CASE REPORT

Access to essential paediatric surgery in the developing world: a case of imperforate anus with rectovaginal and rectocutaneous fistulas left untreated

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SUMMARY
Anorectal malformations consist of a wide spectrum of conditions which can affect both sexes and involve the distal anus and rectum as well as the urinary and genital tracts. Patients have the best chance of a good functional outcome if the condition is diagnosed early and efficient anatomic repair is promptly instituted. This report describes a rare case of imperforate anus associated with both rectovaginal and rectocutaneous fistulas in a 6-year-old Filipino girl. The case highlights shortcomings in the healthcare delivery system combined with socio-economic factors that contributed to the delay in both diagnosis and the institution of adequate treatment. Care and preventive measures that can be implemented in low-resource settings to reduce the impact of birth defects are also discussed.

BACKGROUND
A birth defect is defined as any abnormality affecting body structure or function that is present from birth. Congenital malformations of single systems, organs or limbs comprise the majority of birth defects with a prevalence of 36.5 per 1000 births.1 In the Philippines, a review of in-patient records from 1996 to 2000 at the Philippine General Hospital (PGH) listed congenital heart defects as the most common birth defect. This was followed by Hirschsprung disease, congenital atresia, and stenosis of anus without fistula.2

Anorectal malformations (ARM) include a wide spectrum of defects in the development of the lowest portion of the intestinal and urogenital tracts. Children with these malformations have no opening where the anus should be and are said to have an imperforate anus.3 However, the use of the latter term belies the complexity of this condition. Imperforate anus is an embryonic birth defect that results from incomplete formation of the hindgut, resulting in the absence or abnormal localisation of the anus around the eighth week of development. The rectum may end in a blind pouch or have openings to the urethra, bladder or vagina.4

Imperforate anus occurs in about 1 in 5000 live births; its aetiology remains unknown and is likely multifactorial. A genetic component is also possible because as early as the 1950s, it was recognised that there was an increased risk (as much as 1/100) for a sibling of a patient with ARM to be born with a malformation, compared with the incidence of about 1 in 5000 in the general population.5 There is a reported association between imperforate anus and prenatal thalidomide exposure, maternal diabetes mellitus and maternal age.6 Additionally, a recent systematic review and meta-analysis indicated that paternal smoking and maternal obesity were associated with increased risks for the development of ARM.6 Although no factors are known to prevent or reduce the risk for imperforate anus during pregnancy, a study conducted in China showed that daily maternal consumption of 400 μg of folic acid before and during early pregnancy may reduce the risk for imperforate anus.7

Imperforate anus can be categorised based on sex specific classifications and the site of the associated fistula. In females, imperforate anus may be associated with fistulas to the perineum or vaginal vestibule and rarely to the vagina.7 The most common anomaly in females is a rectovestibular fistula.5 A fistula in the perineum or between the blind lower end of the rectum and urethra (bulbar or prostatic) or the bladder neck may occur with imperforate anus in males. Isolated imperforate anus can also occur in both sexes, while cloaca is a rare variant seen in females where the rectum, vagina and urethra open together in a common channel.7

ARM has the unique aspect of having a higher association with congenital anomalies, with the reported range varying between 20% and 80%. The most common associated anomalies reported worldwide were related to the urogenital system, the most frequent of which are renal agenesis, ectopic kidney, vesicoureteral reflux, hypospadias and undescended testes. Other systems such as the cardiovascular, gastrointestinal tract and nervous systems, can also be affected.8 Complex defects that are associated with other anomalies have a poor functional prognosis compared to minor defects without accompanying anomalies.8

