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SPECIAL ARTICLE

The primary care of a patient with a history of a gastrointestinal malformation and abdominal wall or diaphragmatic defects
[[es]]Atención primaria del paciente con una malformación congénita☆

Jordi Prat-Ortells*, **Xavier Tarrado**

Hospital Sant Joan de Déu, Universitat de Barcelona, Esplugues de Llobregat, Barcelona, Spain

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Abstract Survival of patients with congenital malformation has improved over the last decades. Primary Care paediatricians must be aware of the most common problems that this group of patients suffers. More importantly, paediatricians can offer a holistic view that is often lost in specialised consultation.

This article is focused on common congenital malformation, such as oesophageal atresia, abdominal wall defects, anorectal malformation, Hirschsprung disease, and congenital diaphragmatic hernia. The main problems are shown, with special emphasis on long-term complications and all the dimensions of the individual.

Resumen La supervivencia de los pacientes con antecedentes de malformaciones congénitas complejas ha aumentado en las últimas décadas. El pediatra de atención primaria debe conocer los problemas más habituales que pueden sufrir este grupo de pacientes. Además, puede ofrecer una visión global que, a menudo, se pierde en las consultas especializadas.

En este trabajo se recogen algunas de las malformaciones congénitas digestivas y respiratorias más habituales como la atresia de esófago, defectos de pared abdominal, malformación

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* Corresponding author.
E-mail address: joprat@sjdhospitalbarcelona.org (J. Prat-Ortells).

anorrectal y enfermedad de Hirschsprung y la hernia diafragmática congénita. Se señalan los problemas de mayor interés para el pediatra, haciendo hincapié en las complicaciones a largo plazo y en todas las dimensiones de la persona.

Introduction

In recent decades, improvements in neonatal care, anaesthesia and surgery have increased survival of children born with severe congenital anomalies. Once they return home, these patients will be assigned a primary care paediatrician that will act as the point of contact with the health care system. Since in many cases these anomalies are rare, paediatricians are often inexperienced in the management of patients with congenital malformations. In this article, we discuss relevant aspects that primary care paediatricians ought to take into account in managing this group of patients. Specifically, we will discuss the management of infants born with oesophageal atresia, intestinal atresia, abdominal wall defects (gastroschisis, omphalocele) or congenital diaphragmatic hernia, or those who have had necrotising enterocolitis or who have undergone a bowel resection.

Overview

It is only logical to expect that patients with congenital malformations will be offered a combined follow-up involving the primary care paediatrician and more specialised care delivered in the reference hospital for the patient's condition. Patients that are more severely ill and have more complex malformations require follow-up by a multidisciplinary team including paediatricians affiliated with a tertiary care hospital.

However, many malformations involve mainly one organ or system. In these cases, the work of the primary care paediatrician is very similar to the work performed in the care of other children: the routine ''well-child'' follow-up with management of intercurrent disease. In fact, specialists expect to receive the general overall perspective of the patient that can only be offered by the paediatrician.

Some patients with congenital malformation exhibit weight gain below what would be expected. This is particularly frequent in patients with a history of preterm birth, low birth weight or gastrointestinal problems. If there is significant nutritional compromise, the patient should be managed at the hospital level. Otherwise we should not be excessively concerned by our patient being in a low percentile in weight charts. Growth charts represent the general population of healthy children, so their data cannot be extrapolated to infants born with severe malformations. Therefore, what matters is whether the growth of the infant runs parallel to the standard curve, as opposed to the percentile in the curve.

We now proceed to analyse the role of the primary care paediatrician in the management of patients with a number of specific congenital malformations. Table 1 summarises the main comorbidities associated with the malformations discussed. The text provides an overview of the most significant malformations and offer practical information for the management of these patients.

- Oesophageal atresia (OA)

Respiratory problems are common in OA. The respiratory symptoms associated with OA (tracheomalacia, bronchitis...) usually improve with age and are frequent in the first years of life. Many patients with OA have a characteristic cough or breathing sounds reminiscent of asthmatic patients, but these symptoms do not improve with salbutamol. This can be attributed to tracheomalacia of varying severity.[1]

Recurrent instances of aspiration accompanied by coughing fits during consumption of fluids are suggestive of tracheo-oesophageal fistula.[2] This complication may also develop late. If suspected, the patient must be referred to a specialist.

