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Audit of Adherence to Best Practice Guidelines for Children born with Down Syndrome

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Abstract

Aim

The aim of this audit is to determine adherence to the Down Syndrome Medical Interest Group (DSMIG) Neonatal Best Practice Guideline in a tertiary neonatal unit over a 5-year period. Our secondary objective was to describe patterns in the demographics, admissions and clinical course.

Methods

This is a retrospective audit of infants with a diagnosis of DS between 2018 and 2022 in Cork University Maternity Hospital (CUMH). Ethical approval was obtained prior to data collection. Demographics and clinical data were obtained from the electronic health records (EHR). Data was collated and analysed using a password encrypted Microsoft excel dataset. Continuous data was reported as mean or median. Categorical data was outlined as counts. Dichotomous variables were summarised as proportions. A Chi Squared Test and Fishers exact test were used to assess dichotomous variables.

Results

73 patients were included in this audit of which 37 (51%) were female. The median gestation was 38 and 3 days and median birthweight was 3100g. 60 (82.2%) required admission to NICU with 43 (58.3%) admitted immediately after birth. Median hospital stay was 10 days. 8 (11%) had growth parameters documented during their admission. 66 (90%) of patients had an ECHO performed (63 (95.4%) abnormal). 19 (26%) were exclusively breastfeeding and 7 (9.7%) required feeding support on discharge. All babies underwent screening for haematological and thyroid abnormalities, but 13 (18%) babies had unnecessary bloods taken for serum TFT levels. Referrals were made for audiology (60, 82.2%), ophthalmology (17, 23.3%) and other relevant healthcare professionals.

Discussion

This audit shows good adherence to the DSMIG guideline in areas of cardiovascular and haematological monitoring but indicates a need for improvement in other categories.

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Introduction

In Ireland, Down Syndrome (DS) affects 1 in 444 births.[1] DS effects most organ systems in the body which may result in predictable complications in the neonatal period including congenital cardiac disease, feeding difficulties, congenital hypothyroidism and transient abnormal myelopoiesis.[2] Early diagnosis of health conditions that affect people with DS can result in improved quality of life and prolong life expectancy.[3]

The Down Syndrome Medical Interest Group (DSMIG) Neonatal Best Practice Guideline for infants with DS was developed to provide a standard of medical surveillance for children with this condition in the UK and the Republic of Ireland (ROI).[4]

The primary objective of this audit was to determine adherence to the DSMIG Neonatal Best Practice Guideline for infants born with DS in a tertiary neonatal unit over a 5-year period and to potentially improve outcomes for this vulnerable population. Our secondary objective was to describe patterns in the demographics, admissions, and clinical course of the population of babies with DS born in the tertiary unit.

Methods

This retrospective audit of infants with a diagnosis of DS, was conducted in Cork University Maternity Hospital (CUMH) over a 5-year period, from January 2018 to December 2022. Ethical approval was obtained from University College Cork Clinical Research and Ethics Committee (CREC) and the Local Information Governance Group (LIGG) for audit application prior to collection of data. Consent was not required for the purpose of this retrospective audit.

Neonatal demographics and clinical data were obtained from the electronic health records (EHR) available on the Maternal and Newborn Clinical Management System (MN-CMS). The following variables were recorded: gestation, birth weight, Apgar scores at 1 minute and 5 minutes, mode of delivery, need for resuscitation at delivery, antenatal versus postnatal diagnosis, and mode of feeding. Admission to the NICU versus the postnatal ward and duration of admission were also recorded. Delayed admission was defined as any infant admitted beyond 4 hours of age.

We assessed adherence to the DSMIG Neonatal Best Practice Guideline in the following areas: growth parameters recorded on the DS growth chart, cardiovascular assessment (echocardiogram, ECG), thyroid function assessment (TFTs, newborn bloodspot screening), haematological assessment (FBC), referral to audiology and referral to ophthalmology services. We also documented any gastrointestinal morbidities.

