Dissertation

Evaluation of the Motor Activity, Cardiopulmonary Performance Capacity and QoL in Patients Born with a Congenital Abdominal Wall Defect

submitted by

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Abbreviations

AWD.....Abdominal Wall Defect
BMI.....Body-Mass-Index
BR.....Breathing Reserve
CPET .....Cardiopulmonary Exercise Performance Testing
DKT.....Dordel-Koch-Test
EMG.....Electromyography
EQO_2.....Ventilatory Equivalent for Oxygen
FEV_1.....Forced Expiratory Volume in One Second
GIQLI .....Gastrointestinal Quality of Life index
GS.....Gastroschisis
HR.....Heart Rate
IQR.....Interquartile Range
OC .....Omphalocele
OSAS .....Observer Scar Assessment Scale
POSAS.....Patient and Observer Scar Assessment Scale
PSAS .....Patient Scar Assessment Scale
RER .....Respiratory Exchange Ratio
TPN.....Total Parenteral Nutrition
VO_2 .....Peak Oxygen Uptake

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Abstract in German

Einleitung: Das Überleben von Patient*innen mit angeborenen Bauchwanddefekten (abdominal wall defects, AWD) hat sich in den letzten Jahrzehnten deutlich verbessert, aber es fehlen umfassende Untersuchungen, die strukturiert die kardiopulmonale Leistungsfähigkeit, motorische Fähigkeiten, Bauchwandfunktion sowie patient*innenberichtete Ergebnisse gemeinsam erfassen.

Methoden: Patient*innen, welche zwischen 2002 und 2013 mit AWD geboren wurden, wurden in die Studie eingeschlossen und mit einer alters-, geschlechts-, BMI- und aktivitätsgematchten gesunden Kontrollgruppe verglichen. Die Untersuchungen inkludierten Spirometrie, kardiopulmonalen Belastungstest, motorische Leistungsdiagnostik, Oberflächenelektromyographie, Sonographie der Bauchwand, Stand- und Ganganalyse sowie Erhebungen zur gastrointestinalen Lebensqualität und kosmetischen Zufriedenheit.

Ergebnisse: 18 Patient*innen mit AWD sowie 18 Kontrollen wurden eingeschlossen. Lungenfunktion, kardiopulmonale Leistungsfähigkeit sowie Stand- und Ganganalyse zeigten keine signifikanten Unterschiede zwischen den Gruppen. Im Gegensatz dazu war die motorische Leistungsfähigkeit (Dordel-Koch-Test) in der AWD-Gruppe signifikant reduziert (AWD: Median 3,54/IQR 1 vs. Kontrolle Median: 2,8/IQR 1; p=005). Die Oberflächenelektromyographie zeigte großteils eine vergleichbare Muskelaktivierung, obwohl Unterschiede im Seitenvergleich bei zwei statischen Untersuchungen aufgefallen sind. Der gastrointestinale Lebensqualitäts-Score war in der AWD-Gruppe signifikant niedriger (AWD: Ø137,2 ± 6,8 vs. Kontrolle 141,4 ± 4,9; p=0,038). Die kosmetischen Ergebnisse wurden insgesamt überwiegend positiv bewertet.

Diskussion und Schlussfolgerung: Patient*innen mit AWD zeigen eine erhaltene kardiopulmonale Leistungsfähigkeit und Bauchwandmorphologie, weisen jedoch relevante Einschränkungen der motorischen Leistungsfähigkeit sowie der gastrointestinalen Lebensqualität auf. Diese Ergebnisse unterstreichen die Bedeutung einer strukturierten, multidisziplinären Langzeitnachsorge, die über die rein anatomische Rekonstruktion hinaus funktionelle Aspekte berücksichtigt, um Einschränkungen frühzeitig zu erkennen und gezielte Interventionen einzuleiten.

Abstract in English

Introduction: Survival of patients with congenital abdominal wall defects (AWDs), including gastroschisis and omphalocele, has improved substantially over recent decades. While short-term outcomes are well documented, data on long-term functional performance and quality of life remain limited and fragmented. In particular, comprehensive assessments combining cardiopulmonary performance, motor abilities, abdominal wall function, and patient-reported outcomes are lacking.

Methods: Patients with a history of gastroschisis or omphalocele diagnosed between 2002 and 2013 were evaluated and compared with age-, sex-, BMI-, and activity-matched healthy controls. Assessments included spirometry, cardiopulmonary exercise testing, standardized motor performance, surface electromyography, ultrasound of the abdominal wall, stance and gait analysis, gastrointestinal quality of life, and cosmetic satisfaction.

Results: 18 AWD patients and 18 controls completed the full study protocol. Motor performance as measured by the Dordel–Koch test was significantly lower in the AWD group, especially in tasks involving coordination and trunk control (AWD median 3.54 [IQR 1] vs. control median 2.8 [IQR 1]; p = 0.005). Anthropometry, pulmonary function, and cardiopulmonary exercise performance as well as gait and stance analyses did not differ significantly between patients and controls. Surface electromyography revealed comparable muscle activation between groups, but side-to-side asymmetries were observed in AWD patients. Gastrointestinal quality of life score was significantly lower in patients (AWD mean 137.2 ± 6.8 vs. control mean 141.4 ± 4.9; p=0.038). Cosmetic outcomes were generally rated favorably.

*Discussion and Conclusion***:** Patients born with AWDs demonstrate preserved cardiopulmonary capacity and abdominal wall morphology but exhibit significant impairments in motor performance and gastrointestinal quality of life. These results highlight the importance of a structured, multidisciplinary long-term follow-up to allow early detection of impairments to identify limitations early and initiate targeted interventions.

1 Introduction

Congenital abdominal wall defects (AWD), such as gastroschisis (GS), omphalocele (OC), ectopia of the heart, exstrophy of the bladder and cloaca, and Pentalogy of Cantrell are rare congenital abnormalities affecting the ventral abdominal wall. While various forms exist, GS and OC are the most frequent, with incidences of 4.5 per 10,000 and 0.6-4.8 per 10,000 live births, respectively (2, 3).

1.1 Gastroschisis

GS is a congenital anomaly of the ventral body wall, first described in 1733 by James Calder (4). It is characterized by the presence of a hole in the abdominal wall, which allows the intestinal loops, and sometimes parts of the colon and other organs to eviscerate (**Figure 1**). The intestine is directly exposed to amniotic fluid, with consecutive swelling and possible damage to the seromuscular layer (5, 6).

GS primarily occurs to the right of the umbilical cord insertion, though it can occasionally be found on the left side (7, 8). Unlike other ventral body wall abnormalities such as OC, ectopia of the heart, exstrophy of the bladder and cloaca, and Pentalogy of Cantrell, GS is not distinctively located in the midline.

Figure 1: Gastroschisis on day of birth shortly before surgical repair.

1.1.1 Prevalence, Embryology and Etiology

Over the past decades, the prevalence of GS has been on the rise. Kirby et al. reported that its occurrence nearly doubled in the United States between 1995 and 2005 (9). In Europe, the prevalence has increased in the last decades and is currently estimated at approximately 3 per 10,000 live births, in contrast to reported declines in cases in Colombia and China (10-12). To better understand true prevalence trends on a global level, standardized data collection through registries, and close collaboration between healthcare professionals, such as within ERNICA (European Reference Network for Rare Inherited Congenital Anomalies) are required (13).

The exact etiology of GS remains unclear, but it is believed to have multifactorial pathogenesis including genetic, environmental, and vascular factors affecting fetal development. Various theories have been summarized by Chuaire Noack (11). Three principal mechanisms are described: disturbances in embryonic folding or anatomical development, ischemic events, such as those caused by occlusion of the right vitelline artery, and rupture of the amniotic membrane during physiological umbilical herniation between the 6th and 10th weeks of gestation (11). Each mechanism leads to weakening of the abdominal wall close to the umbilical cord insertion, resulting in herniation of uncovered bowel into the amniotic cavity. Although these mechanisms differ in their underlying pathophysiology, they share the concept of a localized developmental insult rather than a generalized failure of abdominal wall formation (14).

Several epidemiological studies have identified factors associated with an increased risk of GS. One of the strongest associations is maternal age, with teenage mothers showing the highest prevalence (2, 15-17). It has been hypothesized that younger maternal age might be linked to environmental exposures or specific genetic susceptibilities that influence embryonic development (16). In addition, maternal smoking, alcohol consumption, and recreational drug use, particularly vasoactive substances such as cocaine and amphetamines, have been implicated in increasing risk of GS (16, 18, 19).

Environmental influences alone, however, are unlikely to fully explain the occurrence of GS, and genetic contributions have therefore been explored. Familial clustering reported in a small number of cases suggests a potential hereditary component (20). In a systematic review by Salinas-Torres et al. described the importance of not focusing solely on environmental factors, even though genetic causes are unlikely to be the sole explanation for GS. Their study identified four single nucleotide polymorphisms in three genes. These genes are all involved in blood pressure regulation. Based on these findings, they propose that vascular pathogenesis could be the most plausible explanation for the development of GS

(21). A study by Marquart et.al. concludes that the cause of GS remains unclear but is most likely due to complex gene–environment interactions and further studies are needed to clarify specific genetic mechanisms (22).

1.1.2 Classification and Associated Malformations

Most of the GS cases occur as isolated anomalies and are classified as isolated or simple GS (23).

In 10-15% other malformations can be found and GS is classified as complex (23-26). The most common associated anomalies involve the gastrointestinal tract. Intestinal atresia, a condition where a segment of the intestine is absent or closed, occurs in approximately 10-15% of cases. Other complications such as malrotation, stenosis, and perforation are also frequently observed, potentially affecting postoperative outcomes and nutritional management (25). Extraintestinal associated malformations are very scare in contrast to other AWD (24, 27).

Cardiovascular defects have also been identified in GS cases, with a prevalence of around 2.5%. These range from minor septal defects to more severe congenital heart diseases (24). In fact, the prevalence of minor septal defects seems to be the same in children without GS. Therefore, Danish authors propose if this “associated” minor heart defects without any symptoms might be detected in extended screenings in patients born with GS compared to patients without GS (25).

The presence of associated malformations in infants with GS can significantly influence clinical management and prognosis. Early identification through detailed prenatal imaging and postnatal assessment is crucial for planning surgical interventions and ensuring appropriate multidisciplinary care. Recognizing potential associated anomalies enables better parental counseling and helps anticipate complications (24, 25).

1.1.3 Prenatal Diagnosis and Management

GS is mostly diagnosed prenatally through routine ultrasound examinations. Advances in prenatal imaging over recent decades have significantly improved early detection, and management of this condition (12). The key sonographic feature is the presence of free-floating bowel loops within the amniotic cavity, without a covering membrane. This characteristic distinguishes GS from OC, where herniated organs are enclosed within a membranous sac. Prenatal ultrasound demonstrates a high diagnostic accuracy for GS, with detection rates reported to be approximately 97.7% in isolated cases (28).

More than just confirming diagnosis, prenatal ultrasound is essential for assessing the condition of the herniated bowel. Sonographic findings such as bowel dilation, wall thickening, and polyhydramnios can indicate complications such as bowel atresia, stenosis, or ischemia. Identification of these features is clinically relevant, as complex GS is associated with increased morbidity and mortality (29).

Although the optimal frequency of prenatal surveillance remains unclear, many centers schedule their patients for serial ultrasound examinations to assess bowel appearance and amniotic fluid volume over time. Regarding the mode of delivery, current evidence does not indicate a clear benefit of elective cesarean section over vaginal delivery. As a result, the mode of delivery should be based on standard obstetric indications, with planned delivery at a tertiary care center equipped for neonatal surgical intervention (30-33).

The ERNICA Consensus Statement in Europe suggests vaginal birth between 37+0 and 39+0 weeks in children with uncomplicated GS. The panel recommends assessing fetuses with GS by ultrasound based on a combination of parameters rather than reliance on individual markers. Evaluations shoul include bowel wall thickness, gastric dilation, herniation of the stomach and/or bladder through the abdominal wall defect, the presence of polyhydramnios, fetal growth parameters, fetal movements, and the size of the abdominal wall defect. In addition, both intra-abdominal bowel diameter and extra-abdominal bowel diameter should be considered to improve overall risk stratification (12).

The prognosis for infants with GS has improved significantly in the last decades, with survival rates now exceeding 90% in high-income countries. However, outcomes depend largely on the presence, and severity of bowel injury or associated anomalies. Early and accurate prenatal diagnosis allows for timely multidisciplinary planning, optimizing perinatal management, and improving neonatal outcomes (28).

Prenatal management includes neonatal, anesthesia, and pediatric surgery counseling for the parents to optimally prepare them for the necessary steps after delivery.

1.1.4 Postnatal Management

Delivery should be planned in a tertiary care center after appropriate prenatal counseling. After delivery, the resuscitation team should include pediatric surgeons when immediate interventions to prevent bowel damage become necessary (34, 35). The existence of protocols for a rare disease like GS has improved the management quality, morbidity and mortality (36, 37).

The exposed bowel should be protected by placing the lower part of the neonate's body into a bowel bag (34, 38, 39). Fluid and temperature management is also crucial in these patients due to the elevated fluid and temperature loss through the exposed bowel (38, 40). Initial stabilization should include orogastric tube to decompress the stomach and prevent excessive swallowing of air and thus bowel distension (38). After stabilization, the infant should be brought in right lateral decubitus position to prevent twisting of the mesenteric vascular pedicle (27).

1.1.5 Surgical Approach and Postoperative Complications

The primary objectives in the surgical management of GS include the safe reintegration of the herniated abdominal contents, management of bowel complications—particularly in complex cases—and definitive closure of the AWD, all while minimizing the risk of abdominal compartment syndrome. Closure might be achieved early (within approximately 6 hours of birth) using either sutured or sutureless techniques, or it might be delayed following gradual reduction of the viscera using a prosthetic silo. However, the most effective and optimal technique for defect closure in GS remains uncertain and represents an ongoing gap in clinical knowledge (41).

Because the AWD in GS typically measures only 2–3 cm, initial reduction can be technically challenging. As illustrated in **Figure 2** (17.2), the defect is enlarged superiorly by incising the fascia under direct protection of a finger placed beneath the abdominal wall to avoid bowel injury. Superior extension is preferred, as inferior extension risks injury to the bladder, which lies directly below the defect (42).

After safe enlargement, the midgut can be reduced gradually into the peritoneal cavity (**Figure 2** (17.3)). Reduction is performed gently to minimize venous congestion and avoid twisting or kinking of the mesenteric vessels, especially in cases where the bowel is edematous or covered by a fibrinous peel. Once complete reduction is achieved, primary fascial closure can be attempted (42).

