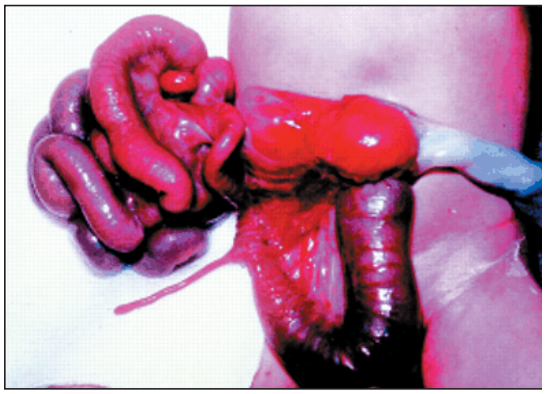
Congenital abdominal wall defects

Background and epidemiology: When the Canadian Congenital Anomalies Surveillance Network holds its second annual scientific meeting in Edmonton, Alta., next week (www.hc-sc.gc.ca/pphb-dgspsp/ccasn-rcsac/ccaswkshop_e.html), one of the main agenda items will be abdominal wall defects. This term refers primarily to gastroschisis (characterized by an intact umbilical cord and evisceration of the bowel through a defect in the abdominal wall, generally to the right of the cord, with no membrane covering) and omphalocele (characterized by herniation of the bowel, liver and other organs into the intact umbilical cord, the tissues being covered by membranes unless the latter are ruptured).[1] Although both are congenital defects of the anterior abdominal wall, they differ in several respects. In cases of gastroschisis, a sac is usually absent, associated anomalies (other than intestinal atresia in 25% of cases and cryptorchism in 31%[1]) are rare, the defect occurs to the right of the umbilicus, and the mother is young (less than 25 years). In contrast, in cases of omphalocele, a sac is present, associated anomalies resulting from chromosomal abnormalities (most commonly trisomy 18) are common, the defect occurs within the umbilicus, and the mother is generally older.[2]

Obtaining accurate data on the frequency of abdominal wall defects is complicated by the fact that, in a certain proportion of cases, the pregnancy is ended by elective termination. Estimates of the birth prevalence of gastroschisis (1 in 10 000 births) and omphalocele (2.5 in 10 000) in Western countries[1,3] are comparable to those of Down's syndrome (14 in 10 000), neural tube defects (6 in 10 000), congenital heart defects (5 in 10 000), orafacial clefts (11 in 10 000) and limb reduction defects (3 to 8 in 10 000).[4] Although the birth prevelance of omphalocele has remained generally stable over the past 20 years,[3,4] reports from Europe, the United States and Japan suggest that the rate of gastroschisis has increased as much as 10-fold over the past decade.[3,5–7] This increase may be partly due to increased detection and ascertainment resulting from increased use of prenatal ultrasonography; however, if this were the only factor, a similar rise in omphalocele rates would be expected. Retrospective analyses of case series of gastroschisis indicate that the risk is greatest for low-income, young mothers who are heavy smokers,[8] who are undernourished[9] and who use over-the-counter medications with vasoactive properties (e.g., pseudoephedrine, phenylpropanolamine, ephedrine, methylenedioxymethamphetamine) in early pregnancy.[8,9] The cause of gastroschisis is multifactorial and seems to involve vascular disruption of the fetal mesenteric vessels.

Clinical management: An abdominal wall defect is often diagnosed during routine prenatal ultrasonography. In early embryogenesis, the intestines are normally extra-abdominal, returning to the abdominal cavity by 11 weeks' gestation; therefore, a diagnosis of abdominal wall defect should be delayed until 14 weeks' gestation to allow for possible errors in gestational dating. Advances in maternal biochemical screening for open neural tube defects have aided in the antenatal diagnosis of fetal abdominal wall defects. Both gastroschisis and omphalocele are associated with elevation of maternal serum α-fetoprotein (MSAFP). The median value for MSAFP is 9.42 multiples of the median (MOM) in gastroschisis and 4.18 MOM in omphalocele.[3]

Once an abdominal wall defect has been identified, a careful ultrasonographic assessment for associated structural anomalies and, in some cases, amniocentesis for karyotype analysis should be undertaken. The prognosis for a child with omphalocele is related to the presence of associated chromosomal and structural anomalies; in cases with associated cardiac defects the mortality rate approaches 80%.[1] The prognosis for an infant with gastroschisis is determined primarily by the condition of the exteriorized bowel; the contemporary mortality rate is around 8%.[10]



Gastroschisis defect in an infant. Reproduced with permission of the BMJ Publishing Group.