The diagnosis of imperforate anus is usually made shortly after birth by routine physical examination. The majority of ARMs in girls can be diagnosed correctly on physical examination alone. An anal orifice visible on the perineum with a normal vagina and urethra is indicative of a perineal fistula. A rectovestibular fistula, on the other hand, occurs if the opening is in the posterior vestibule and...
outside the hymen. If only one orifice is seen between the labia, cloaca is highly likely. Rectovaginal fistulas may initially appear as imperforate anus with no fistula (normal-appearing vagina and urethra, but no visible anal orifice) but closer inspection will usually show the presence of an orifice in the posterior wall of the vagina or internal to the hymenal ring. Radiological evaluation of the newborn with an imperforate anus should include abdominal ultrasound to detect urological abnormalities (i.e., distended vagina or hydrocolpos associated with persistent cloaca) and plain radiographs of the spine and sacrum to assess for anomalies. Sepsis, aspiration, abdominal distension, colonic perforation, respiratory distress, electrolyte imbalance and even death are complications that can arise from the delayed diagnostic and therapeutic management of ARM. The reported mortality rate of imperforate anus is between 1.4% and 30%. Undoubtedly, the importance of early diagnosis of imperforate anus cannot be over-emphasised. Immediate diagnosis will allow timely planning of subsequent management. Staged surgery, the most widely accepted approach for the management of ARM, consists of three operations: colostomy at birth, definitive operation after 2–3 months of age, and colostomy closure at the age of around 6 months. A posterior sagittal approach with anorectoplasty (PSARP) is the ideal definitive method to repair ARM. Regular follow-up in a bowel management treatment centre is needed in all these cases in order to achieve better continence. A combination of successful anatomical repair and appropriate programmes of bowel care can lead to socially acceptable continence in a majority of patients, especially those with an intact sacrum.

We present an unusual case of imperforate anus presenting with rectovaginal fistula and concomitant rectocutaneous fistula. We also discuss the socio-economic factors that impeded the patient from getting access to early paediatric surgical care. Care and preventive measures that can be implemented in low and middle-income countries to reduce the impact of birth defects are also discussed.

CASE PRESENTATION

A 6-year-old Filipino female patient from a rural village in Northern Samar, the Philippines, presented with history of pasty-green to brownish stools per her vagina since birth. The patient was born full term to a then 36-year-old G4P3 (3-0-0-3) mother via cephalic spontaneous vaginal delivery. The mother had regular prenatal care check-ups at a rural health unit. The pregnancy was not complicated with gestational hypertension or gestational diabetes mellitus. The mother took iron supplements and denied tobacco, alcohol or illicit drug use during the pregnancy. The birthing process was attended by a midwife at home and denied tobacco, alcohol or illicit drug use during the pregnancy. The child had a good appetite but was underweight for her age. Her social skills were also greatly affected as she refused to play with other children due to embarrassment about the malodorous discharge coming from her vagina. In 2012 the parents sought the help of a private donor (AGR) who was conducting clinical research in the community. Arrangements were made and the patient was subsequently admitted to the University of the East-Ramon Magsaysay Memorial Medical Center (UERMCC) for evaluation and further management.

Upon admission in late 2012, the patient had stable vital signs with a blood pressure of 90/60 mm Hg, pulse rate of 102 bpm, respiratory rate of 22 cycles/min, and a core temperature of 36°C. Pertinent physical findings included coarse crackles in the left lower lung field on auscultation. The measured abdominal circumference was 60 cm, consistent with a globular and distended abdomen (figure 1A). Bowel sounds were normal, but there was a palpable 20×21 cm firm, non-tender mass in the right upper quadrant. No direct or rebound tenderness was elicited on palpation. The rest of the physical examination was unremarkable. Haematological tests revealed mild anaemia (10.5 mg/dL), normal white blood cell count with no left shift, and slight thrombocytosis. Serum sodium and potassium were within normal limits. Chest X-ray showed hazy opacities in the left upper lobe consistent with the diagnosis of pneumonia. Plain abdominal X-ray (figure 1B) demonstrated faecal stasis and focal areas of lucencies in the anterior right lateral and superior aspect of the abdomen which was indicative of pneumoperitoneum. A working diagnosis of imperforate anus with rectovaginal fistula was made.

A subsequent referral to paediatric services for further evaluation and pre-operative care was carried out. Cefuroxime 250 mg intravenous every 8 h was started to treat pneumonia. Nutritional supplements were initiated by gradually increasing the patient’s caloric intake. On the 20th hospital day, the child underwent exploratory laparotomy with sigmoidectomy, appendectomy and colostomy. Intra-operative findings showed the sigmoid colon to be markedly distended with faeces and...
The intraoperative functional life (the said procedure, the patient is expected to live a normal and healthy life), is scheduled to be completed in 2015. While waiting for the re-operation, the patient is under strict and explicit instructions on stoma care provided by the hospital staff to the mother, the family returned home to Northern Samar while waiting for the second stage of the operation.}

With strict and explicit instructions on stoma care provided by the hospital staff to the mother, the family returned home to Northern Samar while waiting for the second stage of the operation. No peristomal skin infection was noted. The patient’s nutritional status continued to improve and she has gained a tremendous amount of self-confidence, enabling her to play and interact with other children. She also currently attends school as a grade one pupil.