If tension permits, primary closure is performed by suturing the rectus fascia with absorbable or non-absorbable material depending on the degree of tension. **Figure 2** (17.4) illustrates placement of interrupted figure-of-eight sutures into well-defined fascial edges. A thin ribbon retractor might be placed beneath the fascia to prevent accidental incorporation of bowel during suture tying (42).

A key technical point is appropriate suture placement lateral to the umbilical ring to prevent postoperative umbilical defects—one of the most common long-term sequelae after GS repair (43).

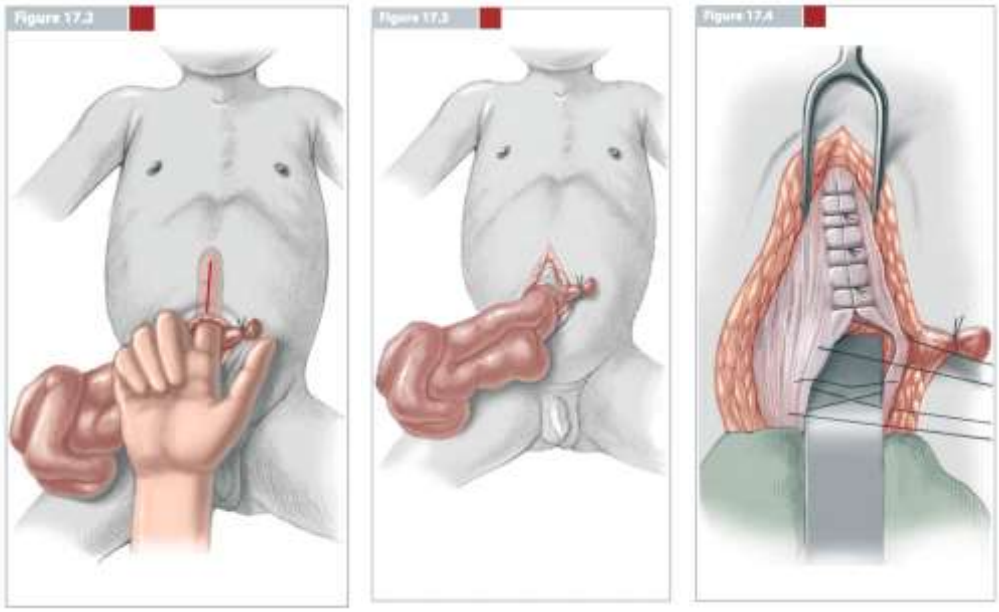

Figure 2: Primary Reduction (17.2), fascial enlargement (17.3) and primary fascial closure (17.4) Sources: P. Puri; M. Höllwarth, Pediatric Surgery, Edition 1, 2006, Springer Nature. Reproduced with permission from Springer Nature.

When primary closure is not feasible—due to bowel edema, a small abdominal cavity, or intolerance to reduction—silo placement is the preferred technique. As shown in **Figure 3** (17.5), a reinforced silastic sheet can be sutured circumferentially to the fascia using horizontal mattress sutures of 3-0 silk, creating a temporary silo. The sheet is then wrapped around the herniated viscera and secured using a continuous running suture (42).

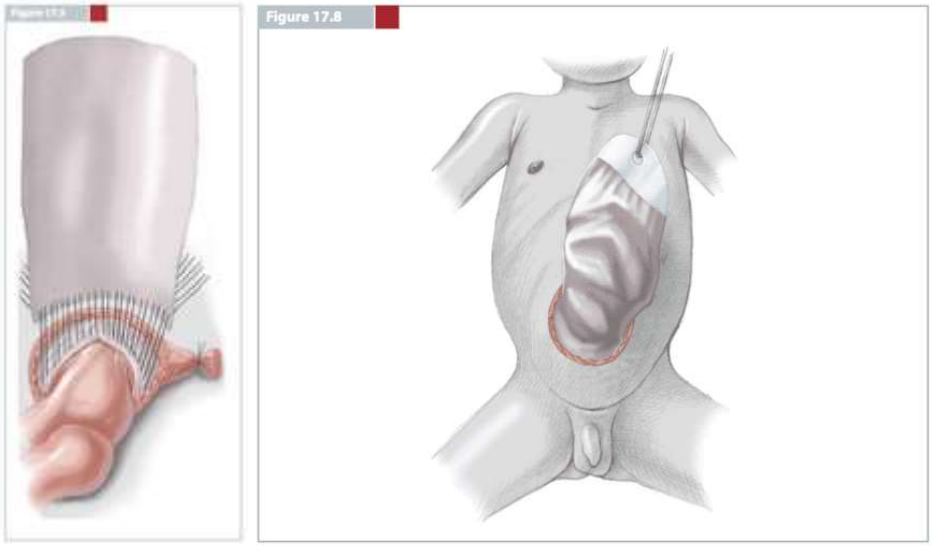

Figure 3: Use of a Hand-Sewn or Preformed Silo (17.5 and 17.8) Sources: P. Puri; M. Höllwarth, Pediatric Surgery, Edition 1, 2006, Springer Nature. Reproduced with permission from Springer Nature.

The preformed spring-loaded silo (**Figure 3** (17.8)) has become the most widely used modern approach. It can be placed at the bedside without general anesthesia and has been associated with increased rates of successful fascial closure, fewer ventilator days, more rapid return of bowel function and lower complication rates such as intestinal venous congestion or infection (44, 45). Daily gradual reduction is performed by compressing the silo contents and advancing the silastic ring to progressively reduce the bowel until complete reduction allows definitive fascial closure (42).

In the last years, discussion about sutureless closure of the abdominal wall bed side either with general anesthesia or analgesia with sedation is widely discussed in cases of uncomplicated GS (46, 47). Sutureless repair involves the initial manual reduction of the herniated bowel into the abdominal cavity, after which the residual umbilical cord is positioned over the defect and secured with an occlusive dressing to facilitate natural epithelialization and closure (48). A retrospective study of the Midwest pediatric surgery consortium showed reduced need for general anesthesia, fewer courses of antibiotics, a lower incidence of superficial, deep surgical site infections, and shorter durations of mechanical ventilation (49).

Importantly, these improvements occur without prolonging the time to initiation of enteral feeding, achievement of full feeds, or overall length of hospitalization in appropriately selected patients. Authors mentioned that longitudinal follow-up of these patients is needed to better define long-term outcomes and potential complications (49).

The variety in the literature suggests that no single operative strategy is universally superior. Outcomes depend primarily on patient selection and defect characteristics (50). Primary closure is favored when reduction is easily tolerated, whereas staged silo reduction offers greater safety in infants with viscero-abdominal disproportion, severe bowel edema, or hemodynamic instability. Sutureless closure represents an additional option in selected cases and might reduce anesthesia exposure and postoperative wound morbidity.

Common complications are prematurity, central line sepsis, necrotizing enterocolitis, and wound infections. Because of the potentially long time until achieving full enteral feeding, total parenteral nutrition (TPN) is often required which itself can lead to TPN-related complications (51-53).

Infections, like postsurgical wound infections and central line sepsis, are the most common complications in patients born with GS. These infections increase morbidity and costs in these patients (54-56). Postsurgical infection rate is reported up to 32% in initial hospital stay using the Clavien-Dindo-Classification (54, 57).

Delayed closure, mostly defined as closure later than 24 hours postnatally, seems to be a risk factor for postsurgical infection. Several studies indicate that delayed closure is associated with higher infections rates (55, 58-60).

1.1.6 Long-Term Sequelae of Gastroschisis

Advances in neonatal intensive care and pediatric surgery have markedly improved the survival of infants born with GS. As mortality has decreased, the focus of research and clinical care has shifted toward long-term outcomes, including growth, gastrointestinal function, neurodevelopment, motor performance, and overall quality of life.

Growth and Physical Development

Infants with GS are typically born preterm as well as small for gestational age, and many experience prolonged hospital stays due to feeding intolerance and the need for parenteral nutrition. De Bie et al. reported that 81% of infants with GS had weights below the 10th percentile at discharge; however, most children demonstrated catch-up growth within the

first three years of life. This recovery was most consistent among those with simple GS, whereas children with complex GS often remained smaller throughout childhood (61, 62).

Persistent nutritional vulnerability, feeding difficulties, and short bowel syndrome are predominantly seen in complex GS and are major determinants of long-term morbidity (63). These patients often require extended follow-up by a multidisciplinary team.

Gastrointestinal Morbidity

Gastrointestinal problems represent the most common long-term sequelae. A study shows that at a median follow-up of four years, 34% of children had gastroesophageal reflux, 33% reported chronic constipation, and 4% experienced fecal incontinence (61). Small bowel obstruction and adhesive ileus have been reported in approximately 3% of patients overall, with a higher incidence observed among those with complex GS.

A recent Swedish study from 2024 confirmed that children with complex GS have a significantly higher risk of intestinal failure and adhesive bowel obstruction compared to those with simple GS (64). Moreover, many long-term survivors report intermittent abdominal pain or episodes of subileus decades after repair, emphasizing the need for life-long clinical awareness of adhesive disease as adhesions and volvulus can occur years after the initial repair (26, 63-65). Although relatively infrequent, small bowel obstruction remains one of the leading causes of hospital readmission.

Neurodevelopment and Motor Function

Neurodevelopmental outcomes in GS patients have become a growing area of interest. A systematic review found that data remain limited but suggest a slightly increased risk of neurodevelopmental delay, attention-deficit symptoms, and mild motor coordination problems in some cohorts (66). Early neonatal factors, such as prolonged hospitalization, sepsis, or parenteral nutrition, might contribute. Preterm birth and low birth weight, both common in GS, contribute independently to later neurodevelopmental and metabolic vulnerability (62, 66).

Gorra et al. observed that at two years of age, GS patients showed no major cognitive impairment compared to other neonatal intensive care survivors, but long-term studies beyond preschool age are scarce. Current evidence indicates that most children with simple GS achieve normal cognitive development, although light motor and coordination deficits might persist into adolescence (66, 67).

Genitourinary Sequelae – Cryptorchidism

Cryptorchidism (undescended testes) is markedly more common in boys with GS than in the general population (68). A 2022 systematic review and meta-analysis reported a pooled prevalence of ~19% (95% CI 13–26) among boys with GS and detailed the anatomical presentations (non-palpable, prolapsed through the defect, and palpable testes). Early conservative observation might allow spontaneous descent in a subset, while approximately half of the patients ultimately require orchidopexy; outcomes after appropriately timed surgery are generally good (69).

The literature suggests that rates might be higher in some cohorts (~30%) as prematurity, larger AWD, and “complex” GS correlate with increased risk. These studies also note that spontaneous descent can occur during infancy, supporting early surveillance followed by timely orchidopexy if descent is not achieved by 12–18 months (68, 70, 71).

Testicular position should be routinely assessed during follow-up examinations and patients and parents should be informed about the need for early surgical management if spontaneous descent does not occur.

Quality of Life (QoL)

Some studies demonstrated good long-term health-related QoL, with most children and adolescents perceiving themselves as healthy and active (72). These findings highlight that psychosocial adjustment is usually satisfactory, provided early medical and psychological support is available (73, 74).

In De Bie et al.’s cohort, children with simple GS had PedsQL™ scores that were slightly higher than those of healthy controls (61). Nevertheless, children with complex GS or those who underwent multiple operations reported lower physical health scores and some cosmetic concerns related to abdominal wall scarring.

While majority of children with simple GS achieve normal growth, and development, those with complex defects show increased risk for gastrointestinal, neurodevelopmental, and genitourinary complications. Therefore, a structured, multidisciplinary long-term follow-up program, including pediatric surgery, pediatric gastroenterology, nutrition, pediatric urology, and developmental medicine, is essential to identify long term complications early and optimize overall quality of life.

1.2 Omphalocele

OC is a congenital defect of the anterior abdominal wall characterized by herniation of abdominal viscera into the base of the umbilical cord, covered by a three-layered sac consisting of amnion, Wharton's jelly, and peritoneum (6, 75). The umbilical cord inserts at the apex of the sac, distinguishing OC from GS, where the defect is paraumbilical and lacks a protective membrane (**Figure 4**).

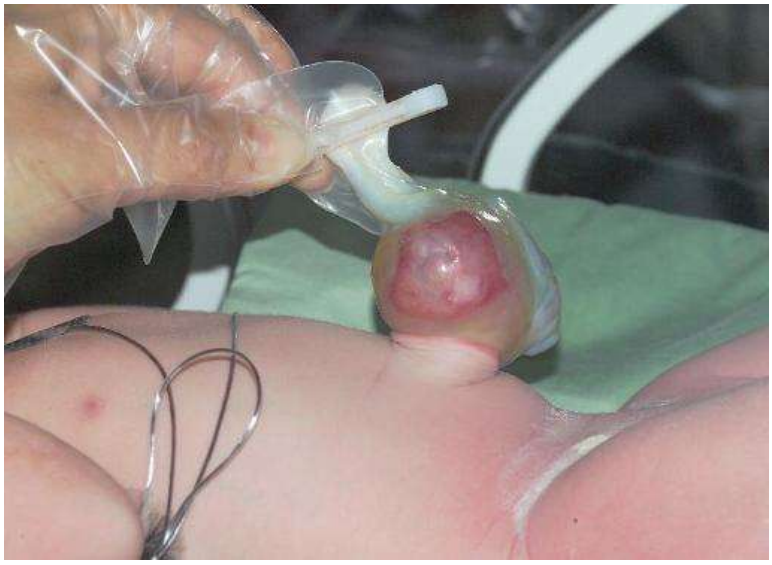

Figure 4: Omphalocele at intensive care unit.

1.2.1 Prevalence, Embryology and Etiology

Contemporary population-based studies estimate the prevalence of OC to range between 0.6 and 4.8 per 10,000 live births, with reported differences largely reflecting variations in registry inclusion criteria and regional reporting practices (76-78). In contrast to the rising incidence of GS, the prevalence of OC has remained stable or even slightly decreased during the past decades. This trend might occur due to improvements in prenatal diagnostic accuracy and the increased detection of severe cases during early gestation(77, 78).

The etiology of OC remains not fully understood. Literature consistently notes that there is no consensus regarding the precise embryological mechanisms leading to OC, with multiple theories proposed and ongoing debate among clinicians and embryologists (79, 80).

