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Fetal and perinatal implications of anomalies in the gastrointestinal tract and the abdominal wall

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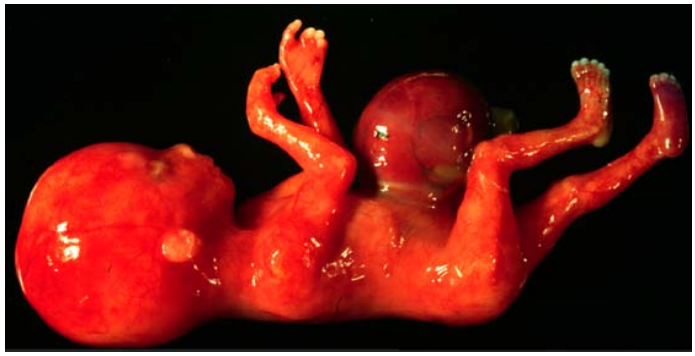

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SUMMARY

Background

Anomalies in the gastrointestinal tract and abdominal wall are relatively common. The present studies include the diagnoses of gastroschisis, omphalocele, imperforate anus, esophageal and duodenal obstruction. In all these conditions, the newborns are in need of postnatal surgery. A prenatal diagnosis may improve the outcome by optimizing the pre- and postnatal care for the newborn. Historically, the clinical course has been based on postnatally diagnosed cases and, until recently, knowledge has been lacking about the intrauterine development and thereby the possibility for optimal surveillance for certain diagnoses. There is a wide variety in the accuracy and sensitivity of detecting different anomalies prenatally.

The aims of the studies were

Selected population

To describe the outcome from the time of prenatal diagnosis into the postnatal period and to identify risk factors for poor outcome and determine whether surveillance may be beneficial. An additional aim was to describe the rate of prenatal diagnosis and elucidate possible strategies for improved diagnosis.

Non-selected population

To describe the incidence, prenatal detection rate and accuracy of the diagnoses in our non-selected population.

Materials and methods

The study populations of duodenal obstruction, gastroschisis and omphalocele were followed from the time of a prenatal diagnosis, through birth and into the postnatal period. The studies of imperforate anus and esophageal obstruction were based on all cases that had been through at least one prenatal ultrasound examination at the National Center for Fetal Medicine at any time during pregnancy and been given the diagnosis either pre- or postnatally.

All studies were based on a selected population consisting of referred cases in addition to our own non-selected population. The non-selected population was also investigated separately.

Results and comments

Duodenal obstruction

The study comprised 29 prenatally diagnosed fetuses. Trisomy 21 was present in 21%. Altogether, associated anomalies were present in 18/29 (62%). Isolated duodenal obstruction, with an expected good outcome, was present in 11/29 (38%). Of these, 5/11 (45%) died in utero or had a substantially impaired neurological development most probably due to intrauterine asphyxia. Our hypothesis is that a vagal overactivity due to distension in the lower esohageal tract leads to bradycardia, asphyxia and in some cases even asystolia and death. Fetal duodenal obstruction is a serious condition with a high risk of adverse outcome, also in cases with normal karyotype and no other anomalies. Surveillance of fetuses with duodenal obstruction is difficult because the fatalities seem to occur instantly. The prenatal detection rate in the *non-selected population* was high (82%). The incidence was approximately 1:3500. There was one false positive case.

Gastroschisis

Sixty-four fetuses with a prenatal diagnosis of gastroschisis were included. All cases had normal karyotype. Associated anomalies were present in 6%. Thirteen infants (22%) were small for gestational age. Prenatal intra-abdominal dilatation of the bowel was associated with obstruction of the bowel. Pathological CTG indicating fetal distress and leading to emergency CS developed in 22% of the fetuses already prior to labor. Intensive CTG monitoring of fetuses with gastroschisis in the third trimester may reduce the risk for fetal distress and IUFD. Of the 64 cases, 56 (87.5%) survived and all had a life without any significant gastrointestinal problems at follow-up over a period of 6-180 months. The prenatal detection rate in the *non-selected population* was 100% and there were no false positive cases. The incidence was 1:2290.

