Abstracts

613 CONGENITAL DIAPHRAGMATIC HERNIA SURVEILLANCE IN IRELAND
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1K Thorup, 2V Monk, 3E Gourlay, 4N Aiton. 1Brighton and Sussex Medical School; 2Brighton and Sussex University Hospitals NHS Trust, Brighton, UK

Background Fetal Alcohol Syndrome (FAS) comprises a triad of growth impairment, central nervous system dysfunction and characteristic facial features. Diagnosis is complex and often not recognised at an early age. The three facial features: short palpebral fissures, smooth philtrum and thin upper lip, are unique to FAS. Clinical examination is inherently subjective and apart from palpebral fissure length, minimal reference data is available in neonates. Establishing a standardised method and normal range would promote an objective assessment. Earlier diagnosis would enable earlier effective interventions.

Methods Standardised digital facial photographs were taken of normal term Caucasian neonates. Mothers completed anonymous questionnaires about alcohol consumption during pregnancy. Photographs were assessed using Facial Analysis Software to obtain values for palpebral fissure length (PFL) and upper lip circularity (LC). Upper lip thinness and philtrum smoothness were ranked according to 5-point Likert Scale.

Results 29 infants were studied, 17 male: 12 female. Mean gestational age 40.3 weeks (range 37.1–42.3), mean weight 3559 g, 23 (79%) had no prenatal alcohol exposure whilst 6 had minimal exposure (1–2 units/week). PFL measurements could be obtained from 21 photographs (72%) with mean of 15.6 mm (range 13.7–18.7 mm). Upper lip and philtrum values could be determined in 24 (83%). Mean LC was 57.21 (range 31.4–128.2). Mean rank scores for upper lip and philtrum were both 3.

Conclusion It has been possible to gain measurements of facial features in just over ¾ of neonates studied, showing the feasibility of this technique in this age-group. Further results are needed to establish reference ranges.

614 THREE SIBLINGS WITH NEONATAL PRESENTATION OF GRISCELLI SYNDROME
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BR Qandalji. Pediatrics, Ministry of Health, Amman, Jordan

Introduction Griscelli syndrome type 2 is a rare disorder characterised by pigment dilution (silvery hair), variable immune deficiency, and tendency to develop a life threatening hemophagocytic syndrome. Presentation in neonatal life is even more rare.

Materials and Methods These are three siblings to first cousin parents who presented with Griscelli syndrome in the neonatal life with desemel outcome.

Results J, A, S, are three siblings, two sisters and one brother born to first cousin parents. All had normal vaginal full term delivery. All presented on day one of life with pallor, silvery hair, and hepatosplenomegaly. Investigations revealed variable degrees of anemia & thrombocytopenia. BM revealed hemophagocytic syndrome. There was no HLA matched BM donor so they were treated conservatively. They had repeated admissions for infections and received several blood and platelet transfusion. They had strom courses and died by two months of age.

Conclusion This is probably the largest series of Griscelli syndrome presenting in neonatal life. BM is the only hope for this syndrome.

A thorough family history is always helpful in diagnosing difficult cases.

615 CONGENITAL LUNG MALFORMATIONS PRESENTING WITH SIMILAR CHEST X-RAY CHANGES AT BIRTH
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1G Nepali, 2M Dattani, 1S Somisetty. 1Neonatal Unit, Luton and Dunstable Hospital NHS Foundation Trust; 2Radiology, Luton & Dunstable Hospital NHS Foundation Trust, Luton, UK

Con genital defects of diaphragm or malformations of lung usually present in first few hours of birth and if left undiagnosed can lead to significant morbidity and mortality. Early accurate recognition is paramount for subsequent management.

Aim To highlight cases presenting to tertiary neonatal unit with respiratory distress on admission and similar chest x-ray changes.

Case 1 Term infant, one of dichorionic diamniotic twins with antenatal history of polyhydramnios admitted with respiratory distress soon after birth. Initial Chest X-ray (CXR) showing homogeneous opacification of left hemithorax with mediastinal shift to right. Subsequent CT chest revealed Bronchogenic cyst.

Case 2 Term infant admitted with respiratory distress. Initial CXR showed homogeneous opacification of left hemithorax with mediastinal shift to right. Subsequent CXR revealed left sided diaphragmatic hernia.