Two main embryologic mechanisms have been proposed for the development of OC. The first is failure of the midgut to return to the abdominal cavity after its physiological herniation into the umbilical cord between the 6th and 10th week of gestation. During normal development, the rapidly elongating midgut herniates into the umbilical cord from weeks 6–10 and by weeks 10–12, it returns to the abdominal cavity as the body wall closes in the midline (81, 82). The second proposed theory is about abnormal body folding or fusion, in which incomplete closure of the cephalic, caudal, or lateral folds prevents proper formation of the ventral abdominal wall (81, 83). These mechanisms are discussed in recent reviews and are supported by embryological studies, but definitive evidence for either remains lacking (79, 80, 82).

However, these classical embryologic explanations do not fully account for the wide phenotypic variability observed in clinical practice. Increasing evidence suggests that genetic, epigenetic, and environmental factors act in combination to disrupt ventral body wall development (77, 78, 84). Chromosomal abnormalities, imprinting defects, and altered gene expression during early folding have all been implicated (85, 86). Despite ongoing advances in molecular genetics, the precise developmental mechanisms leading to OC remain incompletely defined, reflecting the heterogeneous and multifactorial nature of this congenital anomaly.

Maternal risk factors reported in epidemiologic analyses include advanced maternal age, pre-existing diabetes, and obesity (77, 87, 88). Overall, OC arises from a multifactorial interplay of developmental arrest, chromosomal or epigenetic defects, and maternal influences.

1.2.2 Classification and Associated Malformations

OCs can be usually categorized according to size and content. A small OC generally contains only intestinal loops, whereas a large or “giant” OC contains the liver and might exceed 5 cm in diameter(63, 89-91). Another useful classification distinguishes between isolated and non-isolated OCs. Isolated cases occur without additional malformations or chromosomal anomalies and tend to have a more favorable prognosis. In contrast, non-isolated OCs are associated with genetic or structural abnormalities and carry a higher risk of perinatal mortality(75, 78, 92). Prenatal differentiation between these groups is crucial for counseling and management.

OC is frequently associated with additional congenital anomalies, reflecting its complex developmental origin. Approximately 50–70% of cases present with one or more associated malformations and in about half, these anomalies determine prognosis more than the OC itself (75, 78, 93).

The most common accompanying abnormalities involve the cardiac, chromosomal, and genitourinary systems. Congenital heart disease occurs in up to 50% of affected infants, most commonly ventricular septal defect, atrial septal defect, tetralogy of Fallot, and left ventricular outflow tract anomalies (83). Chromosomal abnormalities are present in roughly 30–50% of cases, with trisomy 13, 18, and 21 being the most frequent (83, 93, 94). Other structural malformations might include neural tube defects, limb anomalies, and cleft palate.

Syndromic associations are well documented. The two most prevalent are Beckwith–Wiedemann syndrome and Pentalogy of Cantrell. Beckwith–Wiedemann syndrome, resulting from dysregulation of imprinted genes at chromosome 11p15.5, is characterized by OC, macroglossia, visceromegaly, and hemi-hyperplasia (85, 86). Pentalogy of Cantrell comprises a combination of midline defects: OC, lower sternal defect, anterior diaphragmatic defect, pericardial defect, and intracardiac malformation(95). Other syndromic links include the OEIS complex (omphalocele–exstrophy–imperforate anus–spinal defects), trisomy 18 (Edwards syndrome), and trisomy 13 (Patau syndrome) (96-98).

The size and content of the OC strongly influence perinatal management and prognosis. A study demonstrated that the OC–to–thoracic ratio obtained by prenatal imaging correlates with postnatal respiratory outcome and survival (15). Isolated small OCs generally have an excellent prognosis, whereas complex or syndromic OCs carry increased risk for preterm delivery, respiratory distress, and neonatal mortality (2,6,13).

1.2.3 Prenatal Diagnosis

Persistence of herniated viscera beyond this developmental stage, particularly when accompanied by a covering membrane and a central umbilical cord insertion, is considered diagnostic for OC (76, 99, 100). First-trimester ultrasound already allows differentiation from physiological midgut herniation, which resolves by the 12^{th} week. Persistence of herniated viscera beyond this point, especially when associated with a membrane and a central cord insertion, confirms the diagnosis (99). In addition to standard ultrasound, advanced imaging techniques such as fetal magnetic resonance imaging are increasingly applied in cases of large or complex OC. These modalities allow more detailed assessment of sac contents, fetal lung volume, extent of liver herniation, and the risk of pulmonary hypoplasia (101, 102).

Information obtained from advanced prenatal imaging supports individualized counselling and plays an important role in delivery planning, especially in fetuses with giant OC (103, 104).

Given the high prevalence of chromosomal and syndromic abnormalities, comprehensive fetal evaluation, including a detailed anatomic survey and the offer of diagnostic genetic testing should follow diagnosis of OC. Aneuploidies, most commonly trisomy 13, 18, and 21, are frequent, and in addition the presence of increased nuchal translucency or multiple associated anomalies further raises suspicion of underlying chromosomal disease

Once the diagnosis of OC has been established, multidisciplinary counselling involving neonatology, pediatric surgery, and clinical genetics is crucial. Serial growth assessments are advised throughout pregnancy. Whenever possible, delivery should be planned in a tertiary perinatal center with immediate access to neonatal intensive care and pediatric surgical expertise (99, 100).

1.2.4 Postnatal Management

Postnatal management of infants with OC begins with immediate stabilization after birth. The herniated viscera require protection using warm, sterile, moisture-retaining dressings or transparent coverings to minimize evaporative heat and fluid loss, and careful positioning is necessary to avoid external pressure on the defect (99). Routine nasogastric decompression is routinely performed to minimize gastric distension and resulting respiratory compromise. Infants with giant OC often present with respiratory insufficiency resulting from pulmonary hypoplasia and limited abdominal domain, which may necessitate early ventilatory support and cautious transfer to intensive care or the operating room, depending on clinical stability (105).

Because OC is frequently associated with additional congenital anomalies, a systematic postnatal workup is essential. This includes cardiac, abdominal, and cranial ultrasound, as well as genetic assessment when syndromic features are present. Data from large institutional cohorts highlight the high rate of associated cardiac and renal malformations, which substantially influence surgical timing, perioperative risk and early morbidity (91).

Postoperative management focuses on careful monitoring for respiratory deterioration, hemodynamic instability, and the development of abdominal compartment syndrome. Infants with giant OC often require prolonged ventilation and delayed enteral feeding. Early postoperative challenges include ileus, gastroesophageal reflux, feeding intolerance, and the need for extended parenteral nutrition. Several studies have identified low birth weight, cardiac

anomalies, reduced fetal lung volume, and ruptured sacs as key predictors of postoperative complications and prolonged hospitalization (91).

Respiratory management is a central component of early postoperative care. Limited pulmonary reserve due to pulmonary hypoplasia frequently necessitates mechanical ventilation in infants with giant OC. Studies correlating prenatal imaging findings with postnatal outcomes have demonstrated that reduced fetal lung volumes are associated with longer durations of ventilation and more complex postoperative recovery (105).

Ruptured OC represents a neonatal emergency requiring immediate intervention. Management priorities include rapid resuscitation, thermoregulation, fluid replacement, and the early administration of broad-spectrum antibiotics, followed by urgent surgical treatment. Despite modern intensive care, ruptured sacs remain associated with higher morbidity and mortality (106).

Overall, postnatal management of OC demands a tailored, physiology-driven, multidisciplinary approach. Across the spectrum of disease severity, optimized respiratory support, thorough diagnostic evaluation, and vigilant postoperative monitoring are essential to improving neonatal outcomes.

1.2.5 Surgical Approach and Postoperative Complications

The surgical management of OC aims to achieve safe reduction of the herniated viscera and tension-free closure of the abdominal wall while avoiding excessive intra-abdominal pressure. Surgical strategy is primarily determined by defect size, viscero-abdominal disproportion, associated anomalies, and the infant's physiological stability. In small and medium-sized defects, primary fascial closure shortly after birth is often feasible and is associated with a low risk of abdominal compartment syndrome when reduction is performed gently and without undue tension (99). In contrast, giant OC poses significant challenges due to limited abdominal domain, reduced thoracic volume and varying degrees of pulmonary hypoplasia. Early primary closure in such cases is rarely feasible and might result in hemodynamic compromise, respiratory failure, or abdominal compartment syndrome (99, 107). If primary closure is not feasible due to viscero–abdominal disproportion, multiple staged approaches have been described, including prosthetic silos, temporary synthetic patches such as Gore-Tex and cutaneous flap techniques (108, 109). Elevated intra-abdominal pressure following surgical reduction can impair diaphragmatic excursion, reinforcing the need for individualized planning and staged repair when intolerance to primary closure is anticipated (90).

In small and moderate OCs without significant liver herniation, primary fascial closure shortly after birth is usually feasible. After opening or excising the sac, the bowel and liver are gently reduced, and the fascial defect is closed in the midline. The skin is then adapted with or without an umbilicoplasty, as illustrated in **Figure 5** (16.2, 16.3) (42).

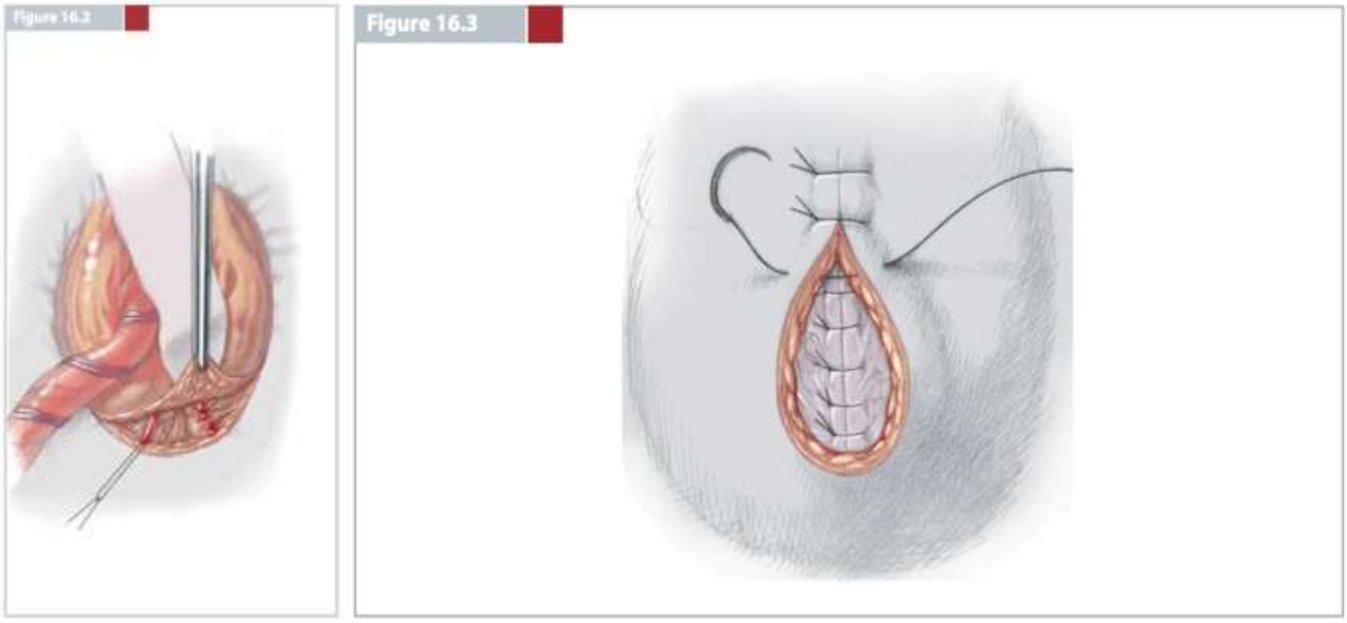

Figure 5: Excising sac (16.2) and primary closure (16.3) Sources: P. Puri; M. Höllwarth, Pediatric Surgery, Edition 1, 2006, Springer Nature. Reproduced with permission from Springer Nature.

When primary fascial closure is not possible but there is sufficient skin to cover the viscera, staged skin-only closure is an option. In this approach, the sac is partially or completely excised, wide subcutaneous undermining allows advancement of bilateral skin flaps over the viscera and the fascia is either left open or bridged with a prosthetic patch as **Figure 6** (16.6) shows. The infant is left with a planned ventral hernia, which is repaired electively later in childhood. For larger or ruptured OCs, prosthetic silo placement remains a widely used staged surgical technique (110). A Silastic or Dacron silo is sutured circumferentially to the fascial edges **Figure 6** (16.7), allowing gradual daily reduction of the herniated viscera before delayed fascial and skin closure once intra-abdominal pressure is tolerated (42).

In selected infants, component-separation techniques and intra-abdominal tissue expanders have been employed to enlarge abdominal domain and facilitate midline fascial closure, although these approaches require multiple procedures and are generally reserved for specialized centers.

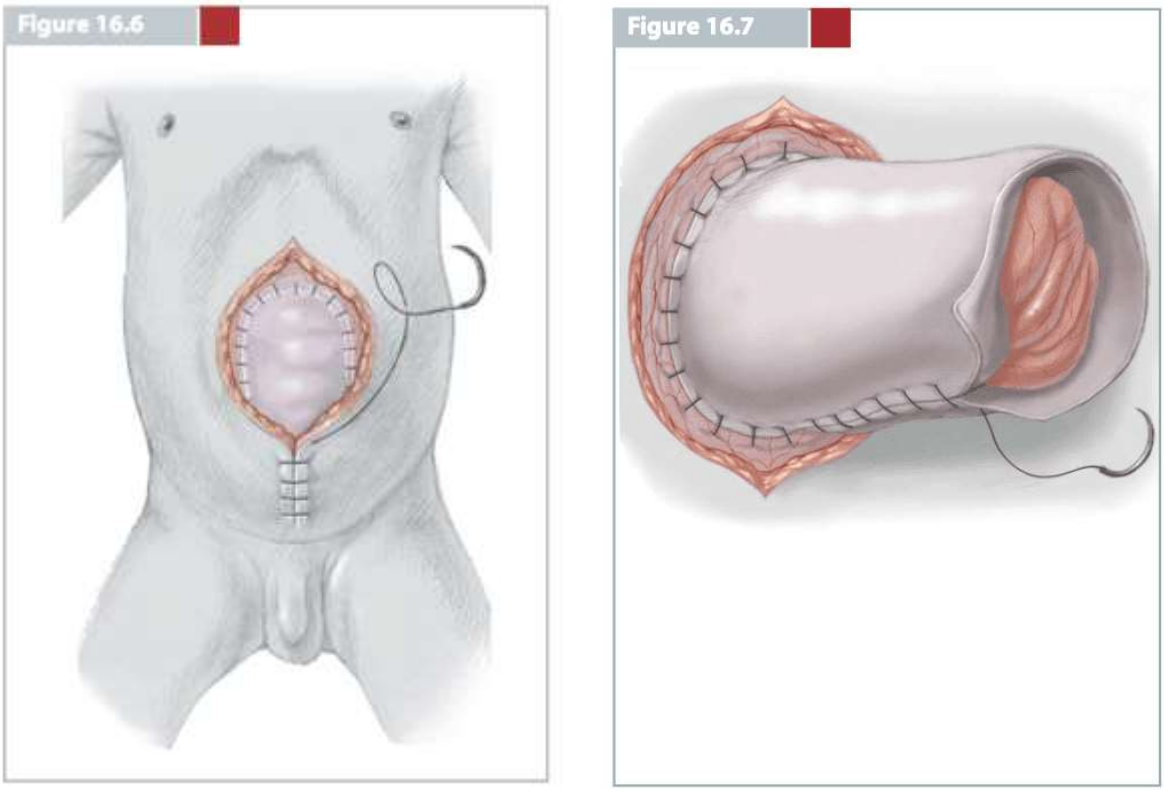

Figure 6: Closure with prosthetic patch (16.6) or Silo technique (16.7) Sources: P. Puri; M. Höllwarth, Pediatric Surgery, Edition 1, 2006, Springer Nature. Reproduced with permission from Springer Nature.

