Abstract:
Delayed diagnosis of anorectal malformation (ARM) is an avoidable event associated with significant complications and morbidity. Previous studies have suggested higher than expected rates of delayed diagnosis, especially when a threshold of 24 hours of life is used to define delayed diagnosis. The aim of this study is to highlight the prevalence of delayed diagnosis of ARM in Ireland and to determine if any improvement in rates of delayed diagnosis of ARM has occurred since we previously examined this problem over a 10 year period in 2010. We compared trends in the incidence of delayed diagnosis of ARM between two cohorts, A (1999–2009) and B (2010–2012). Delayed diagnosis was defined as one occurring after 48 hours of life. Delayed diagnosis occurred in 29 cases (21.3%) in total, with no difference in the incidence of delayed diagnosis between the two periods (P = 0.8) and none of the patients (21.3%) being females. The rate of bowel perforation in patients with delayed diagnosis was 10.3% (3 cases). Our findings highlight the importance of perineal examination was seen as early as the second century BC, when Socrates recommended anal palpation for examination of all neonates. While routine neonatal examination has been strongly advocated in the literature, and practiced widely, the delayed diagnosis of ARM continues to be a common problem. Female neonates with ARM frequently exhibit anatomical anomalies more facilitative to the passage of meconium than in males and thus delayed diagnosis appears to be more common in this population.

Introduction
Anorectal malformation (ARM) is a common paediatric congenital malformation with an incidence of approximately 1:2500 to 1:5000. Its occurrence has a male preponderance and is associated with several syndromes such as trisomy 21 and Campomelic dysplasia. Early diagnosis and timely surgical intervention is the key to successful outcome. It is expected that most cases should be diagnosed within the first 24 hours of life on routine inspection of the perineum, although some are only diagnosed following the development of features such as abdominal distension or bilious vomiting. The importance of perineal examination was seen as early as the second century BC, when Sonarus recommended anal palpation for examination of all neonates. While routine neonatal examination has been strongly advocated in the literature, and practiced widely, the delayed diagnosis of ARM continues to be a common problem. Female neonates with ARM frequently exhibit anatomical anomalies more facilitative to the passage of meconium than in males and thus delayed diagnosis appears to be more common in this population.

The delay in diagnosis is associated with early life-threatening complications such as sepsis, bowel perforation and death. Late complications include constipation and megarectum. The complications not only alter the surgical management but cause significant social and psychological morbidity. We have previously carried out a 3 year review demonstrating an unacceptably high rate of delayed diagnosis of ARM with a median time to diagnosis of day 4 of life. The aim of this study is to highlight the current prevalence of delayed diagnosis of ARM in Ireland and to determine if any improvement in rates of delayed diagnosis of ARM has occurred since our previously published study in 2010.

Methods
The medical records of 136 cases with a recorded diagnosis of ARM between 1999 and 2012 were reviewed. The study population was previously published in 2010. Cohort B consisted of cases diagnosed between 2010 and 2012. Demographic and clinical data were recorded with particular attention to clinical presentation (clinical features suggestive of anorectal malformation, such as constipation, abdominal distension, bilious vomiting, sepsis, and failure to pass meconium), timing of diagnosis (given in hours of life), type of ARM (high vs low; rectourethral, anovestibular etc.) and any complications as a consequence of late diagnosis, such as perforation and sepsis. Delayed diagnosis was defined as diagnosis made after 48 hours of life. Data collection and statistical analysis were carried out using a statistical software package (Microsoft Excel 2007).

Results
The medical records of 136 cases with a recorded diagnosis of ARM between 1999 and 2012 were reviewed in the study. The ratio of males to females was 1:1.7. The commonest symptoms at presentation were abdominal distension (58%), vomiting (35%), bilious vomiting (35%), constipation (14%), sepsis (14%), and delayed passage of meconium (10.3%). Ten patients (10.3%) had a high ARM with 17 patients (65.5%) having a low malformation (Table 2). Unfortunately there was no improvement in the rate of delayed diagnosis of ARM between the 2 cohorts (21.2% vs 21.6%).

Associated congenital anomalies
Patients from both cohorts who had delayed diagnosis had a high incidence of synchronous congenital anomalies. In cohort A only 4 (19%) cases had no associated anomalies, while none of those in cohort B were without an associated anomaly. Examples of encountered pathologies from cohort B included bilateral cleft lip (1 case), VSD/ASD (4 cases), tracheo-oesophageal fistula/oesophageal atresia (TOF/OA) (1 case). Conversely, in this same cohort, 8 of 22 patients (36%) whose diagnoses were not delayed had no associated congenital anomaly.