Approximately 9 months after the initial operation, the child was re-admitted at UERMMC for the second stage of surgery. She underwent PSARP with an unremarkable intra-operative and post-operative course. During the second operation a rectovaginal fistula was also noted (figure 1E). The current plan is to allow the anorectal wound to heal followed by repeated and regular dilatation of the canal to prevent stricture of the neo-anus. The third stage of the surgery, which consists of the regular dilatation of the canal to prevent stricture of the neo-anus, is scheduled to be completed in 2015. While waiting for the said procedure, the patient is expected to live a normal and functional life (figure 1F).

GLOBAL HEALTH PROBLEM LIST

▸ How should rare birth defects, such as anorectal malformations or imperforate anus, be managed in developing countries with limited healthcare resources?
▸ How does a parent’s perception of the quality of healthcare services impact on their health seeking behaviour for their children?
▸ Should a national healthcare policy be created to ensure that all patients have access to essential surgical services as part of their basic human rights?

GLOBAL HEALTH PROBLEM ANALYSIS

Management and prevention of birth defects in low and middle-income countries

A report issued by the WHO in 1999 contended that governments in the developing countries should devote attention and resources to the control of genetic disorders and birth defects. Public health authorities should recognise the fact that these conditions are major causes of disease, disability and mortality in their countries. Programmes for the management and prevention of birth defects that can significantly reduce their burden in a cost-efficient manner should be initiated.

In 2008, the March of Dimes Organization proposed cost-effective interventions that can be carried out in low-income countries to reduce the impact of birth defects. Care and prevention can be implemented by expanding current maternal and child health programmes and developing medical genetic services in the secondary and tertiary tiers of health systems. The recommended measures are divided into two phases that can be tailored based on a country’s financial state, health-care delivery status and specific health priorities.

Phase one consists of improving the diagnosis, treatment and prevention of birth defects. Adequate training of primary-care providers on the screening and detection of birth defects should include learning to assess risk factors in a pregnant woman by conducting a thorough and complete newborn examination and identify recognisable dysmorphic features and patterns. Collating physical signs of birth defects that are visible externally is one way of rapidly diagnosing structural birth defects such as imperforate anus.

The treatment of birth defects consists of improving human resources at the primary-care level and establishing surgical services for children with birth defects in allocated units in secondary and tertiary healthcare. The former heavily emphasises the role of primary-care practitioners, who should be able to aid in the follow-up of patients cared for in secondary and tertiary healthcare. In addition, training on genetic counselling, psychosocial support and basic rehabilitation for people with birth defects should be promoted. It is also recommended...
that individuals in the community are trained to assist in the rehabilitation process and lay support organisations are encouraged.13

Preventive measures include family planning, promotion of adequate and proper nutrition, food fortification and supplementation with essential micronutrients specifically iodine and folic acid, prevention of infections such as syphilis and rubella, and adequate control of maternal chronic illnesses such as diabetes mellitus and epilepsy. These steps should target both pregnant women and women of reproductive age.13

Depending on the country’s needs and resources, phase two, which involves the employment of medical genetic services for risk identification and management, can be subsequently implemented.11 Prior to conception, the following are recommended: use of family history as a screening tool for risks for birth defects and cascade screening (carrier risk identification using family pedigrees and DNA analysis of identified individuals),13 antenatal screening procedures including assessment of Rhesus negativity, screening for Down syndrome and neural tube defects using maternal serum screen and ultrasound, and the use of fetal anomaly scanning to detect major malformations.11 Population carrier screening for common recessive disorders such as full blood count and electrophoresis for haemoglobin disorders can be carried out during the pre-conception and antenatal periods. Newborn screening for conditions such as congenital hypothyroidism and G6PD deficiency is also strongly advocated.13 These priority actions are intended to integrate seamlessly with a country’s existing women’s, maternal, newborn and child health programmes, and so new vertical programmes do not need to be developed.13

An effective programme of primary care can be expanded successfully (even in low-resource settings) to incorporate additional interventions such as genetic screening, into basic reproductive care. However, this can only be realised with government support and political commitment, and decisions regarding budget priorities should be guided with accurate data on the disease burden of birth defects and the cost-effectiveness of the proposed programmes. The successful thalassaemia prevention and treatment programme in Iran shows that a strong political will and accurate epidemiological data can result in cost-effective measures that reduce the impact of birth defects in a resource-poor setting.14