Non-operative “paint-and-wait” management constitutes the main alternative to early surgical reconstruction for giant OC. Here, the intact sac is treated with topical escharotic and antimicrobial agents such as silver sulfadiazine or povidone–iodine, promoting gradual eschar formation, granulation, and epithelialization; definitive repair of the resultant ventral hernia is postponed until later infancy or childhood - **Figure 7** (16.10). A systematic review comparing staged surgical closure with primary non-operative delayed management found similar mortality but shorter time to full enteral feeds in the conservative group, supporting “paint-and-wait” as a valid first-line strategy for many giant OCs. Modifications of conservative care include a serial taping technique in which the intact sac is progressively compressed with adhesive dressings in the intensive care unit allowing gradual reduction and early fascial closure within approximately two weeks in most infants.

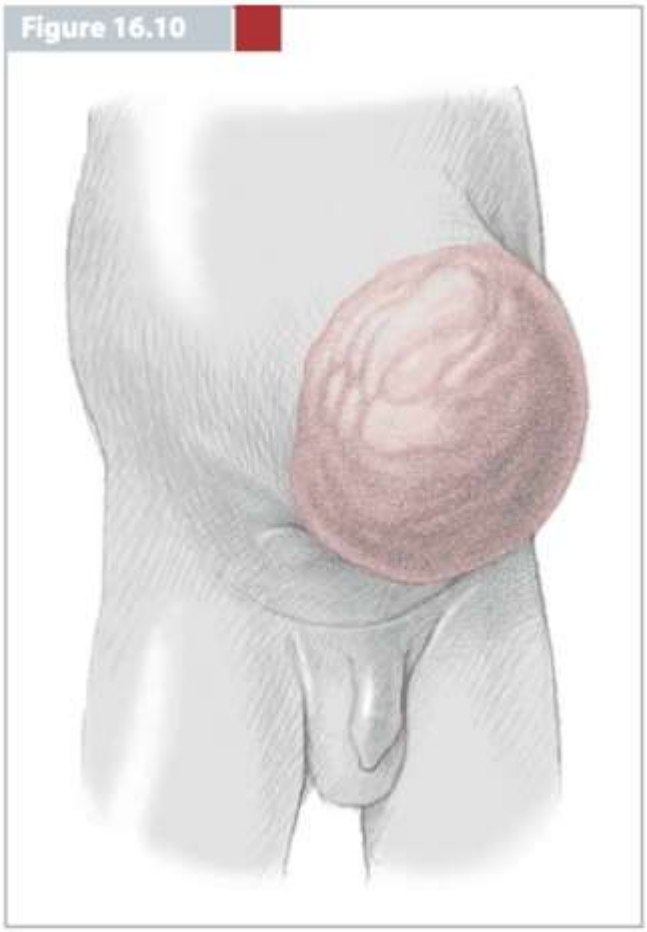

Figure 7: “Paint-and-wait” management of omphalocele. Sources: P. Puri; M. Höllwarth, Pediatric Surgery, Edition 1, 2006, Springer Nature. Reproduced with permission from Springer Nature.

Ruptured OC presents an acute surgical emergency. The absence of the protective sac exposes viscera to contamination, heat and fluid loss, and mechanical trauma. Initial management focuses on resuscitation, sepsis prevention, thermal support, and temporary coverage of the defect (106).

Postoperative complications in OC repair vary by defect size, associated anomalies, and surgical technique. In giant OC, respiratory insufficiency is common due to limited lung volume, and increased intra-abdominal pressure after repair, often resulting in prolonged ventilation and respiratory support (105). Feeding difficulties, failure to thrive, and the need for long-term gastrostomy feeding occur particularly in infants with giant OC or complex anomalies.

Wound-related complications include infection, seroma, dehiscence, necrosis, and, in mesh repairs, enterocutaneous fistula formation following OC repair (91). Hernia recurrence is common when primary closure is attempted under tension or when synthetic mesh is used as a bridging technique, with reported recurrence rates exceeding 50% in some series (111). Compartment syndrome is rare but represents a serious complication and might lead to renal failure, reduced splanchnic perfusion, bowel ischemia, or even loss of bowel, requiring extensive resection (112).

Overall, postoperative outcome is strongly influenced by the presence of associated cardiac, renal, or chromosomal anomalies, the degree of pulmonary hypoplasia and the need for prolonged mechanical ventilation. Infants with isolated OC generally have favorable outcomes, whereas those with complex giant OC or major anomalies experience significantly higher morbidity, prolonged hospitalization, and increased need for multidisciplinary follow-up (90, 105).

1.2.6 Long-Term Sequelae of Omphalocele

Respiratory Morbidity

Long-term respiratory morbidity is one of the most relevant sequelae in children with OC, particularly giant OC. Pulmonary hypoplasia, caused by reduced thoracic volume and viscero-abdominal disproportion, persists beyond the neonatal period and might manifest as chronic respiratory insufficiency, recurrent lower-respiratory-tract infections, reactive airway disease, and exercise intolerance (113). Prenatal studies using three-dimensional ultrasound and fetal MRI consistently show that observed-to-expected lung volumes below 50% predict prolonged ventilation, prolonged hospitalization, and increased risk of long-term oxygen dependency (102, 113, 114).

Several series report chronic pulmonary hypertension, which can persist into infancy and childhood. Pulmonary hypertension incidence varies widely, from 8% to over 50%, depending on patient selection and severity of lung underdevelopment (113, 115). Late-onset pulmonary hypertension has been described even after initial stabilization, sometimes associated with sepsis or chronic lung disease and can necessitate long-term pharmacologic therapy or respiratory support (115). Children with giant OC might remain at risk for recurrent respiratory symptoms, wheezing, sleep-disordered breathing, and need for tracheostomy (116). These findings highlight the need for long-term pulmonology follow-up in this cohort, particularly for infants demonstrating severe neonatal respiratory insufficiency.

Growth and Nutritional Problems

Growth delay and persistent nutritional challenges are frequently observed in children with OC, particularly in those affected by giant defects. Feeding-related problems, including oral aversion and prolonged reliance on enteral or parenteral nutritional support, may extend well beyond infancy and persist into later childhood (63). Previous studies have reported that up to two-thirds of infants with giant OC require long-term enteral feeding assistance, most commonly via nasogastric or gastrostomy tubes, whereas gastrostomy placement is rarely necessary in cases of small OC (63). Feeding dysfunction might be linked to prolonged mechanical ventilation, oral-motor delays, neurologic impairment, and altered gastrointestinal motility. Feeding dysfunction in this population is thought to be multifactorial and has been associated with prolonged mechanical ventilation, delays in oral-motor development, neurological impairment, and alterations in gastrointestinal motility. Consequently, impaired growth or “failure to thrive” has been described in a considerable proportion of affected children, many of whom require ongoing nutritional monitoring and support throughout childhood (115).

Gastrointestinal Complications

Patients with OC are confronted with a broad spectrum of gastrointestinal long-term complications, including gastroesophageal reflux disease (GERD), chronic constipation, recurrent abdominal pain, adhesive bowel obstruction, and intestinal malrotation. The prevalence of GERD among OC survivors exceeds that observed in the general pediatric population, frequently necessitating prolonged medical treatment (63). In severe cases, surgical management is often necessary, especially in infants with associated anomalies or prolonged respiratory morbidity. Severe feeding symptoms, such as vomiting, poor weight gain, or esophagitis, necessitate further evaluation (105).

Late adhesive small bowel obstruction can occur years after neonatal surgery. Long-term series demonstrate a notable incidence of adhesional morbidity, requiring surgical intervention in some patients (92). Malrotation is common and persist even after abdominal wall repair, with a risk of volvulus described in multi-institutional analyses, warranting long-term vigilance for obstructive symptoms (117, 118).

Neurodevelopmental Outcomes

Neurodevelopmental impairment represents an important long-term concern in children with OC, particularly among those affected by giant defects. Several studies employing standardized developmental assessment instruments, including the Bayley Scales, have reported that a substantial proportion of survivors with giant OC exhibit delays across cognitive, motor, and language domains during infancy and early childhood (119). These developmental

delays have been shown to correlate closely with indicators of disease severity, such as pulmonary hypoplasia, pulmonary hypertension, prolonged mechanical ventilation, neonatal sepsis, and the need for tracheostomy(119).

In contrast, children born with small OC generally demonstrate favorable neurodevelopmental outcomes. Large longitudinal cohort studies have reported school attendance and academic performance in this group that are comparable to those observed in the general pediatric population(120).

Cosmetic and Abdominal Wall Sequelae

Cosmetic outcome is an important aspect of long-term follow-up in children with OC, particularly in those presenting with extensive scarring or residual ventral hernias. A study published in 2024 using the Patient and Observer Scar Assessment Scale (POSAS) demonstrated significantly increased scar stiffness, thickness, and irregularity in patients with giant OC when compared with those with smaller defects. While cosmetic results in minor defects are generally favorable, individuals with giant OC more frequently require secondary surgical revision or consultation with plastic surgery specialists (121). Dissatisfaction with aesthetic appearance may adversely affect self-esteem and psychosocial well-being, especially during adolescence. In addition, residual ventral hernias can necessitate delayed abdominal wall reconstruction, and persistent abdominal wall weakness may contribute to reduced core strength or postural impairment (111, 122).

Children born with OC, particularly those affected by giant defects, are at risk for a wide range of long-term morbidities. Despite substantial improvements in survival, a proportion of patients continue to experience chronic health-related challenges. Importantly, many of these complications evolve gradually and may not become evident during infancy, highlighting the dynamic and lifelong nature of the condition. Coordinated care involving pediatric surgery, pulmonology, cardiology, gastroenterology, nutrition, developmental pediatrics, psychology, and physiotherapy facilitate early identification of emerging complications and timely intervention. Such integrated longitudinal care has the potential to improve functional outcomes and quality of life while providing sustained support to affected children and their families.

1.3 Quality of Life and Long-Term Outcome of Patients with AWD – Background, and Rationale for the Present Study

AWDs, particularly GS and omphalocele OC, have been extensively investigated with respect to prenatal diagnosis, neonatal management, and short-term surgical outcomes. By comparison, data addressing long-term functional outcomes and health-related quality of life (QoL) remain relatively scare. Existing studies have described long-term complications, including the need for repeat surgical interventions due to fascial defects or umbilical and incisional hernias (43, 65). In addition, gastrointestinal symptoms such as stool irregularities, recurrent abdominal pain, and repeated hospital admissions related to ileus or subileus have been reported (65). Cosmetic dissatisfaction has also been documented, with approximately half of affected individuals reporting unfavorable aesthetic outcomes in some cohorts (73, 74, 123). Other studies, however, have revealed that children born with an AWD have the same QoL compared with the healthy population (74).

These heterogeneous findings show important gaps in the current understanding of long-term functioning following AWD repair and highlight the lack of standardized outcome measures across published studies. Many reports rely primarily on parent-reported outcomes, focus on isolated organ systems, include small and heterogeneous cohorts, or lack appropriately matched control groups.

Beyond QoL, substantial uncertainty persists regarding long-term physical performance, particularly cardiopulmonary capacity and core muscle function. Research in children with other congenital anomalies, such as esophageal atresia or anorectal malformations, has demonstrated reduced cardiopulmonary exercise capacity and impaired motor performance when compared with healthy controls (124, 125). These findings raise the possibility that similar functional limitations may also be present in individuals with AWDs. To date, however, only a single study has assessed cardiopulmonary performance in patients with large AWDs. That investigation, which included 18 children, reported overall preserved cardiorespiratory function despite the severity of the underlying defects (126). Importantly, no previous study has systematically evaluated abdominal wall muscle morphology, muscle activation patterns, gait and stance characteristics, or objectively measured motor performance in patients with AWDs. This represents a relevant knowledge gap, given the potential long-term impact of early abdominal wall reconstruction on trunk stability and biomechanics. Moreover, although cosmetic outcomes have been addressed in selected studies, parameters such as core strength, motor coordination, and functional trunk stability, dependent on the integrity of the rectus abdominis, oblique, and transversus abdominis muscles, have not been examined in a structured and comprehensive manner.

To address these gaps, we conducted a single-center observational case–control study with a uniquely broad methodological approach. We assesed:

- cardiopulmonary performance using standardized protocols (127)
- static lung function (spirometry)
- detailed gait and stance analysis
- electromyographic activity of the abdominal wall
- ultrasound-based abdominal muscle morphology
- validated assessment of motor abilities by Dordel-Koch-Test (DKT) (128)
- gastrointestinal quality of life using the gastrointestinal quality of life index (GIQLI) (129)
- cosmetic satisfaction using POSAS

No prior study has combined objective biomechanical, cardiopulmonary, morphological, and QoL assessments within the same cohort in AWD patients before. By comparing our patients to an age-, sex-, BMI-, and activity-matched healthy control group, we sought to generate high-quality evidence to clarify whether AWD survivors experience measurable long-term impairments that are not captured in routine clinical follow-up.

Hypothesis

Null hypothesis (H0): Patients born with congenital AWDs do not exhibit impairments in motor abilities compared with matched healthy controls.

Alternative hypothesis (H1): Patients born with congenital AWDs demonstrate reduced motor performance compared with matched healthy controls.

In addition, cardiopulmonary exercise capacity, abdominal wall muscle function, gait and stance parameters, gastrointestinal quality of life, and cosmetic satisfaction were systematically assessed to provide a comprehensive long-term functional characterization of this patient population and to identify potential domains requiring targeted follow-up or intervention.