Case 3 Premature infant born at 34 weeks of gestation with multiple congenital anomalies. Initial CXR showed almost complete opacification of right hemithorax. Chest ultrasound was suggestive of severe right sided diaphragmatic entervation.
Methods

Defects are unknown.

Results

ALRI hospitalisation rates among children with various birth defects (OR 2.29, 95% CI: 1.89, 2.78; IRR 2.00, 95% CI: 1.84, 2.17 respectively). They had a birth defect than children with no birth defects (IRR 3.21, 95% CI: 2.36, 4.39).

Conclusion

Aboriginal children were more likely to be hospitalised for ALRI if they had a birth defect than children with no birth defects (IRR 2.96, 95% CI: 2.39, 3.69).

Methods

We conducted a retrospective cohort study of 245,249 singleton births in WA (1996–2005). Population-based hospital morbidity data and the WA Register of Development Anomalies were linked through the Western Australian Data Linkage System to investigate ALRI hospitalisations in children with and without birth defects. We used negative binomial regression to estimate incidence rate ratios (IRR) for the association between birth defects and number of ALRI hospitalisations over the first 2 years of life, adjusting for known risk factors.

Results

Overall 11% of non-Aboriginal children and 40% of Aboriginal children with birth defects had a least one ALRI admission before age 2 years. In adjusted analyses, Aboriginal and non-Aboriginal children were more likely to be hospitalised for ALRI if they had a birth defect than children with no birth defects (IRR 2.29, 95% CI: 1.89, 2.78; IRR 2.00, 95% CI: 1.84, 2.17 respectively).

Conclusions

WA children < 2 years with birth defects are at greater risk of morbidity due to ALRIs, when compared to children with no birth defects. Risk of ALRI hospital admission varies between different birth defect categories.

Introduction

Although uncommon, congenital lobar emphysema (CLE) is a potentially life threatening pulmonary abnormality affecting infants. Lobectomy, sometimes done under emergency conditions, is the universally accepted treatment of CLE with severe symptoms. However, in the developing world trained pediatric thoracic surgeons are not everywhere available. On the other hand, the use of flexible fiberoptic bronchoscopy (FFB) in children has shown an excellent safety record. Furthermore, it can be performed under sedation in most cases. We present here a case of CLE, in which we were able to relieve the acute respiratory distress using FFB.

Case presentation and procedure

A distressed 4.5 months old female infant was referred to us for repetitive prolonged bronchopneumopathy since birth. Successive chest X-rays showed increasing expansion and hyperinflation of the right hemithorax, mediastinal shift, and compression of the left lung. At FFB the apicoposterior segmental bronchus of the right upper lobe (APSB/RUL) was narrow, flaccid and showed an expiratory check valve obstruction. By manipulating and rotating the bronchoscope tip into APSB/RUL and applying suction, we had succeeded to release the trapped air in the RUL. Clinical and radiological manifestations resolved completely following the procedure.

Conclusion

Flexible bronchoscopy in the acute management of congenital lobar emphysema is a safe and effective technique in selected cases.

Aim

The aim of this study was to report an outbreak of Neural Tube Defects (NTDs) in Iraq, and to highlight the possible responsibilities of international and local authorities for an action for the control of this outbreak in this region.

Methods

Information for the occurrence of NTDs was gathered from reports published from an Iraqi western region, and from other countries from reports published by the International Clearinghouse for Birth Defects and European Network for Surveillance of Congenital Anomalies.

Results

Prevalence rate of NTDs was 33 (per 10,000 births, CI95%: 21–44) in Iraq while different rates have been reported from various parts of the world ranging from 12.6 (per 10,000 births) in Cuba, 9.6 (per 10,000 births) in Norway, 8.7 (per 10,000 births) in China, 7.05 (per 10,000 births) in Canada, 3.03 (per 10,000 births) in Canada, and 1.7 (per 10,000 births) in the United States.

Conclusion

In addition to the role it might play in the diagnostic workup of patients with CLE, FFB can be used as a tool for relief of obstruction. In certain situations, this may be life saving, especially in places and settings where emergency lobectomy cannot be arranged.