Referrals to medical social work (MSW) and physiotherapy services were documented, as well as the time to the first outpatient appointment with a neonatologist.

Data was collated and analysed using a password encrypted Microsoft excel dataset. Continuous data was reported as mean (standard deviation) or medians [interquartile range], as appropriate. Categorical data was outlined as counts (%). Dichotomous variables were summarised as proportions and displayed in contingency tables where appropriate. A Chi Squared Test was used to assess dichotomous variables. Fishers exact test was utilised for the assessment of dichotomous variables when the counts in one or more cells had an expected frequency of five or less (for 2 by 2 tables).

Results

Demographics

There were 73 live born infants with DS identified by the MSW department between 2018 to 2022. This yields an incidence rate of approximately 2 per 1000 live births over these 5 years in CUMH. The demographic data of our group is displayed in Table 1.

The median age of delayed admission from the PNW was 21 hours. Those requiring immediate admission to the neonatal unit had a longer duration of stay than those with a delayed admission (17 days versus 7 days).

7 babies required a transfer to another tertiary paediatric centre (4 for cardiac lesions, 2 for duodenal atresia and 1 for necrotising enterocolitis).

Variable	DS Group (n= 73)
Female, n (%)	37 (51%)
Gestational age, median [IQR]	38+3 weeks [37+2- 39+5]
Birthweight grams, median [IQR]	3100 [2696-3415]
Postnatal diagnosis, n (%)	47 (64%)
Delivery by Caesarean section, n (%)	42 (58%)
Emergency Caesarean section, n (%)	12 (16%)
Apgar scores, median [IQR] - 1 minute - 5 minutes	 8 [7-9] 9 [8-10]
Admission to NNU, n (%) - Immediate admission	61 (84%) 36 (50%)

- Delayed admission (>4 hours of age)	25 (34%)
Duration of admission, median [IQR]	
- Immediate admission	17 days [8 - 27]
- Delayed admission	7 days [3 – 11]
Transfer to another unit, n (%)	7 (9.5%)
- Cardiac	4 (5%)
- GI	3 (4%)

Table 1: Demographic and Admission data

Growth and Feeding

Only 8 babies (11%) had their growth parameters documented on the DS Growth Chart. 19 (26%) babies were exclusively breastfed and 45 (62%) received term formula via a bottle, of which 2 required a high calorie formula. 5 babies were combination fed with term formula, of which one required a high calorie formula. One baby was fed enterally via a nasogastric tube (NGT) and 3 babies were bottle fed with NGT feeds.

Cardiovascular Assessment

90% of babies had an echocardiogram performed prior to discharge home. The ECHO findings are summarised in Table 2.

Echocardiography	**DS Group (N = 73) N (%)**
Echocardiogram performed	65 (90%)
Normal echocardiogram	3 (4.6%)
PDA and/or PFO alone	12 (18.5%)
Cardiac anomalies detected	n= 50
Pulmonary hypertension	13 (26%)
Atrioventricular septal defect (AVSD)	16 (32%)
Ventricular septal defect	15 (30%) *
Atrial septal defect	27 (54%) *

Other (Transposition of the great arteries)	1 (2%)

Table 2: Echocardiography Findings.

** some infants had both VSD and ASD detected*

Babies with a delayed admission to the NICU were less likely to have a cardiac abnormality on ECHO than those admitted immediately following delivery (16 versus 32, P value 0.02). Two babies had septal defects (ASD and VSD, and VSD alone) diagnosed on ECHO but did not require admission to the NICU.

Thyroid Function Assessment

All babies had a newborn bloodspot screening for diagnosis of congenital hypothyroidism. However, 13 (18%) babies had blood tests send for TFTs.