2 Patients and Methods

2.1 Study Design and Setting

This single-center observational case–control study was carried out at the Department of Paediatric and Adolescent Surgery, Medical University of Graz. The aim was to evaluate long-term motor performance, cardiopulmonary capacity, abdominal wall function, and health-related quality of life in children and adolescents with a history of GS and OC compared with healthy peers.

All clinical investigations and functional tests were performed in the outpatient setting of our pediatric surgical and sports medicine units. To minimize day-to-day variability, each participant completed the entire examination program within one study visit.

The study was conducted in accordance with the Declaration of Helsinki and national regulations in minors. The institutional review board of the Medical University of Graz approved the protocol (EK 32-231 ex 19/20). Written informed consent was obtained from all participants aged ≥8 years and from parents or legal guardians of all minors, with age-appropriate assent from younger children.

2.2 Study Population

The institutional database was screened for all patients with a diagnosis of GS or OC who were born between January 2002 and December 2013 and treated primarily at our department. All individuals meeting these criteria and available for follow-up at the time of investigation formed the initial study population (n=55).

Clinical records were reviewed to confirm the diagnosis, type of AWD, associated anomalies, surgical management, and postoperative course. Perinatal data (gestational age, birth weight, APGAR scores, and details of neonatal management (type and timing of defect closure, need for silo placement or prosthetic patch, duration of mechanical ventilation, length of neonatal intensive care stay) were abstracted from the medical records using a standardized case report form. For each participant, we documented the total number of subsequent abdominal operations, indication (e.g. adhesive small-bowel obstruction, incisional or umbilical hernia, stoma closure), and age at the last surgery. Complications during the first year of life and late complications (beyond 1 year) were recorded separately.

GS cases were categorized as simple or complex according to widely used criteria. Complex GS was defined by the presence of associated malformation such as intestinal

atresia, perforation, necrosis or volvulus requiring bowel resection or stoma formation (23, 26, 130). For OC, we differentiated giant from non-giant defects. In line with recent pediatric surgical literature, a giant OC was defined as an AWD ≥5 cm in diameter (63, 91).

2.2.1 Inclusion and Exclusion Criteria

Patients with major chromosomal aberrations or syndromic diagnoses were not excluded a priori, as these conditions are part of the OC spectrum. At the time of evaluation, children and adolescents were eligible if they had been born with GS or OC and undergone initial repair at our institution and:

- were between 6 and 18 years of age
- were able to understand study instructions and to perform exercise testing
- agreed (together with their legal guardians) to participate and provided written informed consent.

Exclusion criteria were:

- hemodynamically relevant congenital or acquired heart disease (e.g. uncorrected complex cardiac malformations, significant valve disease)
- neurological or psychiatric disorders interfering with motor testing or cooperation
- acute respiratory infection within 2 weeks prior to assessment

Application of these criteria resulted in 18 participating AWD patients (12 with GS, 6 with OC).

2.2.2 Control Group

For each patient, a healthy control subject matched for sex, age (±1 year), body mass index (BMI; ±2 kg/m²), and habitual physical activity level was recruited. Potential controls were from social circles of hospital staff. None had a history of congenital malformation, chronic pulmonary or cardiac disease, abdominal surgery or neuromuscular disorder. Matching for habitual activity was based on a short questionnaire asking about the frequency of organized sports or vigorous play (categories: “daily”, “several times per week”, “once per week” or “less than once per week”).

2.3 Study Protocol

All examinations were scheduled in the morning or early afternoon. Participants were advised to refrain from strenuous exercise on the day before the visit and to consume only a light meal at least 2 hours before cardiopulmonary exercise testing. Medication was not routinely withheld, and any regular drugs (e.g. inhaled bronchodilators) were documented.

After arrival, a brief medical history was taken, focusing on respiratory symptoms, exercise tolerance, gastrointestinal complaints, and musculoskeletal problems. Participants were instructed to classify their physical activity into one of four categories: “daily,” “several times per week,” “once per week,” or “once per month. This was followed by the measurement sequence described below.

2.3.1 Anthropometry, Body Composition and Laboratory Parameters

Body height and weight were measured with participants barefoot and wearing light clothing, using a wall-mounted stadiometer and a calibrated digital scale. BMI was calculated as weight divided by height squared (kg/m²).

Segmental multi-frequency bioimpedance spectroscopy (Combyn™ ECG device, Academic Technologies at the Institute of Cardiovascular Medicine, Graz, Austria) was used to estimate total body water, extracellular fluid, fat mass and appendicular skeletal muscle mass according to the manufacturer’s algorithms and published validation data (131). The measurements were obtained in the supine position after at least 5 minutes of rest (**Figure 8**).

A 12-lead resting electrocardiogram (ECG) and non-invasive brachial blood pressure were recorded to exclude relevant arrhythmias or arterial hypertension prior to exercise testing.

If patients agreed, a blood sample was taken to a standard serum vial for determination of liver enzymes.

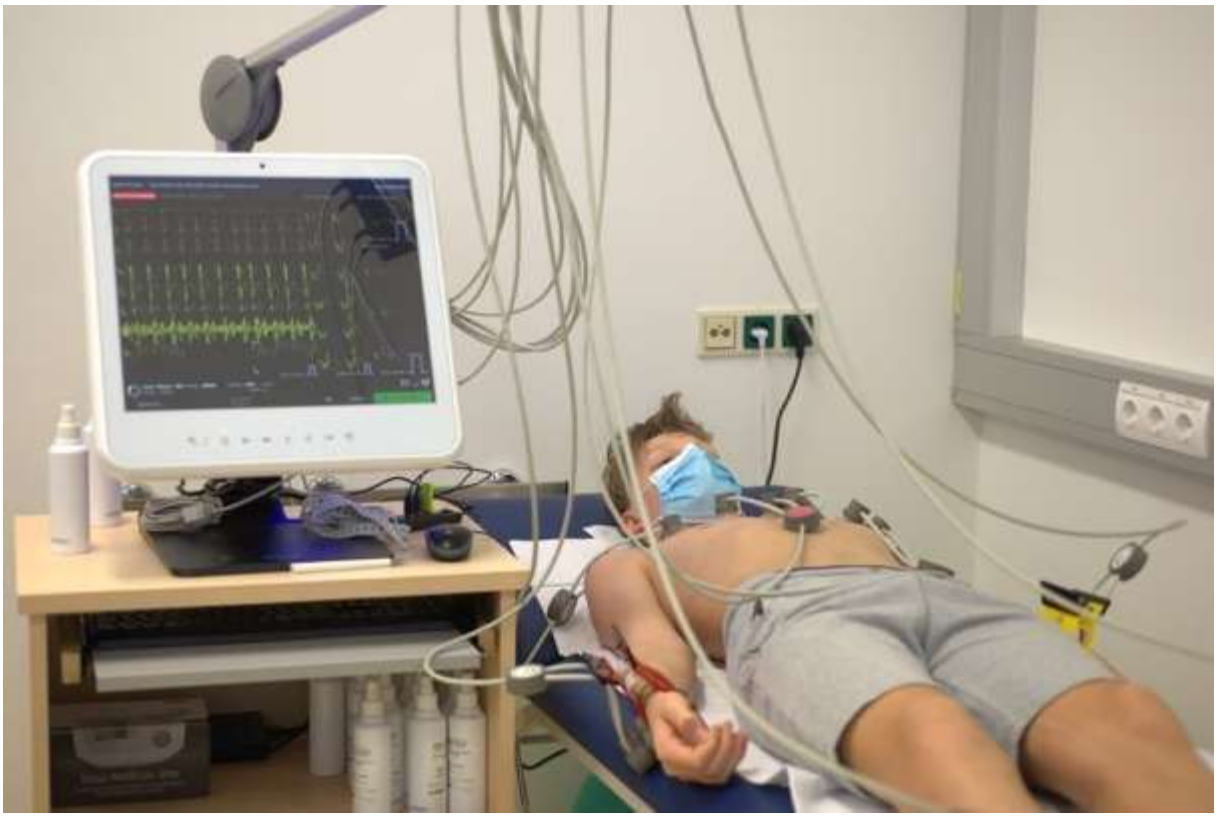

Figure 8: Recording ECG and body composition.

2.3.2 Pulmonary Function Testing

Lung function was assessed with a pediatric spirometer (Oxycon Pro®, Carl Reiner GmbH, Vienna, Austria). After instruction and demonstration, children performed at least three technically acceptable forced expiratory maneuvers in the sitting position, following international recommendations for spirometry in children.

The highest values of forced vital capacity (VC_max) and forced expiratory volume in the first second (FEV_1) were used for analysis. The Tiffeneau index (FEV_1/VC_max, %) was calculated. Predicted values were derived from age-, height-, and sex-specific reference equations. A pattern with reduced VC_max and normal or increased FEV_1/VC_max was classified as restrictive, whereas a reduced FEV_1/VC_max with or without reduced FEV_1 indicated an obstructive pattern (132).

2.3.3 Cardiopulmonary Exercise Performance Testing (CPET)

CPET was performed on an electronically braked cycle ergometer suitable for children (Excalibur Sport®, Lode, Groningen, Netherlands). Participants sat upright with appropriate saddle and handlebar adjustment. Breath-by-breath gas exchange and ventilatory parameters were recorded during the exercise (**Figure 9**).

After a resting phase of 3 minutes sitting quietly on the ergometer, subjects started pedaling at 60 revolutions per minute (rpm) with an initial workload of 20 Watt (W) (younger children) or 25 W (older children and adolescents). Resistance was then increased in a stepwise ramp protocol every minute, using increments adapted to age, sex, and anticipated fitness in accordance with previously validated pediatric protocols (127). The test was terminated when the child reached volitional exhaustion, was unable to maintain the target cadence (>60 rpm) despite encouragement, or when clinical symptoms (e.g. chest pain, severe dyspnea, dizziness) occurred.

The following variables were derived:

- peak oxygen uptake (peak VO_2, ml/kg/min), defined as the highest 30-s average of VO_2
- oxygen pulse (O_2/Heartrate (HR))
- respiratory exchange ratio (RER=VCO_2- /VO_2)
- ventilatory equivalent for oxygen (EQO_2=minute ventilation ($\dot{V}E$)/oxygen uptake (VO_2))
- ventilatory reserve index (breathing reserve (BR)/FEV_1%)

A cool-down phase of 3 minutes at the starting workload followed each test. Heart rate was monitored continuously by 12-lead ECG; peripheral oxygen saturation was recorded with a finger pulse oximeter. Capillary blood samples (20 µl) from the heparinized earlobe were taken before exercise, at the end of each workload step, immediately after exhaustion and at the end of the cool-down period to measure lactate concentration using an enzymatic amperometric method (Biosen C_line®, EKF Diagnostics, Cardiff, UK).

Relative performance capacity was expressed as the percentage of age- and sex-specific reference values for maximal workload on cycle ergometry (133). A peak RER >1.10 was considered indicative of a maximal effort (134).

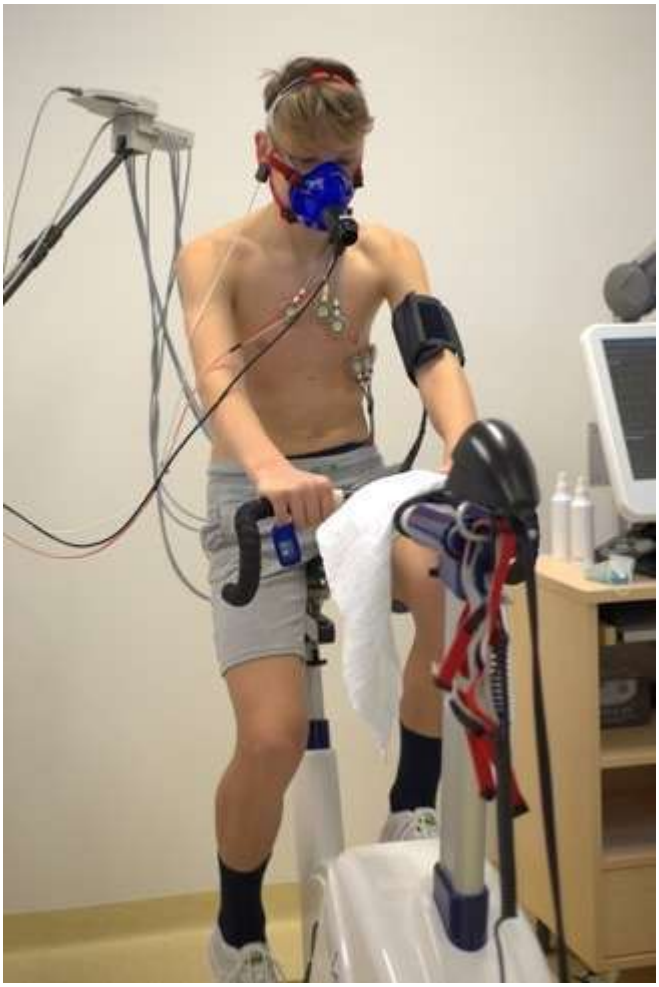

Figure 9: Patient performing CPET.

2.3.4 Motor Performance: Dordel–Koch-Test

Global motor performance and basic motor skills were assessed using the Dordel–Koch-Test, a standardized test battery developed for school-aged children (128).

The DKT comprises the following subtests (**Figure 10**):

- Lateral jumping *(coordination and speed)*
- Sit-and-reach test *(flexibility of the posterior chain)*
- Sit-ups in 40s *(trunk flexor endurance)*
- Standing long jump *(explosive leg power)*
- One-legged stand with eyes open *(static balance)*
- Push-ups in 40s *(upper body strength)*
- Endurance run

In our study, the endurance component was replaced by the CPET results, because cycle ergometry provided a more detailed and objectively measured assessment of endurance capacity. For each item, age- and sex-adjusted grades from 1 (excellent) to 6 (poor) were assigned according to published normative data and an overall DKT score was calculated as the arithmetic mean of all items. Lower scores indicate better motor performance (128).