Delayed diagnoses
An unusual case recorded involved a 7 year old girl with developmental delay and a history of perinatal hypoxic insult who was diagnosed following presentation with chronic constipation. She ultimately underwent a primary modified posterior sagittal anorectoplasty (PSARP). Another neonate was transferred on day 3 of life from a regional hospital with abdominal distension and suspected intestinal obstruction, having passed meconium and been documented to have a high ARM with 17 patients (65.5%) having a low malformation (Table 2). Unfortunately there was no improvement in the rate of delayed diagnosis of ARM between the 2 cohorts (21.2% vs 21.6%).

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The proportion of patients whose initial surgical management following delayed diagnosis involved diverting colostomy formation was similar in both cohorts, with 11 patients (52.4%) in cohort A and 5 patients (62.5%) in cohort B being managed this way. Five patients (23.8%) in cohort A and 2 patients (25%) in cohort B underwent primary analplasty. Four patients (19.1%) in cohort A and one aforementioned patient (12.5%) from cohort B were treated with a primary modified PSARP (mini-PSARP). One patient in cohort A (4.8%) was treated with primary diverting ileostomy due to gross large bowel dilatation and perforation of the sigmoid colon. No patient in either group underwent a primary PSARP.
Discussion

Anorectal malformations are a spectrum of conditions ranging from imperforate anal membrane to complete caudal regression. Most of these anomalies can be easily identified by inspection of perineum during routine neonatal examination and immediate referral should be made to a tertiary care centre for appropriate management 17,18. If the diagnosis of anorectal malformation is delayed, the child is likely to have a significantly higher incidence of serious complications with associated major stress to carers. A study on Cohort A was published in 2009 which revealed that approximately 21% of all children presenting to a paediatric surgical tertiary referral centre had a delay in diagnosis of their ARM in excess of 48 hours, with an associated burden of morbidity including bowel perforation, which occurred in 2 patients (9.5%). The similar trend observed in cohort B (Table 1) is disappointing. One study of 52 consecutive cases of anorectal malformation found that rates of delayed diagnosis are approximately double this when 24 hours of life is used as the threshold for defining delayed diagnosis. Of note the rate of bowel perforation in that study population, at approximately 12%, is similar to that recorded in our population. It appears somewhat fortunate that we have not experienced any mortality in either cohort when mortalities have been described in the setting of delayed diagnosis in other papers.

Nonetheless, widespread problems exist internationally regarding the diagnosis of anorectal malformations, something which is recognised in the literature 19,20. One retrospective review of 75 cases, which used 24 hours of life as the threshold criterion for delayed diagnosis, suggests that this phenomenon is much more common than was previously thought while another review of 36 cases suggested the problem is particularly prevalent in the developing world 15,16. A number of other congenital anomalies (cardiac, congenital cataracts and developmental dysplasia of hip) may be missed on routine neonatal check 17. One aforementioned case in our study was transferred on the third day of life from regional hospital with not only a previously undiagnosed anorectal malformation but also an undiagnosed TOF/OA. However it could be argued that an ARM is a far less subtle congenital anomaly than congenital cardiac or hip pathology.

Current guidelines indicate that detailed examination for significant congenital anomalies should be performed by an appropriately trained clinician between 24 and 48 hours of birth 21. The passage of meconium alone should not be taken as an indication of normal anal anatomy as meconium may also be passed via a fistula. Similarly, the presence of an anus does not exclude ARM. We emphasize that the neonate should be fully undressed and the perianal area must be cleaned of meconium. Careful, comprehensive clinical examination of a baby should include documentation of the position, appearance and patency of the anus. The Royal College of Physicians of Ireland (RCPI) has recently constructed an algorithm to aid in the diagnosis of anorectal malformations (Figure 1). We strongly recommend adherence to this algorithm to improve diagnosis of ARM, and improve the current unacceptably high rate of delayed diagnosis. At present in this country delayed diagnosis of ARM is a common occurrence and carries the risk of severe life-threatening complications in the short term, and increased morbidity in the longer term. This situation is unacceptable. Despite our previous study highlighting the issue, there has been no improvement in pick-up rates. Adherence to a clinical algorithm may be the only way to make delayed diagnosis of anorectal malformations the rarity it should be.

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