Gastrointestinal problems vary depending on age.[3] They start with narrowing of the suture,[2] which manifests as difficulty swallowing. It is rare for this tendency toward stenosis to persist past the first or second year of life (in which case it is necessary to rule out gastro-oesophageal reflux disease [GERD]).

Gastro-oesophageal reflux disease is the most frequent long-term complication. It may go undetected and be asymptomatic. For this reason, it is recommended that assessments of GERD be performed routinely at different points in childhood.[4,5] All patients with OA should undergo an upper endoscopy with collection of biopsy samples, possibly combined with multichannel intraluminal impedance/pH monitoring once they reach 1 year of age.[3,6] Until this time, patients usually receive omeprazole (1 mg/kg/day). If GERD is ruled out, the medication is discontinued, and the patient remains in follow-up. In asymptomatic patients, the upper endoscopy with biopsy should be repeated at age 8-to-10 years and before their transition to adult gastroenterological care at age 17–18 years.[6] These evaluations may identify patients with OA and mucosal inflammation changes or even Barrett's oesophagus. Cases of adults with OA that develop oesophageal cancer have been described in the literature.[7,8]

Table 1 Summary of the most frequent comorbidities in congenital malformations.

Morbidity	Congenital diaphragmatic hernia[c]	Oesophageal atresia	Intestinal atresia and abdominal wall defects	Anorectal malformation and Hirschsprung disease
Pulmonary				
Recurrent infections	Frequent; improves in the first years of life	Frequent; improves in the first years of life		
Tracheomalacia		Frequent; (hasta up to 80%[4]) improves in the first years of life		
Lower endurance	7–35%[21]	Restrictive pattern and bronchial hyperresponsiveness are very common		
Recurrence of TOF[a]		Around 10%[2]		
Gastrointestinal				
Oral aversion	Related to prolonged intubation or ventilatory support	Related to the duration of feeding through a nasogastric or gastrostomy tube		
Food impaction/suture stenosis		Very common after surgery (60–70% at 1 year of age[2,3]). Rare from age 1 year. In case of late onset, rule out GERD or eosinophilic oesophagitis		
GERD[b]	Frequent (50–100%[21]). We recommend routine screening with impedance/pH monitoring	Very frequent; routine screening in children and monitoring during adulthood		
Risk of cancer	Difficult to quantify; associated with GERD and Barret's oesophagus*			
Intestinal obstruction	Possible		One of the most frequent complications. Attempt conservative treatment	
Nonspecific abdominal pain			Frequent. There may be episodes of partial obstruction	

Table 1 (*Continued*)

Morbidity	Congenital diaphragmatic hernia[c]	Oesophageal atresia	Intestinal atresia and abdominal wall defects	Anorectal malformation and Hirschsprung disease
Failure to thrive Short bowel syndrome	Up to 50–60%[21]	Up to 30%[3]	In complex patients This complication is more severe in these patients. A small percentage may become dependent on TPN or even require a bowel (or multivisceral) transplant	
CD recurrence	Related to severity and the need for repair with a hernia patch			
Constipation				Very frequent, especially in low ARMs[d]; requires active management
Incontinence				In cases of severe ARM or pseudoincontinence due to constipation
Musculoskeletal				
Scoliosis	Relatively frequent (4–50%[21])	Associated with thoracotomy (may be minimised with careful technique or a thoracoscopic approach)		
Chest wall deformities	Frequents			
Neurodevelopment				
Neurosensory	Hearing loss		Hearing loss	
Motor impairment	Associated with history of ECMO[e]			

Table 1 (*Continued*)

