consecutive moving of SAN from inner to the outer part of the sternocleidomastoid muscle's by the rubber holder and forceps.

Conclusion As a result of regular use of suggested RLN, SAN dissection and IONIM technique, the postoperative morbidity rate decreased significantly.

OC55 INHIBITORS IN ALBANIAN CHILDREN WITH HEMOPHILIA A

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Objective To identify the data related with therapeutic and patients characteristics and their correlation with the inhibitor's presence in hemophilia A in Pediatric Hematologic/Oncologic Unit. University Hospital Centre 'Mother Teresa', Tirana, Albania, the unique national service that treat pediatric coagulopathies.

Design This analytic, observational, retrospective and case – control study analyses 32 patients (0–14 years old) diagnosed and treated in our center for hemophilia A, that had been tested for inhibitor presence (32 from 45 children with hemophilia A in Albania).

Outcome measures Inhibitor's presence (qualitative test) after 50 days of exposure, severity of the disease, breastfeeding, family history for hemophilia, age and purpose of first FVIII exposure, age of 50th exposure, time between first and 50th exposure, surgery up to 50th day of exposure, type and number of exposures episode (minor or major) and FVIII average dosage.

Results Inhibitors were present in 13 from 32 patients. The mean age of the first exposure to FVIII is 20.1 months old and in 25 of 32 patients hemorrhage was the reason for the first exposure. There was no significant statistical difference between inhibitors presence and age and reasons of first exposure. Neither nor the age of the 50th exposure wasn't correlated with inhibitors presence. The mean age between first and 50th day of exposure was 24.2 months and there was a positive correlation between this data and inhibitors presence (t -Test = 2.267; df = 30; p = 0.031), also the number of episodes of exposition until the 50th day of exposition (avg. 6.5) was correlated with inhibitors presence (t -Test = 3.270; df = 30; p = 0.003). We had more major exposure

episodes (78.1%) and this data influenced in inhibitor's presence (Fisher's Test = 6.131; df = 1; p = 0.015). The average dosage of FVIII was also correlated with inhibitor's presence (χ^2 = 12.799; df = 2; p = 0.002). Severity of the disease and family history for hemophilia weren't correlated with the inhibitors. Breastfeeding with associated with lower risk for inhibitors presence (t = 7.166; df = ; p = 0.011).

Conclusions Inhibitor's presence percentage in Albanian pediatric patients with hemophilia A is 40.6%. This data is much higher than the literature reports. Factors influencing more in the development or presence of inhibitors in our population are: high dosage of factor VIII, the high frequency of exposure episodes and the short time between first and the 50th exposure day.

OC56 HYPERTHERMIC INTRAPERITONEAL CHEMOTHERAPY FOR TREATMENT OF DESMOPLASTIC SMALL ROUND CELL TUMOURS IN A CHILD; A U.K. FIRST

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Introduction Desmoplastic small round cell tumours are rare, aggressive neoplastic tumours with a predisposition to affect the intra-abdominal cavity.

Hyperthermic intraperitoneal chemotherapy (HIPEC) is a mode of delivering concentrated, heated chemotherapy directly into the abdominal cavity. This has been used previously in adult patients and we describe the first UK case of HIPEC for a desmoplastic small round cell tumour in a child.

Case summary An 8 year old girl (AB) was diagnosed with desmoplastic small round cell tumours after presenting with a ten day history of constipation and abdominal discomfort. Initial assessment revealed a large palpable abdominal mass. An ultrasound demonstrated a large heterogenous mass with central cystic necrosis. Two additional lesions were noted within the liver in addition to a 22 mm omental deposit. Bone marrow aspirates and biopsies confirmed the diagnosis.

AB had eleven rounds of chemotherapy in total with the most recent cycle completing in April 2018. A follow-up MRI showed a residual pelvic tumour of 3.1cm indenting the superior wall of the bladder.

After MDT discussion the decision was made to proceed with macroscopic resection of tumours followed by cisplatin HIPEC. Intraoperatively, lesions were noted in the following sites: omentum; pouch of Douglas; left ovary and fimbriae; jejunum and diaphragm. After resection was completed, cisplatin heated to 41 degrees C was washed over the peritoneum. This was tolerated with noradrenaline used to optimise haemodynamics. Postoperatively she was admitted to PICU for monitoring and observation.

During PICU admission AB remained clinically stable. Active interventions included GCSF, thoracic epidural and PCA for analgesia, electrolyte optimisation and hyperhydration. AB was ready for discharge from PICU after 4 days.

Conclusion We described the first case of HIPEC being used in paediatrics in the UK. This technique has been well tolerated and could be considered in other cases of intra-abdominal neoplasia with metastases.