

Trisomy 21 - Incidence and Outcomes in the first year, in Ireland today

Abstract:

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Abstract

Incidence of Trisomy 21 in Ireland, 1:546 live births, is the highest in Europe. This project aimed to define the incidence of T21 amongst liveborn infants at Cork University Maternity Hospital (CUMH), and to describe neonatal outcomes and progress in their first year. Infants were identified from Social Work department records. A retrospective review of the neonatal inpatient database, outpatient letters and medical charts was performed. Forty three infants with T21 were born in CUMH in 2010 and 2011. Incidence of T21 was 1:411. Antenatal diagnosis was uncommon at 14% (6). 34 (79%) were admitted to the neonatal unit. Co-morbidities included congenital heart disease 22 (51%) and duodenal atresia 2 (5%). Thirty four were followed-up in CUMH outpatient department. Of these, 34 (100%) had thyroid function testing, 29 (85%) ophthalmology and audiology referral, and 7 (21%) were referred for hip review. Mortality rate was 9% (4). Readmission to hospital in the first year of life was 42% (18).

Introduction

Trisomy 21 (T21), also known as Down syndrome, is the commonest chromosomal disorder in liveborn infants.^{1,2} Incidence is approximately 1-2 per 1000 livebirths worldwide but in Ireland is 1 in 546. T21 is associated with variable degrees of cognitive impairment and a wide range of disorders, including congenital heart disease, gastrointestinal atresia, feeding difficulties, hearing loss, ophthalmologic disease, thyroid disease and hip abnormalities.³ Surveillance is recommended throughout early childhood.^{4,5} Outcomes of infants with T21 have not previously been audited in our department. This study aims to document outcomes in the first year of life, for infants with T21 born in our hospital.

Methods

The primary study site was Cork University Maternity Hospital (CUMH). Additional study sites included the paediatric departments at Cork University Hospital (CUH), Bon Secours Hospital Cork (BSH), and Mercy University Hospital (MUH). All liveborn infants with T21, born at CUMH in 2010 and 2011, were chosen as the study population. A retrospective review of the neonatal inpatient database, medical charts, outpatient letters and i.Laboratory Web Enquiry system, was performed. CUH, MUH and BSH were contacted to identify admissions in the first year of life.

Results

There were 17,684 liveborn infants in CUMH in 2010 and 2011. 43 were diagnosed with T21, giving an incidence of 1:411. The mean (range) maternal age was 36 (22,44) years. 6 (14%) were diagnosed antenatally by amniocentesis and 37 (86%) postnatally. 34 (79%) were admitted to the neonatal unit. Reasons for admission are outlined in Table 1. There was more than one reason for admission in 6 cases. Median (IQR) duration of stay in CUMH was 7 (5,20) days. Including time spent in other centres, median (IQR) total duration of hospital stay was 8 (5,25) days. 22 (51%) had congenital heart disease, 14 of whom had an atrioventricular septal defect (AVSD). 4 developed cardiac failure in the neonatal unit and 4 were transferred to other centres for tertiary cardiac care.

7 (16%) were diagnosed with gastrointestinal morbidity, all of whom required surgical management in other centres. Two infants had duodenal atresia. 4 (9%) infants died: three in the neonatal period (cardiac failure 1, necrotising enterocolitis and sepsis 1, and hydrops fetalis 1), and one at four months of age due to cardiac failure. 34 were followed-up in CUMH neonatal outpatient department. All infants followed-up had thyroid function testing. 29 (85%) were referred to audiology and ophthalmology. 7 (21%) were referred to orthopaedics for hip surveillance, 2 of whom had clinical concerns regarding hip instability. 18 (42%) were readmitted to hospital in their first year for reasons including infection and feeding difficulty. Two infants were RSV positive, one of which was during their primary hospitalisation.

Discussion

The incidence (1:411) of T21 at CUMH is similar to recently quoted national data of 1:546.⁴ The 2006-2010 EUROCAT survey demonstrated a 17% antenatal diagnosis rate, similar to the 14% in this study. The majority (79%) were admitted to the neonatal unit at birth. A higher antenatal diagnosis rate would not necessarily improve outcomes as reasons for admission were not immediately life-threatening. The presence of congenital heart disease was either the primary cause or a contributing factor to the deaths of four infants, resulting in a mortality rate of 9%. This is similar to the 7% 1st year mortality reported in the USA.⁶ Guidance for screening in the first year is published and was followed in the majority of cases. T21 is associated with various hip problems, generally during later childhood, but T21 should not be regarded as an independent risk factor for congenital hip dysplasia. The 2011 American Academy of Pediatrics guidelines do not discuss hip dysplasia.⁵ There were multiple causes of the high (42%) readmission rate in the first year. Two infants were identified as RSV positive, one whilst an inpatient in the neonatal unit. The second infant was a term baby with a normal heart readmitted at 16 weeks old with bronchiolitis. Our practice is that infants with T21 and haemodynamically significant heart disease, e.g. AVSD, are recommended to receive Palivizumab prophylaxis.

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References

- McGuire W, Fowlie PW. Routine screening by echocardiography to reduce morbidity and mortality from congenital heart disease in neonates with Down syndrome (Protocol). Cochrane Database Syst Rev 2005, Issue 3. Art No: CD005388. DOI: 1002/14651858.CD005388.
- Weijerman ME, van Furth AM, Vonk Noodergraaf A, van Wouwe JP, Broers CJ, Gemke RJ. Prevalence, neonatal characteristics, and first-year mortality of Down syndrome: a national study. *J Pediatr.* 2008 Jan;152:15-9.
- Vis JC, Duffels MCG, Winter MM, Weijerman ME, Cobben JM, Huisman SA, Mulder BJ. Down syndrome: a cardiovascular perspective. *J Intellect Disabil Res.* 2009 May;53:419-25.
- Down Syndrome Medical Interest Group. Medical management of children & adolescents with Down syndrome in Ireland. University of Dublin, Trinity College, The National Children's Hospital, AMNCH, Tallaght: Approved Guidelines; 2001
- Bull MJ, Committee on Genetics. Clinical report - health supervision for children with Down syndrome. *Pediatrics.* 2011 Aug;128:393-406.
- Prasher VP. Down syndrome and thyroid disorders: a review. *Downs Syndr Res Pract.* 1999 Aug;6:25-42.
- EUROCAT prenatal diagnosis tables [Internet]. 2012 [cited 2013 Jan 3]. Available from: <http://www.eurocat-network.eu/PRENATALSCREENINGAndDIAGNOSIS/PrenatalDetectionRates>.

8. Kucik JE, Shin M, siffel C, Morengo L, Correa A. Congenital anomaly multistate prevalence and survival collaborative. Trends in survival among children with Down syndrome in 10 regions in the United States. *Pediatrics* 2013 Jan;131:27-36.