Haematological Assessment

Every baby had a full blood count (FBC) sent. The majority of babies had normal FBCs. 10 babies had a thrombocytopenia (<100,000 per microlitre). Of these, 7 babies had moderate thrombocytopenia (50-100x10^9/L) and 3 babies had severe thrombocytopenia (<50x10^9/L). Two babies had leukocytosis. No babies had blasts with a leukocytosis seen on their blood films. 1 baby had thrombocytopenia with blasts on their blood film. This baby was referred to a haematologist for management of likely Transient Abnormal Myelopoiesis (TAM).

Audiology

All babies had the Universal Hearing Screen prior to discharge unless they were transferred to another unit. 60 babies (82%) had a formal referral to audiology services either due to an abnormal hearing screen or as a result of their diagnosis of DS.

Ophthalmology

All babies had their red reflexes assessed as part of their discharge examination. 17 babies (23%) had a formal referral to an ophthalmologist prior to discharge.

Other

Two babies had duodenal atresia and required transfer to a tertiary paediatric surgical unit.

56 babies had referrals to a MSW and 54 had referrals to a neonatal physiotherapist documented.

6 babies did not have a follow up plan documented, however 3 of these were transferred to another unit. The majority of infants had follow up planned for 1-2 months after discharge (N=31) while some were seen earlier (N=18) and others by 3 months of age (N=18).

Discussion

The incidence rate of 2 per 1000 live births reported by our study is comparable to other tertiary Maternity centres in the ROI but higher than those quoted for the UK and Europe.[1,4] The high incidence of DS highlights the importance of adhering to a surveillance programme as the morbidities encountered by patients with DS are common but very treatable.[2]

In terms of primary outcome measures, varying degrees of adherence to the DSMIG Neonatal best practice guidelines were observed within each category. Poor adherence to documentation of growth parameters was noted. Evidence shows babies with DS often lose more than 10% of their birth weight and can then struggle to gain weight. Thus, adequate monitoring of growth is essential in providing care to infants with DS.[4] Since carrying out this retrospective audit DS growth charts were incorporated into the EHR. As such a reaudit of adherence to documentation of growth parameters may show more positive results in the coming years.

All babies reviewed underwent newborn bloodspot screening but 13 also had serum TFTs taken. There is a well reported transient physiological rise in Thyroid Stimulating Hormone in the immediate postnatal period in the DS population due to a lower thyroxine level.[7] Therefore, early serum sampling can result in a false positive result for congenital hypothyroidism and can lead to further unnecessary tests.

Nearly all babies (90%) had an ECHO prior to discharge home. The DSMIG promotes an ECHO be performed by 6 weeks of age. We were unable to determine if the remaining 10% of patients had an ECHO within this timeframe as the newborn EHRs do not transfer to the paediatric cardiology department. However, it is standard practice for infants in our unit to undergo an ECHO if they have concerning clinical findings. An American study reviewed postnatal ECHOs in babies with DS who had a reported normal fetal ECHO. Reassuringly, that study demonstrated that no baby had a missed diagnosis of complex congenital heart disease and any baby with a cardiac abnormality noted postnatally had either a murmur or an abnormal ECG.[5] 77% of babies in our cohort had a heart defect of which 32% were AVSDs. The DSMIG surveillance guideline states that it is important to diagnose AVSDs early before the development of irreversible pulmonary hypertension.[4] Furthermore, Al Biltagi et al report that for children with DS congenital heart disease is the highest cause of mortality.[6] In short, although a normal fetal scan in combination with a normal clinical examination is reassuring, obtaining an ECHO prior to 6 weeks of age is optimal.

Every baby in this cohort had an FBC sent in the first days of admission with one baby referred to haematology with suspected TAM. According to Tunstall et al, as many as 5 - 30% of children with DS are born with TAM with a peak incidence at 2 years of age.[8] The survival rate following appropriate treatment can be as high as 90%, thus early diagnosis is key.[4] 11 babies had a documented thrombocytopenia in this cohort. The literature suggests that thrombocytopenia is a common finding in the first week of life for infants with DS, occurring in 11-66%[18, 19]. It is usually mild (>40x10^9) and transient (<2weeks).[18] None of our babies had complications related to thrombocytopenia or bleeding.