Debate exists over the optimal timing (preterm versus term) and mode (cesarean section versus vaginal) of delivery.[1,2] In general, bowel dilatation is an indication for preterm delivery.[2] Upon delivery, the most pressing concern with gastroschisis is continuing fluid loss from the exposed viscera. Recommended management has usually consisted of urgent reduction in the operating room. Overly aggressive attempts at primary closure can greatly increase abdominal pressure, resulting in impaired ventilation and vascular compromise and leading to bowel perforation and necrotizing enterocolitis.[1] To avoid these problems, a prosthetic silo can be sutured around the defect. This permits gradual return of the intestines to the abdominal cavity over 5 to 10 days as the cavity slowly enlarges. In omphalocele, unlike gastroschisis, the extruded abdominal contents are covered by a 2-layer membrane, so fluid losses and electrolyte stability are often more manageable. Cardiac defects often require evaluation before operative treatment of the omphalocele.[1]

Routine prenatal ultrasonography and determination of MSAFP levels permit early diagnosis and mobilization of the multidisciplinary team of obstetricians, geneticists, neonatologists and

pediatric surgeons needed to manage these common congenital anomalies.

Prevention: The rising incidence of gastroschisis reinforces the importance of messages about smoking cessation and adequate nutrition during pregnancy. The possible role of over-the-counter vasoactive medications requires closer examination.

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Patient self-care: helping patients make therapeutic choices

Carole Repchinsky, Christine LeBlanc, editors

Ottawa: Canadian Pharmacists Association; 2002. 896 pp. $125 ISBN 1-894402-03-0

Rating: ***
Audience: Pharmacists, physicians, nurses

Content: In general practice, I'm often most able (and willing) to help those patients who are both able and willing to help themselves. However, empowering patients to problem-solve safely and effectively remains a major challenge, and I'm always on the lookout for tools that will help. This book caught my attention with its theme of teaching patient self-care, and I wasn't too disappointed. Written almost exclusively by pharmacists and pitched mostly at front-line health care providers (particularly other pharmacists), its 65 chapters serve as brief but solid overviews on many unglamorous ailments (e.g., canker sores, infant colic, corns, calluses and bunions, periodontal disease) as well as some more serious concerns (depression, headache, menopause). The chapters, which are generally well referenced, feature many tables and some illustrations and follow a set template: pathophysiology of the condition, common goals of therapy, advice on assessing patients with the problem, and non-pharmacologic and pharmacologic therapies (including herbal and other alternative therapies). A wide breadth of topics relevant to primary care is covered, with nicely done chapters on traveller's health, infant nutrition and infections such as scabies, lice and pinworms. The numerous patient handouts are useful, but an absence of illustrations makes them visually drab.

Limitations: This book presents only sparse evidence for many of the treatments that are recommended, particularly the complementary and alternative therapies. When evidence is cited, the level of evidence is absent. Information on the adverse effects of therapies is also scant. Given these limitations, I wonder whether this book would really enable me to have truly meaningful discussions about informed consent with patients. I was also baffled at times as to why some treatments were discussed in the first place, if they were not being recommended by the authors. For example, the chapter on diabetes states that "current evidence does not support the use of herbal remedies in diabetes." However, the next paragraph goes on to describe how one could use chromium, *Gymnema sylvestre*, ginseng and konjac-mannan to lower blood sugar levels. Despite its non-evidence-based approach and some truly odd parts (like the section on effective patient interactions, which recommends using the term "bum" instead of "rectum"), overall, I do recommend this book. Its selection of down-to-earth topics and its practical tone make it a useful tool for physicians helping patients to help themselves.

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This book is available through your local book retailer or through the publisher at www.pharmacists.ca/function/shopper/Index.cfm?RptCategPassed=Web&RptCodePassed=Pub. It is also available through the Canadian Medical Association at www.cma.ca

Items reviewed are rated on a 4 star scale (4 = excellent)