Omphalocele

The outcome of 90 fetal cases with omphalocele was poor with only 8 (9%) infants alive and healthy. The rate of associated anomalies was high (89%). Omphaloceles were subdivided into epigastric, central and hypogastric based on the location of the defect. The fact that abnormal karyotype was present in 69% of fetuses with central omphaloceles compared to 13% of fetuses with epigastric omphaloceles ($P < 0.0001$) leads us to consider the possibility that central and epigastric omphaloceles are different entities. When a thorough diagnosis was made, it was possible to identify a subgroup with fatal/poor prognosis. Surveillance should be focused on those whose prognosis is believed to be good. In the *non-selected population* the prenatal detection rate was 95% and there were no false positive cases. The incidence was 1:2000.

Imperforate anus

The study comprised 69 cases with imperforate anus. The prenatal detection rate was low (16%). Abnormal karyotype was present in 13%. Altogether, associated anomalies were frequent (86%) and imperforate anus was commonly a part of various syndromes. The outcome was poor with only 35% survival; survival for cases with isolated imperforate anus or only one additional anomaly was 94%, compared to only 14% for cases with more than one additional anomaly. The future detection rate may be improved by heightened awareness of the condition and of the type of anomalies commonly seen with imperforate anus in addition to the ability to recognize the most typical ultrasound findings in imperforate anus. In the *non-selected population,* the incidence was approximately 1:1800; a prenatal diagnosis was made in 11% and there were no false positive cases.

Esophageal obstruction

Forty-eight cases with esophageal obstruction were investigated and the prenatal detection rate was 21/48 (44%). The clinical signs of polyhydramnios were the most important factors for prenatal detection of esophageal obstruction. Consequently, the time of diagnosis was late (median 32+0 weeks). In all prenatally diagnosed cases, the stomach was small or not visible. In addition, an esophageal proximal pouch was visualized in 9/21 (43%) of the cases. Of all 48 cases, abnormal karyotype was present in 23%. Altogether 38/48 (79%) had

associated anomalies. Survival was higher, but not significantly, for cases with isolated esophageal obstruction. An increased awareness of the possibility of esophageal obstruction leading to targeted examinations whenever the suspicion is raised during pregnancy might improve the prenatal detection rate and thereby the outcome. In the *non-selected population*, the incidence was approximately 1:3500, the prenatal detection rate was 43% and there were no false positive cases.

Conclusion

For all the investigated diagnoses, the accuracy of the prenatal diagnoses was high. The prenatal detection rate in the *non-selected population* was high for cases with duodenal obstruction (82%), gastroschisis (100%) and omphalocele (95%) while the detection rate for esophageal obstruction was relatively low (43%) and for imperforate anus very low (11%). Fetal populations are different from neonatal populations. It is important to rule out cases with lethal associated anomalies. Adequate surveillance of the remaining cases gives a possibility to improve the outcome. The surveillance at our center has substantially changed for cases with duodenal obstruction and gastroschisis as a direct consequence of these studies. Knowledge of the development during intrauterine life must be further improved to be able to optimize the surveillance and care for fetuses with various anomalies.

FUTURE ASPECTS

Our aim is to provide a better start for the newborn individual through identifying fetal disease and treating the problem. The most attractive scenario concerning fetal anomalies would be the prevention of occurrence. The increased knowledge in biotechnology and molecular genetics might make that possible for some of the anomalies. A number of anomalies are now mapped genetically and for an increasing number of anomalies we have possibilities for early DNA diagnosis. For families with hereditary conditions, this may reduce their anxiety regarding their pregnancies. However, we will continue to be faced with the unexpected diagnosis of fetal anomalies. A correct, detailed prenatal primary diagnosis and the ability to rule out associated diagnoses will therefore still be the basis for treatment and surveillance. Fetal diagnoses may be made earlier and earlier as ultrasound becomes more widespread and offered to more women earlier in pregnancy**.** In addition, the technology is constantly improving and the 3D ultrasound technique and MRI may be valuable in some cases, complementary to 2D ultrasound. However, if women are to be able to benefit from increased use of prenatal diagnosis we must have the knowledge and skills to offer high quality examinations.

Information and counseling will be of even more importance with earlier diagnoses and easier access for termination of pregnancy. In this context, the socioeconomic situation in the country and the availability of health care for different conditions will play a role.

In cases of diagnosis of a fatal condition, the knowledge and information to parents will be of increasing importance. To dare to become pregnant again, knowing about risks and possibilities is important for the woman's/parents' psychological wellbeing.

Fetal medicine is still in an early phase of development and extensive future research is needed to determine how to best diagnose and treat the fetus.

Link to fulltext (Norwegian)