Morbidity	Congenital diaphragmatic hernia[c]	Oesophageal atresia	Intestinal atresia and abdominal wall defects	Anorectal malformation and Hirschsprung disease
Behavioural problems Language and learning problems Other	Observed more frequently in these patients. The literature also describes memory problems. Autism in up to 11%		May develop in some patients with gastroschisis	
Psychosocial problems	Patients with more severe disease and impaired quality of life may be at higher risk of psychiatric disorders			Severe ARM, patients with prolonged rectal dilation
Sexual problems				Possible in males with severe ARM
Impact on caregiver; economic costs	In severe cases, with ventilation dependency or neurodevelopmental abnormalities	In severe cases; oral aversion produces high levels of anxiety	When multiple surgeries are required, very high if patients are dependent on TPN[f]	Frequent, especially at first and if patients require rectal dilatation for a long period, there are other malformations or patients require complex intestinal management
Quality of life impairments	Usually normalises with increasing age	Early years of life. In older patients, impaired in more severe forms of disease or in cases with associated malformations	Excellent quality of life in simple and uncomplicated presentations; severe impairment in case of short bowel syndrome or TPN dependence NPT	Severe impairment if associated with incontinence, impairment mainly in early years of life, good quality of life in mild or moderately severe forms.

[a] TOF trachea-oesophageal fistula;
[b] GERD, gastro-oesophageal reflux disease;
[c] DH, diaphragmatic hernia;
[d] ARM, anorectal malformation anorectal;
[e] ECMO, extracorporeal membrane oxygenation;
[f] TPN, total parenteral nutrition.
[*] The potential risk of oesophageal cancer is due to GERD and thus associated in equal measure to DH and oesophageal atresia.

The sudden development of food impaction in these patients suggests the diagnosis of eosinophilic oesophagitis.[9] This condition is relatively frequent in patients with OA, and it is diagnosed by mean of upper endoscopy with biopsy.

All patients with OA exhibit abnormal oesophageal motility. As a consequence, they need to drink an abundance of fluids, especially during meals. Physicians must promote fluid consumption and ensure that patients understand this need. Patients with OA often prefer foods with a high water content and a soft texture. It is very important that patients with OA develop the habit of chewing food thoroughly to prevent impaction.

Some patients with OA may develop deformities of the thoracic wall secondary to surgical intervention. This can be prevented with careful muscle-preserving technique or by using a thoracoscopic approach.

- Abdominal wall defects: gastroschisis and omphalocele

The prognosis and quality of life of these patients are excellent. However, there is a small proportion of highly complex patients. Let us consider the example of a survivor of complex gastroschisis. Complex gastroschisis cases are those associated with atresia, intestinal necrosis or perforation, and often involve significant bowel shortening. In these patients, weight and height in particular tend to be affected. In some cases, there is no catch-up growth.

Repeated surgeries are common in these patients[10] for problems that may range from umbilical hernia or cryptorchidism (probably due to a lack of intraabdominal pressure during development) to bowel obstruction. While they may not require surgical intervention, episodes of partial bowel obstruction and chronic abdominal pain can also affect these patients.

In patients presenting with abdominal pain, the following manifestations are considered to be alarming: absence of bowel movements and abdominal distension, bilious vomiting and severe pain, especially if accompanied by vegetative symptoms (nausea, sweating, pallor...), as they suggest bowel vascular compromise or intestinal ischaemia. The initial approach should be conservative management with fluid replacement, nil per os and feeding through a nasogastric tube. Urgent surgery is indicated if the symptoms do not resolve or the patient presents clinical or radiological worsening at any moment concurrently with symptoms of intestinal compromise (lancinating pain, elevation of lactate, metabolic acidosis, poor intestinal perfusion in imaging tests).

Survival is lower and quality of life impaired in patients with intestinal failure and dependent on parenteral nutrition. These are highly complex patients that need to be treated in highly specialised units. In the most simple cases—and this also applies to other patients such as survivors of uncomplicated intestinal atresia or necrotising enterocolitis—patients can lead a completely normal life, but they are more vulnerable if exposed to common diseases such as gastroenteritis. They are at higher risk of dehydration and requiring parenteral fluid replacement, so paediatricians need to be particularly watchful.