All babies had newborn hearing screens performed and were referred to services if results were abnormal. The DSMIG states that all babies with DS should be referred for formal audiology review by 8 months but only 82% of babies in this cohort had a documented referral made.[4]

The standard referral for ophthalmology at birth, according to the DSMIG guideline, is only in cases of abnormalities. 17 babies were documented to have referrals made to ophthalmology in this cohort. These were referred on a basis of their diagnosis of DS rather than any abnormalities found, likely to ensure ophthalmology review by 18 months as recommended by DSMIG.

As this was not a qualitative study it was difficult to ascertain if there was an appropriate level of communication regarding the diagnosis of DS and what conversations were had on the follow up plan. It is clear from this audit that emphasis needs to be placed on ensuring referrals to MSW, physiotherapy and neonatal OPD are complete prior to discharge. Nearly a quarter of infants were not referred to a MSW or physiotherapist and 3 babies who were due to receive ongoing care locally, did not have a follow-up order on their EHR.

Most of the demographics showed a homogenous population with regards sex, birth weight, need for resuscitation and Apgar scores. We noted that the majority of patients were delivered by caesarean section. The incidence of caesarean delivery per total mothers delivered in CUMH in 2021 was 39% but the rate of caesarean delivery in our cohort is much higher. This is not an unreported phenomenon. One study in Nova Scotia, Canada described pregnancies with DS being statistically more likely to end in caesarean section.[10] A similar pattern was noted by Faro et al over a 10-year period in the US.[11]

There was a high total admission rate noted in this audit. This precedent is also described by Martin et al's study in another tertiary neonatal in Ireland where the overall admission rate was 86%.[1] Those requiring immediate admission to the neonatal unit had a longer duration of stay than those with a delayed admission (17 days versus 7 days) which suggests that these babies were sicker or had more issues establishing feeds. Those that were admitted immediately were more likely to have an abnormal ECHO than those admitted after 4 hours

of age (16 versus 32, P 0.02). These findings can guide clinicians in their care of patients with DS as well as answering questions parents may have around their child's prognosis and expected discharge date.

Only 2 babies were diagnosed with duodenal atresia in our cohort (2.7%). This is below the documented rate of 4% in infants with DS.[20,21] A possible explanation for this is that babies were transferred to a surgical centre antenatally however antenatal transfers were not included in this study.

Our study noted breastfeeding rates below the national average of 63% with only a quarter exclusively breastfeeding.[12] Feeding difficulties are well documented in infants with DS[2], however, 62% of infants in our cohort were able to establish bottle-feeding. This suggests that the low breastfeeding rates are not simply explained by a difficulty with oral feeds. Establishing breast feeding in infants with DS has many benefits including immune support and strengthening of facial muscles.[14] One Irish study showed a significant reduction in hospital admissions in babies who received more than ninety days of exclusive breastfeeds.[13] Although it can take longer to establish, studies have shown it is possible for babies with DS to breastfeed successfully.[15,16] A qualitative study of experiences of breastfeeding and breastfeeding support in Sweden showed that healthcare professionals had a predisposed negative view of babies with DS establishing breastfeeds and that mothers often felt unsupported. This study concluded that "It would be of great value if future research focused on designing and implementing support to increase knowledge regarding breastfeeding and DS among healthcare professionals."[17]

Overall, this audit shows good adherence to the DSMIG guideline in areas of cardiovascular and haematological monitoring, but there is a need for improvement in other categories such as monitoring growth correctly, avoiding TFTs, and appropriate referrals to audiology, ophthalmology and local follow-up. The results of this audit will be circulated with a focus on improving adherence within these categories and implementing a plan for educating the staff involved in caring for these infants. Our study also made interesting discoveries around delivery and admission patterns, ECHO findings and breastfeeding rates. All of these findings would benefit from being investigated further with a view to guide resources for the care of these vulnerable infants.

Declarations of Conflicts of Interest:

None declared.

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