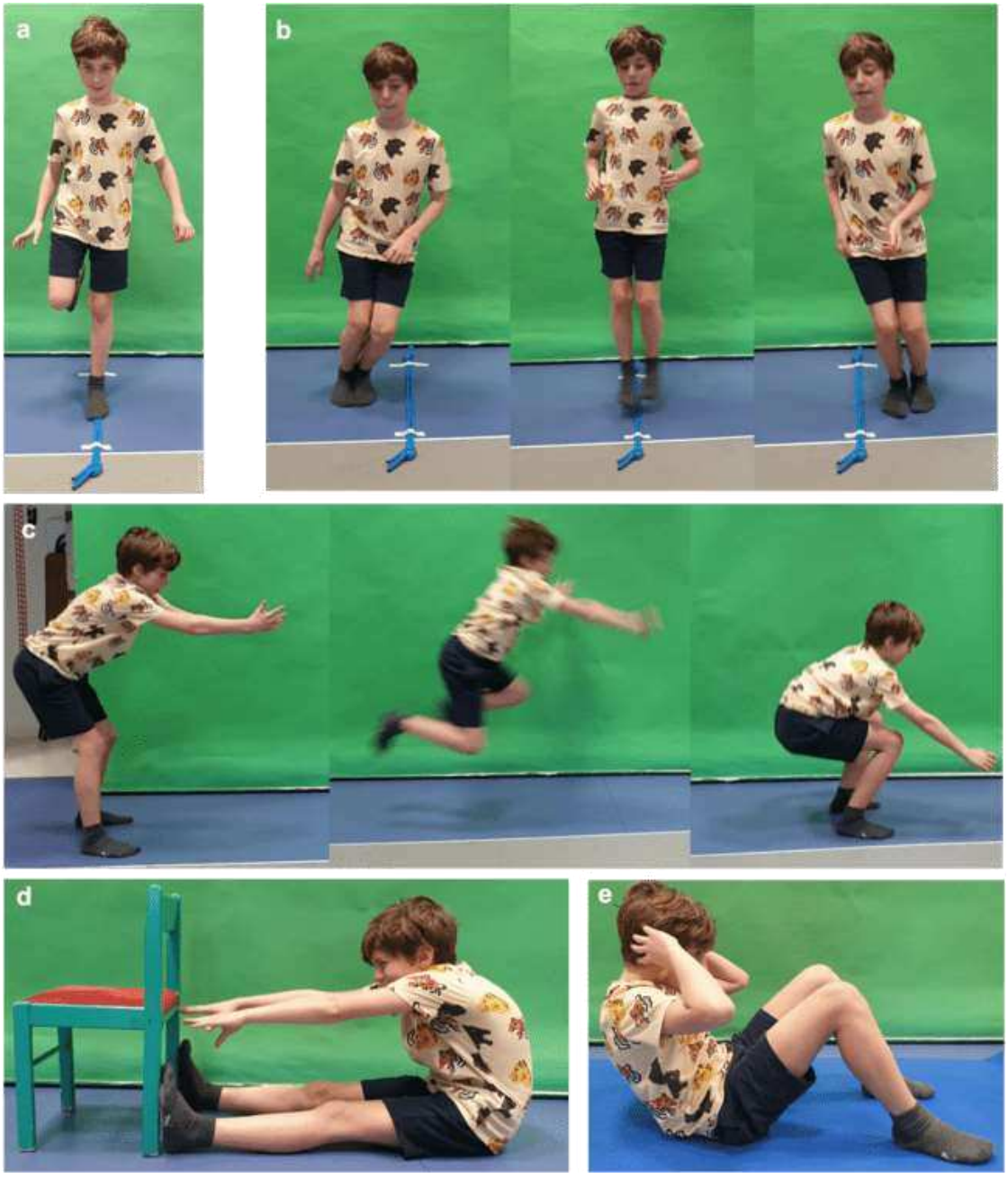

Figure 10: Dordel-Koch-Test - (a) One-legged stand (b) lateral jumping in 40s (c) standing long jump (d) Sit-and-reach test (e) Sit ups in 40s.

2.3.5 Surface Electromyography of the Abdominal Wall

To evaluate abdominal wall muscle activation, wireless surface EMG (Ultium® system, Velamed GmbH, Cologne, Germany) was used (135, 136). Eight self-adhesive bipolar electrodes were positioned bilaterally over the rectus abdominis, the external and internal oblique muscles, and the transversus abdominis (**Figure 11**). After skin preparation, electrode placement was checked visually, and by test contractions.

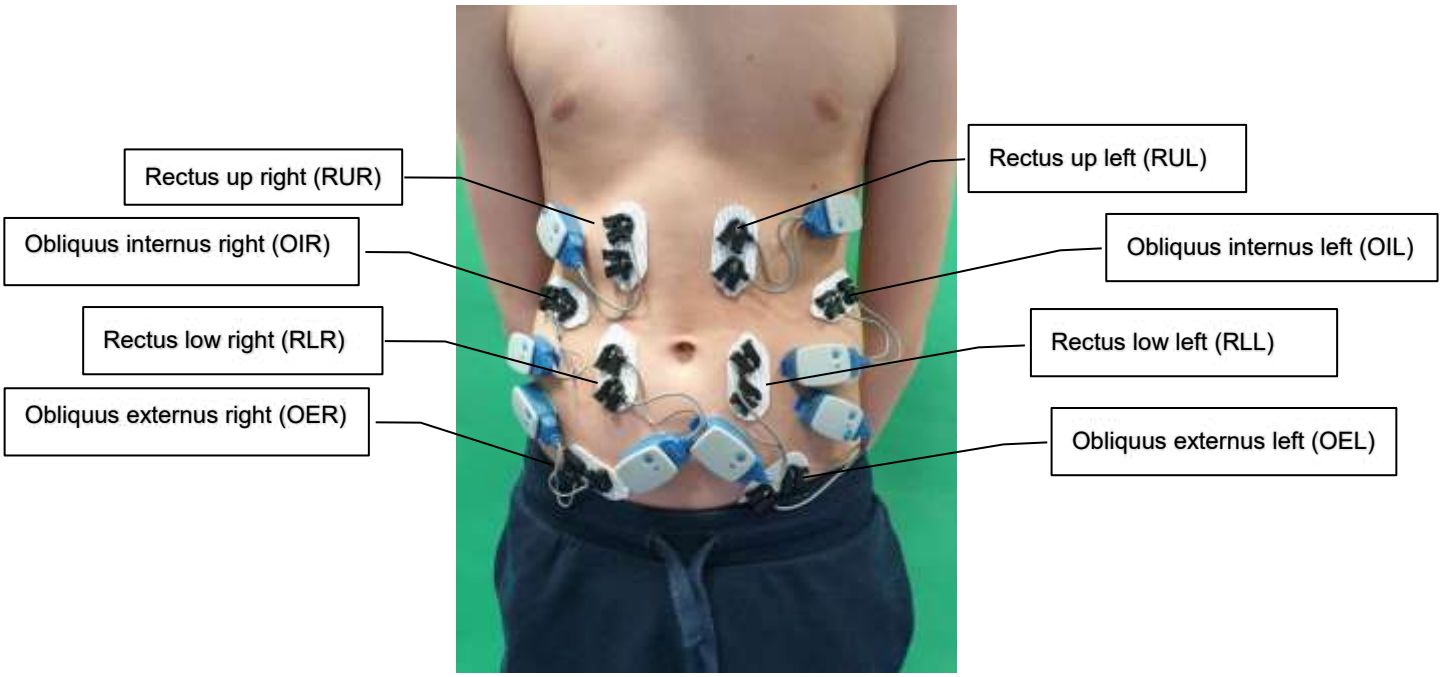

Figure 11: Placement of self-adhesive bipolar electrodes for surface EMG(1). Licensed under CC BY 4.0. http://creativecommons.org/licenses/by/4.0/. Adapted by the author.

Participants performed a series of standardized tasks engaging the core musculature, including relaxed supine breathing, active trunk flexion, straight-leg raise, plank, and side plank (**Figure 12**). For each muscle and task, EMG signals were sampled, band-pass filtered, and rectified. To allow comparisons between subjects, amplitudes were normalized to the maximal voluntary isometric contraction (MVC) recorded during dedicated maneuvers for each muscle group and expressed as percentage of MVC. This normalization approach follows recommendations for trunk EMG studies (137).

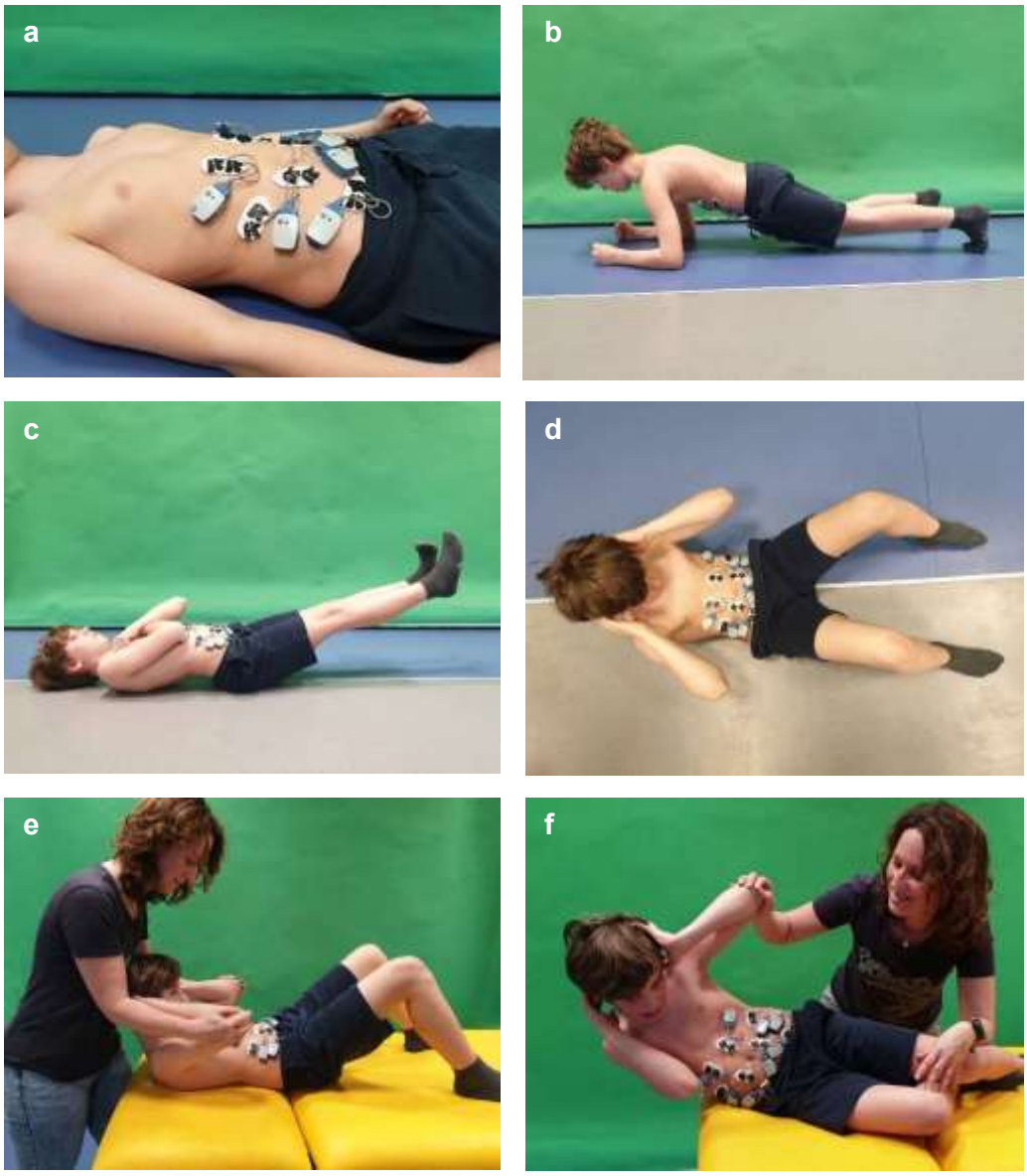

Figure 12: Standardized test series for EMG –(a) relaxed supine breathing (b) plank (c) straight-leg raise and hold (d) active trunk flexion (e) MVC Rectus abdominis (f) MVC oblique muscles (1) Licensed under CC BY 4.0. http://creativecommons.org/licenses/by/4.0/. No changes were made.

2.3.6 Stance and Gait Analysis

For stance and gait analysis, a pressure-sensitive walkway (Zebris FDM system, Velamed GmbH, Cologne, Germany) was used. During ordinary relaxed standing and Matthias' Arm-Raising Test, the trajectory of the center of pressure (COP) was recorded for 30 s, yielding COP path length, sway area, and mean sway velocity as indicators of postural stability. During walking at a self-selected speed for 3 min, spatiotemporal gait parameters (cadence, step length, stride time, stance phase, single-support time) were obtained (**Figure 13**).

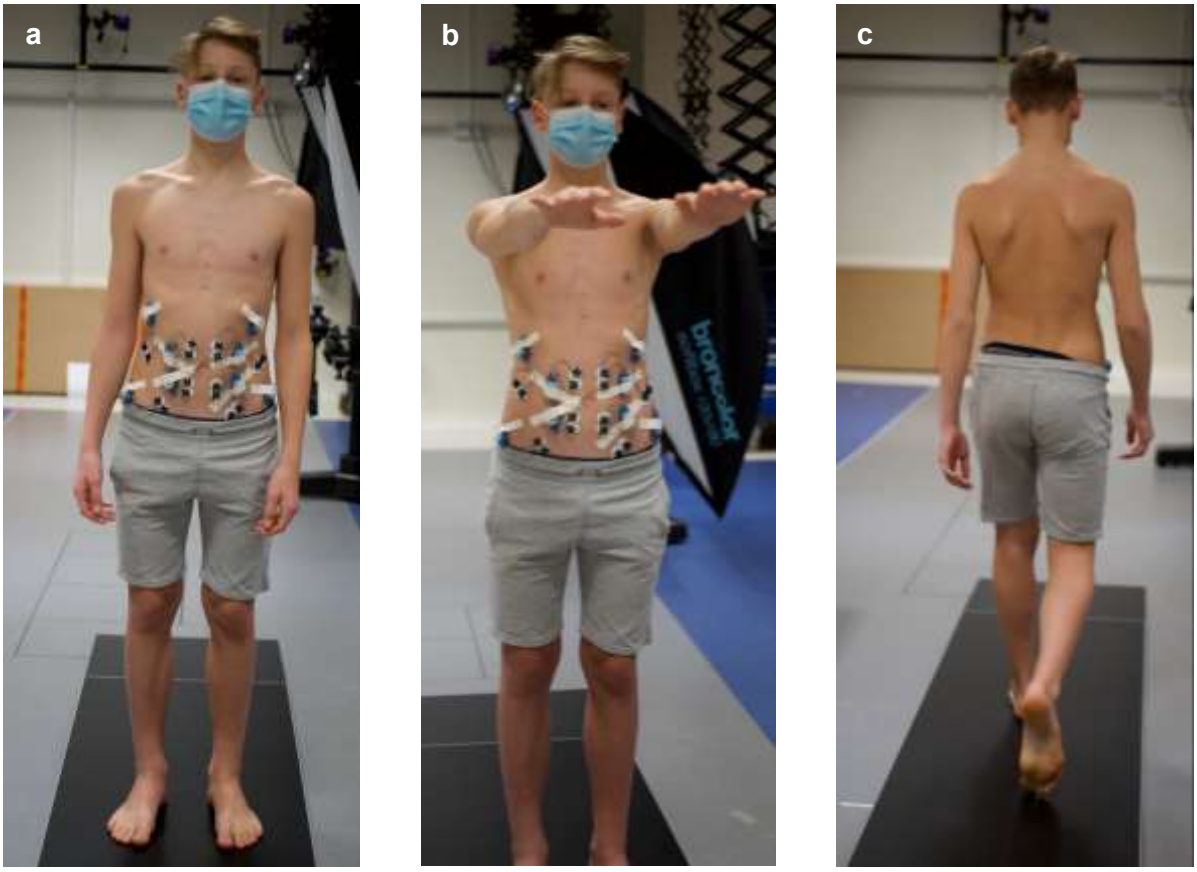

Figure 13: Stance analyses: (a) Ordinary relaxed stand (b) Matthias' Arm-Raising Test (c) Gait analyses: walking over the plate for 3 minutes (1) Licensed under CC BY 4.0. http://creativecommons.org/licenses/by/4.0/. No changes were made.