Many surgical departments routinely excise the appendix in patients with complex abdominal diseases. Appendectomy is all but imperative in patients with associated malrotation (a common finding in patients with gastroschisis, omphalocele or diaphragmatic hernia).[11]

An aspect that may be neglected in these patients is overall neurodevelopmental impairment.[10] In particular, patients with gastroschisis commonly present multiple neurologic risk factors: young maternal age, possible prenatal exposure to drugs, preterm birth and low birth weight, a dysfunctional family environment, long lengths of stay, multiple surgical interventions and exposure to general anaesthesia at a very young age. In general, these patients are similar to other children by age 2 years. However, they may exhibit memory and behavioural problems, learning difficulties or language disorders. A neurodevelopmental evaluation is recommended between ages 1 and 6 years.[12] Some children with this previous history are also at higher risk of hearing loss.[10]

Vaccination of these patients should conform to the routine immunisation schedule applied to any other children. Oral rotavirus vaccines confer immunity by simulating native infection by rotavirus. This vaccine is contraindicated in patients with immunodeficiency (suspected or confirmed), a history of intussusception or an uncorrected gastrointestinal malformation. There is no formal contraindication for vaccination against rotavirus in survivors of gastroschisis, intestinal atresia or necrotising enterocolitis.[13,14] However, we have directly witnessed a patient with a history of gastroschisis develop fulminant necrotising enterocolitis (resulting in death) following the administration of this vaccine, and are also aware of another similar case, although milder. In both cases, testing did not detect the presence of a virus in the stool or blood of the patients, and it was not possible to establish a causal relationship between the vaccine and the observed complications. Gastroschisis is a disease that can in itself predispose to necrotising enterocolitis. Considering the risks and benefits of this vaccine in Spain, it seems reasonable to recommend against the administration of the rotavirus vaccine to patients with gastroschisis or other diseases involving inflammation of the intestine or significant dilation of bowel loops.

- Hirschsprung disease and anorectal malformations (ARMs)

Patients with uncomplicated forms of these anorectal diseases have an excellent prognosis and a quality of life within the normal range.[15] Typical examples include patients with Hirschsprung disease and a rectosigmoid aganglionic colon segment[16] or patients with rectoperineal fistula, rectovestibular fistula (girls) or rectourethral fistula (boys, although they do not have the excellent outcomes observed in the previous examples).

However, the spectrum of patients with more severe malformations or with associated syndromes (Down syndrome is associated with both Hirschsprung disease and ARMs) may have more significant problems, mainly affecting continence and sexual activity.

Based on the evidence of longitudinal studies, intestinal manifestations improve as years go by. Eighty-three percent of patients with ARMs are socially continent (some of them depending on enemas). However, the quality of life of patients with severe ARMs is impaired in 15–36% of cases and severely impaired in 9%.[15]

Incontinence can be attributed to different factors, including the severity of the malformation, the morbidity associated with surgery or the presence of medullary involvement. Specialised units must be able to offer an intestinal management protocol to the patients. Furthermore, patients with severe ARM may also experience urinary incontinence, and functional evaluations must be performed to prevent long-term complications.

Constipation is frequent in both diseases and requires treatment, as allowing it to progress can result in pseudoincontinence with involuntary bowel movements due to overflow. Primary care paediatricians are qualified to manage mild cases of constipation.

In this group of patients, parents should be made aware of the importance of establishing routines to facilitate bowel movements in the patient. We should recommend a diet rich in fruits, vegetables and whole grains, avoiding foods that promote constipation (such as rice, apples or bananas). The patient should be offered plenty of fluids. Lastly, it is advisable to establish times following meals (after lunch or dinner, to take advantage of the gastrocolic reflex) when the patient will be encouraged to go to the bathroom and attempt a bowel movement.

Male patients with a history of severe ARM (for instance, a rectoprostatic fistula) may also suffer from erectile dysfunction and absence of ejaculation, with the associated decrease in fertility.[15] In female patients, ARMs do not cause fertility problems, but in many cases caesarean section will be the preferred method of delivery.[15]

We ought to introduce a new concept in the follow-up of patients with malformations that primary care paediatricians should also be aware of: psychosocial health and mental health disorders. These patients are a clear example of this need.