Unfortunately, most middle- and low-income countries currently lack the comprehensive health programmes necessary to reduce the impact of birth defects.1 In the Philippines, prenatal screening is practiced in a very limited way. The most widely used prenatal diagnostic procedure is prenatal ultrasonography, and first trimester ultrasound screening has been introduced in some urban medical centres. Non-invasive forms of prenatal diagnosis such as maternal serum screen are not routinely offered.15 Preventive programmes such as fortification of staple foods, that is, iron in rice, vitamin A in wheat flour, and iodised salt supplementation, do exist, but there is really no formal preconception care programme in the country.16 On the other hand, population-based screening conducted in the post-natal period, specifically newborn bloodspot screening (NBS), is more successful. NBS was integrated into the public health delivery systems in 2004. The national health insurance programme (Philhealth) initially covered 90% funding of NBS but complete coverage was instituted in September 2011.16 In January 2014, NBS was expanded to include 22 more disorders such as haemoglobinopathies and additional metabolic disorders, in addition to the six previously screened diseases.17

ANATOMY OF RECTAL ANOMALIES

Our case illustrates an unusual type of imperforate anus associated with both rectovaginal and rectocutaneous fistulas. Worldwide, the most common anomaly in females is a rectoves-tibular fistula.3 A review of imperforate anus cases in countries such as Africa,18,19 India20 and Saudi Arabia19 revealed similar findings. A low frequency of rectovaginal fistulas was observed in Africa, with only 1.2% of 1401 patients diagnosed with the condition.19 A study conducted in Saudi Arabia also showed that rectovaginal fistulas were also unusual, occurring only in 2% of 53 females with imperforate anus.7 In the Philippines, a review of 189 imperforate anus cases showed that the most common type of associated fistula in females was of the anovestibular subtype. Cloacal (9%) and rectovaginal fistulas (7%) were actually more common than rectoves-tibular fistulas (1%).21

A retrospective study of 619 cases of ARM in girls aimed to determine the true incidence of rectovaginal fistula. The study revealed that the incidence was only 1%, which contrasts greatly with the literature published over the last 70 years. One explanation for this was that there is a much better understanding of the true anatomy of the different malformations today and that a cloaca is less commonly mistaken for a rectovaginal fistula.3 Differentiation between a cloaca and rectovaginal fistula is very important because its misdiagnosis has important therapeutic consequences. Obstructive uropathy and hydrocolpos commonly present with cloaca, and so diagnosing this as rectovaginal fistula may neglect the problem of obstructive uropathy. Another implication is that only the rectal anomaly would be repaired and the patient will have a persistent urogenital sinus.5 In our case, the demonstration of a rectal orifice internal to the hymenal ring established our diagnosis of a true rectovaginal fistula.

The presence of a concomitant rectocutaneous fistula also makes this case unique. A rectocutaneous fistula is an abnormal communication between the rectum and the peri-rectal skin.22 Our literature search showed only two reports with similar or closely related cases. The most recent report (1994) is of two newborn boys with imperforate anus associated with a recto-bulbar-cutaneous fistula. Both patients had a long fistulous tract originating from the rectal pouch and coursed deeply into the scrotum. In case 1, the midportion of the fistula communicated with the bulbar urethra through a single small opening and exited in the anterior aspect of the penis also near the penoscrotal junction.23 The second report (1983) illustrated an imperforate anus with two fistulas, one ano-urethral and one penile urethro-cutaneous.24 Our case resembled more an H-type fistula (figure 1G) with the fistulous tract from the rectal pouch opening in the perineum, while a short fistula connects the rectal pouch to the distal portion of the vaginal canal and exits internal to the hymenal ring.

There have only been anecdotal reports of H-type fistula cases among paediatric surgeons in the Philippines. As far as we know, this case of a female patient with an imperforate anus associated with two fistulas occurs rarely.

MANAGEMENT OF ARM

Delays in both diagnosis and surgery were apparent in this case. Delayed diagnosis was defined as a diagnosis made after 48 h of life. Despite routine physical examination postpartum, one in five neonates born with imperforate anus had a delayed diagnosis.25 In a study conducted in Malawi, Africa, the median age at
presentation was 24 days. The median age among boys was 3 days (range 1–24 days), while girls presented after a median age of 90 days (range 1–1185 days). A Philippine study also showed the same trend, with males presenting at a younger age (range 1–365 days) and females presenting at a later age (range 1 day–14 years). The wide range in the ages at consultation among the females can be due to the symptoms developing only in late childhood or early adolescence. In contrast, complications in males manifested earlier and their anomalies were more easily detected.