2.3.7 Ultrasound of the Abdominal Wall

High-resolution B-mode ultrasound (Vivid S5, GE Healthcare, 12L-RS linear probe, Düsseldorf, Germany) was used to measure the thickness of the rectus abdominis, external and internal oblique, and transversus abdominis muscles. A standardized measurement grid was established based on the distance between xiphoid and symphysis. This distance was halved (mark C) and then divided into proximal and distal thirds (marks B1 and B2). Additionally, crossings of the medio-clavicular and anterior axillary lines with transverse lines at B1, B2 an C (I–VI) were marked on the skin, as illustrated in **Figure 14**.

With the patient in the supine position and relaxed, three repeated measurements were taken at each site (I-VI) on both sides of the abdomen. For the rectus abdominis, thickness was determined at its maximal width in the sagittal plane at B1, B2, and C. For the lateral abdominal muscles, thickness of the external oblique, internal oblique, and transversus

abdominis was measured in the sagittal plane at each of the six transverse levels (I-VI). All examinations were performed by the same investigator to minimize inter-observer variability.

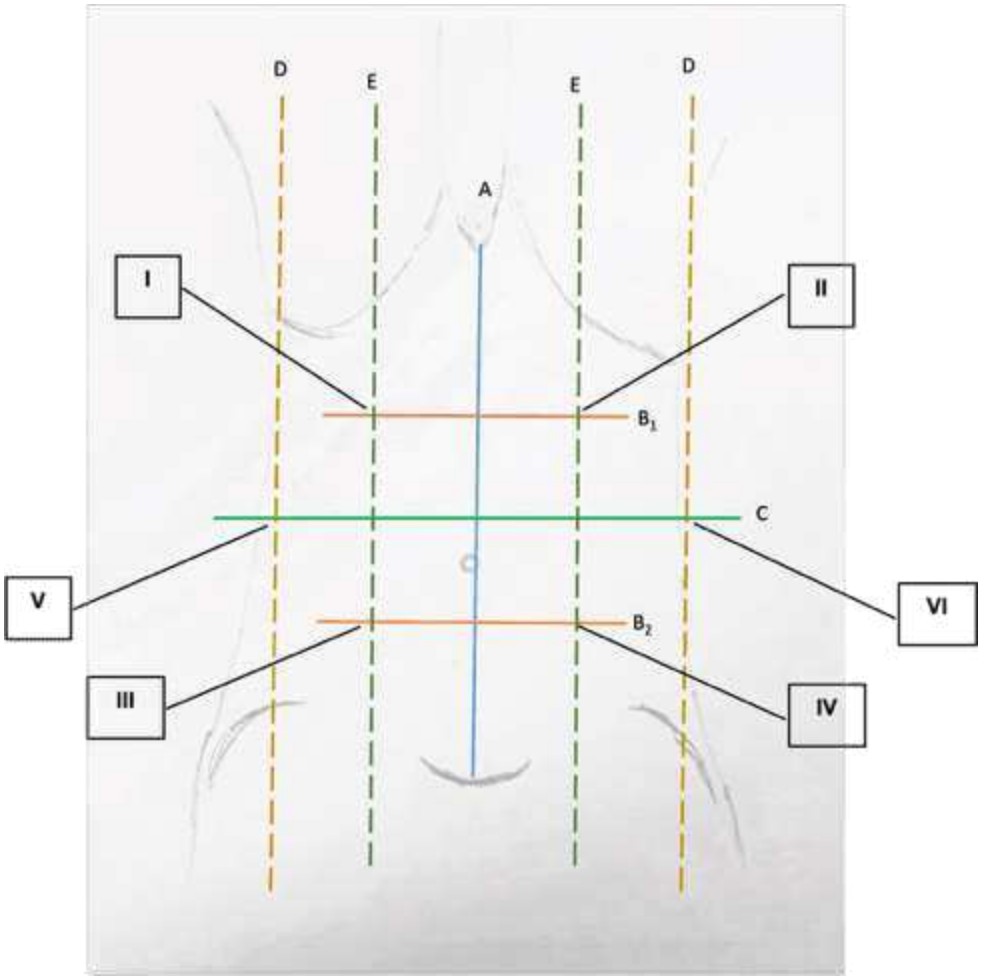

Figure 14: Standardized protocol for ultrasound of the abdominal wall.(1). Licensed under CC BY 4.0. http://creativecommons.org/licenses/by/4.0/. Adapted by the author.

2.3.8 Quality of Life and Cosmetic Satisfaction

Gastrointestinal health-related quality of life was quantified using the GIQLI. The questionnaire contains 36 items covering five domains (core gastrointestinal symptoms, physical, psychological, social, and disease-specific aspects). Each item is scored on a 5-point scale (0–4), resulting in a total score between 0 and 148, with higher scores indicating better quality of life (129). The German version was administered and when necessary, read aloud to younger children (129).

Stool consistency over the preceding 4 weeks was rated by the child or adolescent using the Bristol Stool Form Scale, which classifies stool form into seven categories ranging from hard pellets (type 1) to watery diarrhea (type 7) (138). Stool frequency, episodes of

abdominal pain, symptoms suggestive of bowel obstruction, and presence of gastro-esophageal reflux were assessed using a structured questionnaire. Frequency of back pain was also recorded (never, monthly, weekly, daily).

Cosmetic outcome of the abdominal scar was evaluated using POSAS (139, 140). Both the participant (Patient Scar Assessment Scale, PSAS) and the examiner (Observer Scar Assessment Scale, OSAS) rated scar characteristics (pain, itching, color, stiffness, thickness, irregularity, and surface area) on 10-point scales; the sum score ranges from 6 (best) to 60 (worst). In addition, an overall opinion of the scar appearance (1=very good, 10=very poor) was recorded. The POSAS has been validated for linear surgical scars in children and adults (139, 141).

During clinical examination, the length, and maximal width of the main abdominal scar were measured with a flexible ruler. Presence or absence of an umbilicus, hypertrophic scarring, scar hardening, additional scars, and visible suture marks were documented systematically. A picture of the abdominal wall was taken in each patient.

2.3.9 Statistical Analysis

Data were entered into a dedicated database (Microsoft Excel 2016) and analyzed with SPSS Statistics 27 (IBM Corp., Armonk, NY). Continuous variables were checked for normal distribution using the Kolmogorov–Smirnov test. Normally distributed data are presented as mean ± standard deviation and compared between groups using unpaired two-sided t-tests. Non-normally distributed data are expressed as median and interquartile range (IQR) and analyzed with Mann–Whitney-U tests.

Categorical variables were summarized as counts and percentages and compared with Fisher’s exact test. Correlations between two continuous variables were assessed by Pearson’s correlation coefficient, whereas relationships between ordinal and continuous variables were examined using Spearman’s rank correlation.

All tests were two-sided and a p-value <0.05 was regarded as significant. Given the exploratory nature of this pilot study and the limited sample size inherent to patients with rare congenital anomalies, no adjustment for multiple testing was performed.

3 Results

A total of 55 children with AWDs born between 2002 and 2013 and treated at our center were initially identified. After verification of diagnosis, 35 patients had GS and 20 had OC.

Subsequent stepwise screening reduced the pool of eligible participants. One GS patient had died in the neonatal period and two survivors with GS had severe cognitive impairment that precluded participation in functional testing. Three OC patients were excluded due to hemodynamically relevant congenital heart disease. The most frequent reason for exclusion was inability to contact families because of outdated contact information, affecting nine GS and four OC cases. After all criteria had been applied, 23 GS and 7 OC patients remained reachable and eligible. Of these, 18 patients (12 GS: 8=♀; 4=♂, 6 OC: 3=♀; 3=♂), with a mean age of 12.6 ± 3.5 years (range 7–18 years) agreed to participate and completed all components of the assessment (**Figure 15**).

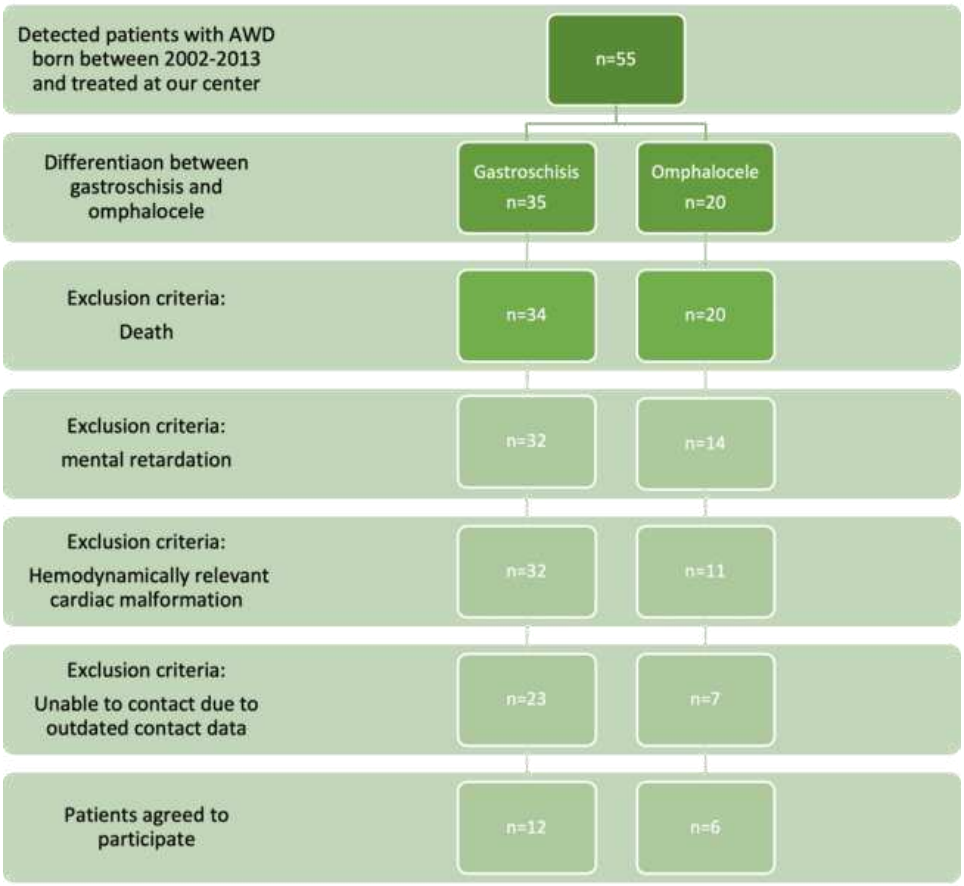

Figure 15: Trial flow diagram from the original publication(1). Licensed under CC BY 4.0. http://creativecommons.org/licenses/by/4.0/. Adapted by the author.

Clinical characteristics, associated malformations, type of neonatal repair and subsequent abdominal operations are summarized in **Table 1**.

Within the GS subgroup, one child (8%) presented with a colonic atresia and was therefore categorized as a complex case, while the remaining eleven children (92%) showed the typical presentation of simple GS without additional intestinal malformations. Among the six participants with OC, two had large defects consistent with giant OC, whereas four exhibited smaller OC. One of the small OC cases had previously required neonatal surgery for an ileal atresia. A detailed list of coexisting malformations is provided in **Table 1** from the original publication.

Regarding initial surgical management, primary abdominal closure during the first day of life was possible in 14 out of 18 participants (77.8%). Two neonates, both affected by intestinal atresia, underwent stoma creation in combination with closure of the abdominal wall. In one infant, the abdominal cavity was initially too small to allow tension-free closure. Therefore, a spring-loaded silo was used until reduction was tolerated. Another child underwent patch-assisted secondary closure after temporary silo placement, reflecting the heterogeneity of early postoperative courses (**Table 1**).

Across the entire cohort, children required an average of 2.2 ± 1.4 abdominal wall–related surgical interventions from birth to the time of follow-up. The highest individual number was five procedures. When comparing defect types, children with OC tended to undergo more surgeries than those with GS, although this difference did not reach statistical significance (OC mean 2.8 ± 2 vs GS mean 1.9 ± 0.9; p=0.334; *unpaired two-sided t-test*).

A total of eight children experienced complications that required operative treatment during the first twelve years of life. Half of these events occurred in infancy, including adhesive bowel obstruction (two cases), midgut volvulus in one child, an umbilical hernia in another and an infected patch that had to be removed at two months of age. Later complications were also documented as two umbilical hernias were repaired during early childhood (ages one, and two years), one child developed volvulus with adhesive obstruction at six years, and another underwent surgery for an adhesive ileus at twelve years. These findings underline the variability in long-term surgical burden associated with AWDs.

The control group included 18 healthy children (♂=7, ♀=11) matched for age, sex, BMI, and activity level. Their mean age was 12.3 ± 3.3 years (range: 7-17 years), which did not differ significantly from the AWD group (p=0.798; *unpaired two-sided t-test*).

Table 1: Clinical data of 18 AWD patients including type of AWD, surgical procedure, associated malformations, complications and total number of operative procedures affecting the abdominal wall(1). Licensed under CC BY 4.0. http://creativecommons.org/licenses/by/4.0/. No changes were made.