In general, patients with malformations have risk factors that may affect their psychosocial health: the malformation itself, repeated exposures to anaesthesia, long hospitalizations, dysfunctional families or a history of traumatic treatments. Overall, these patients have a 3-fold risk of having psychiatric disorders compared to the general population.[17] To provide a framework for comparison, children with chronic diseases such as asthma or diabetes have a 2-fold risk of having psychiatric disorders compared to their healthy peers.

Male patients with bladder exstrophy may be the subset at highest risk of psychiatric illness.[17] Dissatisfaction with their genitals and urinary incontinence are the main cause of the disturbances experienced by these patients in their social and sexual relationships. A study conducted in the clinical setting (without a control group) determined that up to 15% of patients with bladder exstrophy had experienced suicidal ideation; the sample included a patient out of 38 men with bladder exstrophy aged more than 14 years that had completed suicide.[18] Nevertheless, we can allow ourselves to be fairly optimistic in cases of bladder exstrophy, too, as more than 50% of affected patients report a good quality of life.[17]

Patients with ARM also have more mental health problems compared to patients with other malformations. These problems are related to incontinence, but there is also another salient factor: a history of recurrent traumatic experiences, such as rectal dilations.[17] Rectal dilations are performed over a short period of time in infants with Hirschsprung disease or ARMs. However, when problems arise—such as a tendency toward stenosis—performance of dilations can extend for a long period. In some patients, the sequelae of rectal dilations performed for several years are reminiscent of those of children that have experienced sexual abuse, with the aggravating factor that in most cases these dilations are performed by the parents. If the patient truly needs them, perhaps it would be preferable to find a person other than the parents to perform these traumatic practices.

Another aspect that must be considered in regard to psychiatric health is the condition of the main caregivers of these children, who also tend to have more stress and mental health problems.[17]

- Congenital diaphragmatic hernia (CDH)

There is no doubt that patients with CDH benefit from multidisciplinary follow-up in reference units, and primary care paediatricians are one of the links in this comprehensive care chain.[19–21]

As is the case of patients with OA, survivors of isolated or mild-to-moderate CDH have gastrointestinal and respiratory problems (similar in many aspects) that tend to improve with age.[20,22]

However, patients with severe hernias exhibit long-term morbidity of different types: gastrointestinal, pulmonary, neurodevelopmental or musculoskeletal.

Gastrointestinal problems usually manifest as GERD (with a prevalence that may exceed 50%),[20,21,23] growth delay, oral aversion (related to prolonged ventilatory support), intestinal obstruction or even recurrence of hernia. Failure to thrive (observed in 14–63% of cases) may occur in severe cases in patients with a basal energy expenditure above normal, with substantially increased use of energy in the work of breathing.[19]

Patients with CDH are also at increased risk of airway infections and obstructive respiratory symptoms (asthma, wheezing).[19,21] In fact, the obstructive pattern is very common in CDH and does not always improve with time. Nevertheless, this pattern is similar to the pattern exhibited by other infants that required ventilatory support of similar intensity.[19]

Neurodevelopmental delays[19] may be present in patients with CDH, most frequently in the first 2 years of life. In the long term, most patients with CDH are within the normal range. However, 17% score in the lower limit of normal and 17% have a very low intellectual quotient score. Approximately 11% of patients with CDH have autism spectrum disorder (compared to 1.5% of the general population). Nearly 50% of these patients have problems to perform at grade level in school. There are also survivors of CDH with impaired motor skills associated with a history of ECMO. In addition, patients with CDH may suffer neurosensory impairment and hearing loss.

One last aspect that deserves consideration is the impact that this anomaly has on the main caregivers.[21] The parents of these patients devote more time to the care of their child, which sometimes results in missed hours of work or on quitting working altogether. Thus, the disease has a significant economic cost and reduces the quality of life of caregivers.

For this reason, social workers are also an important part of the multidisciplinary care team.

Conclusions

Many patients with congenital anomalies have an excellent prognosis, so their management by paediatricians is very similar to the routine preventive care offered to healthy children. Patients with more severe anomalies require multidisciplinary follow-up with collaboration of the paediatrician and several specialists. In any case, primary care paediatricians must be aware of these conditions and are able to manage these patients in mild to moderate cases.

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