In this case, the diagnosis of ARM was missed in the newborn period because the primary caregiver (midwife) was unable to perform a thorough physical examination. This is expected as local midwives are not routinely trained to conduct a proper examination and their lack of exposure to imperforate anus cases contributes to their lack of awareness and knowledge about the condition. Although the mother had a nursing background, she was also unable to detect the condition and it was only when obvious symptoms were manifested did she actually recognise the medical problem. However, the parents had the perception that the quality of health services in the capital city of Manila were far superior to those being offered in their own province. This led them to bypass their local primary-care services and go directly to a tertiary hospital in Manila.

The household’s meagre income clearly delayed the family from accessing immediate surgical intervention for the patient. Education, especially that of women, is strongly related to many behaviours and choices favourable to good health, even after controlling for income. In this case, the lack of awareness of the parents led to further delays in seeking treatment. Once the parents were aware of their child’s life-threatening complications, a sense of urgency prodded them to take a more proactive attitude in generating the needed out-of-pocket expenses so they could bring their child to PGH for treatment. In an ideal setting, the financial resources that they were able to access via a cooperative loan together with direct patient subsidies from their local elected politician’s Priority Development Assistance Fund (PDAF) allocation, should have been enough to cover the cost of medicines, supplies and diagnostics while the child was in a public hospital. The devolution of healthcare to the local government unit, specifically its impact on the health referral system, also contributed to the delayed access to health services. In addition to a lack of a clearly defined referral system, a decline in the maintenance of infrastructure and under-financing of the operational costs of services also resulted in primary and secondary hospitals no longer being capable of providing referral services to health centres in their catchment areas. Moreover, PhilHealth’s (the national healthcare provider) No Balance Billing (NBB) policy, which provides that no other fees or expenses shall be charged to or be paid for by indigent patients above and beyond the packaged rates, only covers 11 surgical cases and excludes paediatric surgical procedures. Thus, indigent patients such as these must come up with the additional funds needed for surgery. Patients also need to cover their own travel costs and living expenses. This dilemma jeopardises the health of approximately half of the nation’s 100 million people.

Unfortunately, negative perceptions (perhaps justified) about the substandard quality of care provided in rural and geographically poor provinces has led to severe congestion of patients in tertiary hospitals in the capital Manila. The hospitals have limited resources and manpower to cope with the sheer volume of patients admitted each day. The struggle of PGH, which is the largest public hospital in the country, to handle this overwhelming number of patients has resulted in continuous reliance on state subsidies. In 2013, the approved national budget only allocated PHP 275 million (US$6.25 million) for PGH’s capital outlay. This amount is clearly below the PHP 990 million (US$22.5 million) budget requested by PGH. This influx of patients into metro Manila clearly demonstrates an urgent need for a national policy to address the discrepancy in health services provided at the provincial level. The current referral system to Manila is not sustainable and is already impacting on the quality of care provided. Regrettably, many simply do not get the care they need.

**THE THREE DELAY FRAMEWORK**

The three delay framework explains the delay between onset of symptom and delivery of appropriate care in the context of maternal mortality. The first, second and third delays refer to delay in seeking, reaching and receiving care, respectively. This framework can also be used to describe the three types of delay in accessing surgical care. In the above case, the ‘first delay’ occurred due to financial incapacity, low confidence in the health system and lack of awareness about the debilitating and fatal effects of the congenital malformation. The ‘second delay’ ensued due to geographical barriers and the high cost of travel and living expenses needed to reach the health facility. The ‘third delay’ arose when admission to the hospital did not guarantee receipt of care as the medical centre was already performing beyond the capacity of its personnel and resources.

The majority of birth defects are congenital malformations and many are amenable to paediatric surgery. Hence, a comprehensive national programme which aims to reduce the toll of birth defects should also incorporate a robust surgical system. This is timely as the Sustainable Development Goals (2015–2030) will replace the Millennium Development Goals (2000–2015). Universal health coverage by 2030 will also include surgery and the reporting of surgical indicators. The Lancet Commission on Global Surgery presented a national strategy plan addressing five major areas of surgical system development: infrastructure, workforce, service delivery, information management and financing. It is important for low and middle-income countries to gather epidemiological data, which should include both the prevalence of birth defects as well as assessment of the cost-effectiveness of interventions, in order to reduce the impact of birth defects in their countries. Using the template outlined by the Lancet commission, low and
middle-income countries can now develop a national health plan incorporating access to surgical care. A strong and sustained political commitment and active involvement of the international community are also crucial for such a national policy to be effective.

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