ID	Age [years]	Sex	Type of AWD	Classification	Associated Malformations	Procedure	Complications	Total number of procedures
1	7	f	gastroschisis	uncomplicated	-	PC	UH (22 mo)	2
2	7	f	gastroschisis	complicated	colon atresia	PC + colostomy		2
3	8	f	gastroschisis	uncomplicated	-	PC		1
4	10	f	gastroschisis	uncomplicated	-	PC		1
5	11	f	gastroschisis	uncomplicated	-	PC	IDA (1mo)	3
6	15	f	gastroschisis	uncomplicated	-	PC	IDA and MV (6yo)	2
7	15	f	omphalocele	small	-	PC		1
8	16	f	omphalocele	small	-	PC		1
9	16	f	gastroschisis	uncomplicated	-	PC	UH (25 mo)	2
10	18	f	omphalocele	small	ileum atresia, combined immune-deficiency, intractable diarrhea	PC + ileostomy	IDA (12yo)	5
11	18	f	gastroschisis	uncomplicated	-	PC		1
12	8	m	omphalocele	small	-	PC		1
13	12	m	gastroschisis	uncomplicated	-	SC (silo)		2
14	13	m	omphalocele	giant	pulmonary hypo- and dysplasia, ASD, eventration of the right diaphragm	PC		5
15	13	m	gastroschisis	uncomplicated	-	PC		1
16	13	m	gastroschisis	uncomplicated	-	PC	MV/IDA (1wo/3mo)	4
17	13	m	gastroschisis	uncomplicated	-	PC	UH, DR (23mo)	2
18	13	m	omphalocele	giant	pulmonary hypoplasia, ASD	SC (silo+patch)	PI (2mo)	4

ASD – atrial septal defect, PC – primary closure, SC – secondary closure, UH – umbilical hernia, IDA – ileus due to adhesions, MV – midgut volvulus, DR – diastasis recti, PI – patch infection

3.1 Main Analysis

3.1.1 Anthropometry, Body Composition and Laboratory Parameters

Comparison of anthropometric measurements revealed no significant differences between groups. Height, weight, BMI, body fat percentage, and appendicular skeletal muscle mass were comparable between AWD patients, and healthy controls. The distribution of habitual physical activity levels was also not significantly different, indicating successful matching of baseline fitness characteristics (**Table 2**).

Eleven patients agreed to take a blood sample and only in one cases slightly elevated aminotransferases (ALT- alanine aminotransferase 39 U/l [-35 U/l], AST - aspartate aminotransferase=51 U/l [-35 U/l]) were found. This subject had prolonged total parenteral nutrition because of intractable diarrhea.

Table 2: Anthropometric data of AWD patients and age-, sex-, and BMI-matched controls. All data are displayed as mean ± standard deviation unless otherwise specified(1). Licensed under CC BY 4.0. http://creativecommons.org/licenses/by/4.0/. Adapted by the author.

	AWD patients [n=18]	**Controls [n=18]**	**p-value**
Age	12.6 ± 3.5	12.3 ± 3.3	0.798
Anthropometry			
Height [m]	1.59 (0.28)[a]	1.60 (0.24)[a]	0.983**
Body Weight [kg]	48.1 ± 16.7	49.7 ± 16.3	0.785*
BMI [kg/Height²]	19.4 ± 4.4	19.7 ± 4.0	0.787*
Body Fat [%]	13.8 (18.1)[a]	10.8 (20.0)[a]	0.888**
Muscle Mass[b]	6.3 ± 1.3	6.2 ± 1.5	0.943*
Physical Activity Level[c]	2 (1)[a]	2 (0)[a]	0.521**

AWD…abdominal wall defect; m…meter; kg…kilogram;

* unpaired t-test, ** Mann-Whitney-U test,

[a] median (IQR)

[b] n=14; it was not possible to detect muscle mass in 4 cases

[c] Physical Activity Level (once per month=0, once per week=1 several times a week=2, daily=3)

3.1.2 Pulmonary Function

Resting spirometry demonstrated largely preserved pulmonary function across both groups. Neither forced vital capacity (VC_max) nor FEV_1/VC_max ratio differed significantly between AWD patients and controls (**Table 3**). Two individuals in the AWD cohort exhibited a restrictive pattern, both of whom were known to have giant OCs associated with pulmonary hypoplasia.

Table 3: Results of spirometry of AWD patients and age-, sex-, and BMI-matched controls. All data are displayed as mean ± standard deviation(1). Adapted by the author.

	AWD patients [n=18]	**Controls [n=18]**	**p-value**
VC_{max} [L]	2.9 ± 1.0	3.1 ± 1.1	0.558*
Tiffeneau Index [%]	86.4 ± 7.6	86.5 ± 7.6	0.965*

AWD…abdominal wall defect; VC_{max}…maximum vital capacity;

* unpaired t-test

3.1.3 Cardiopulmonary Exercise Performance

Thirteen patients were able to complete CPET and five had to be excluded because of patient height. Across all measured variables, including relative performance capacity, peak oxygen uptake (peak VO_2), oxygen pulse, respiratory exchange ratio (RER), ventilatory equivalent for oxygen (EQO_2), and breathing reserve no significant differences were observed between patients and controls. Mean RER values exceeded 1.10 in both groups, indicating reliable maximal effort (**Table 4**).

Table 4: Results CPET of AWD patients and age-, sex-, and BMI-matched controls. All data are displayed as mean ± standard deviation unless otherwise specified(1). Licensed under CC BY 4.0. http://creativecommons.org/licenses/by/4.0/. Adapted by the author.

	AWD patients [n=13[a]]	**Controls [n=13[a]]**	**p-value**
Relative Performance [%]	97.9 ± 24.3	98.0 ± 20.3	0.986*
RER	1.16 ± 0.08	1.14 ± 0.11	0.600*
peak VO_2 [ml/kg/min]	40.3 (11.0)[b]	39.7 (17.0)[b]	0.555**
O_2/HR [ml]	11.0 (4.0)[b]	11.2 (4.0)[b]	0.751**
EQO_2	21.7 ± 3.1	19.7 ± 2.4	0.071*
BR/FEV_1%	10.5 ± 10.8	14.2 ± 17.7	0.510*

RER…respiratory exchange ratio; peak VO_2…peak oxygen uptake; O_2/HR…oxygen pulse; EQO_2…respiratory equivalent for oxygen, BR…breathing reserve; FEV_1%…Forced Expiratory Volume;

*Unpaired t-test, ** Mann-Whitney-U test,

[a]5 patients were not able to perform CPET because they were too short for ergometry

[b] median (IQR)

3.1.4 Assessment of Motor Abilities

Motor performance differed between the groups. AWD patients displayed significantly higher overall scores in the Dordel–Koch-Test (median 3.4 vs. 2.80; p=0.005; *unpaired two-sided t-test*), representing lower motor function. The most pronounced deficits were observed in the “lateral jumping” and “sit-up” tasks (**Table 5**).

Correlational analyses revealed no association between DKT results and BMI (r=0.213, p=0.213; *Spearman test*), muscle mass (r=0.241, p=0.183; *Spearman test*), body fat percentage or relative performance capacity (r=-0.001, p=0.994; *Spearman test*). However, a significant negative correlation was observed between DKT score and habitual physical activity (r=−0.379; p=0.022; *Spearman test*).

Table 5: Dordel-Koch-Test (DKT) of AWD patients vs. age-, sex-, and BMI-matched controls. All data are displayed as median and interquartile range; statistical comparisons were performed with the Mann-Whitney-U test(1).

	AWD [n=18]	**Controls [n=18]**	**p-value**
Lateral Jumping	4 (1)	3 (2)	**0.037**
Sit and Reach	4 (2)	3 (1)	0.171
Sit-Ups	4 (1)	3 (2)	**0.003**
Long Stand Jump	4.5 (1)	4 (1)	0.068
One-legged Stand	1 (3)	1 (0)	0.252
Push-Ups	3 (1)	2 (1)	0.111
DKT	3.4 (1)	2.8 (1)	**0.005**

3.1.5 Surface Electromyography of the Abdominal Wall

Surface electromyography of the abdominal wall demonstrated no significant intergroup differences in muscle activation across all recorded tasks beside right obliquus internus muscle in exercise “straight leg raise and hold” (**Table 6**). The time participants were able to maintain the “plank” position did not significantly differ between groups (**Table 6**). Likewise, the duration of the “straight leg raise and hold” task showed no significant group difference (**Table 6**). Activation amplitudes of the rectus abdominis, external oblique, internal oblique, and transversus abdominis showed significant difference between left and right side of abdominal wall muscles in the AWD group in exercise “plank” and “straight leg raise and hold” as shown in **Table 7**.

Table 6: Normalized data of surface electromyography of all AWD patients vs. age-, sex-, and BM-matched controls comparing activation of left and right abdominal wall muscles. Data are displayed as median (IQR); statistical comparisons were performed with the Mann-Whitney-U test unless otherwise specified.

	AWD [n=18]		**Control [n=18]**		**p-value**
	Left	Right	Left	Right	Left/Right
Ordinary Relaxed Stand					
Rectus up [%]	2.5 (4.0)	2.5 (4.0)	1.0 (4.0)	2.0 (3.0)	0.164/ 0.772
Rectus low [%]	3.0 (5.0)	3.0 (5.0)	2.5 (4.0)	3.0 (4.0)	0.772/ 0.798
Obliquus internus [%]	8.0 (12.0)	8.0 (12.0)	7.0 (8.0)	8.0 (10.0)	0.281/ 0.851
Obliquus externus [%]	4.0 (8.0)	4.0 (8.0)	5.0 (3.0)	4.0 (5.0)	0.670/ 0.746
Matthias' Arm-Raising Test					
Rectus up [%]	2.0 (3.0)	2.0 (3.0)	1.0 (3.0)	2.0 (3.0)	0.075/ 0.932
Rectus low [%]	2.5 (3.0)	2.5 (3.0)	2.0 (2.0)	2.0 (2.0)	0.297/ 0.224
Obliquus internus [%]	8.0 (15.0)	8.0 (15.0)	8.5 (6.0)	10.0 (12.0)	0.422/ 0.746
Obliquus externus [%]	5.0 (10.0)	5.0 (10.0)	4.0 (4.0)	4.0 (4.0)	0.646/ 0.365
Abdominal Press					
Rectus up [%]	14.5 (27.0)	14.5 (27.0)	6.0 (15.0)	7.0 (13.0)	0.365/ 0.932
Rectus low [%]	13.0 (30.0)	13.0 (30.0)	7.0 (13.0)	9.0 (13.0)	0.422/ 0.798
Obliquus internus [%]	50.0 (83.0)	50.0 (83.0)	29.0 (24.0)	42.0 (41.0)	0.055/ 0.695
Obliquus externus [%]	26.0 (31.0)	26.0 (31.0)	22.0 (16.0)	18.0 (23.0)	0.621/ 0.721
Concentric Phase of Sit-Up					
Rectus up [%]	65.5 (30.0)	65.5 (30.0)	61.5 (41.0)	64.5 (39.0)	0.443/ 0.646
Rectus low [%]	65.5 (53.0)	65.5 (53.0)	72.5 (56.0)	72.5 (55.0)	0.646/ 0.959
Obliquus internus [%]	67.5 (90.0)	67.5 (90.0)	70.0 (65.0)	66.5 (40.0)	0.551/ 0.721
Obliquus externus [%]	50.0 47.0)	50.0 47.0)	54.0 (36.0)	51.5 (65.0)	1.000/ 1.000
Excentric Phase of Sit-Up					
Rectus up [%]	34.5 (21.0)	34.5 (21.0)	28.0 (17.0)	30.5 (19.0)	0.088/ 0.281
Rectus low [%]	43.0 (32.0)	43.0 (32.0)	33.5 (38.0)	39.5 (25.0)	0.224/ 0.443
Obliquus internus [%]	40.0 (66.0)	40.0 (66.0)	29.0 (34.0)	31.0 (24.0)	0.251/ 0.551
Obliquus externus [%]	33.5 (26.0)	33.5 (26.0)	22.5 (19.0)	27.0 (18.0)	0.109/ 0.297
Plank					
Duration of exercise	20.7 ± 14.7[a]		20.8 ± 11.0[a]		0.825[*]
Rectus up [%]	38.5 (80.0)	38.5 (80.0)	25.5 (51.0)	35.0 (36.0)	0.146/ 0.232
Rectus low [%]	58.0 (82.0)	58.0 (82.0)	79.5 (98.0)	43.5 (40.0)	0.602/ 0.415
Obliquus internus [%]	25.5 (53.0)	25.5 (53.0)	52.5 (64.0)	54.0 (30.0)	0.325/ 0.232
Obliquus externus [%]	42.5 (52.0)	42.5 (52.0)	53.0 (59.0)	19.5 (24.0)	0.692/ 0.755

Straight Leg Raise and Hold

Duration of exercise	17.4 ± 11.0[a]		19.5 ± 9.6[a]		0.126[*]
Rectus up [%]	38.5 (80.0)	38.5 (80.0)	58.5 (73.0)	58.0 (66.0)	0.854/ 0.064
Rectus low [%]	61.5 (64.0)	61.5 (64.0)	79.0 (85.0)	60.5 (74.0)	0.728/ 0.093
Obliquus internus [%]	53.0 (60.0)	53.0 (60.0)	62.0 (61.0)	70.0 (61.0)	0.728/ **0.047**
Obliquus externus [%]	38.5 (51.0)	38.5 (51.0)	56.5 (36.0)	10.5 (21.0)	0.294/ 0.759

[*] unpaired t-test

[a] mean ± standard deviation

Table 7: Normalized data of surface electromyography of all AWD patients comparing activation of left and right abdominal wall muscles. All data are displayed as median (IQR); statistical comparisons were performed with the Wilcoxon test.

	Left [n=18]	Right [n=18]	p-value
Ordinary Relaxed Stand			
Rectus up [%]	2.5 (4.0)	1.5 (2.0)	0.081
Rectus low [%]	3.0 (5.0)	3.0 (3.0)	0.452
Obliquus internus [%]	8.0 (12.0)	8.5 (5.0)	0.451
Obliquus externus [%]	4.0 (8.0)	5.0 (1.0)	0.504
Matthias' Arm-Raising Test			
Rectus up [%]	2.0 (3.0)	1.0 (2.0)	0.086
Rectus low [%]	2.5 (3.0)	2.0 (3.0)	0.873
Obliquus internus [%]	8.0 (15.0)	10.0 (7.0)	0.776
Obliquus externus [%]	5.0 (10.0)	5.0 (5.0)	0.305
Abdominal Press			
Rectus up [%]	14.5 (27.0)	10.5 (27.0)	0.102
Rectus low [%]	13.0 (30.0)	11.0 (32.0)	0.379
Obliquus internus [%]	50.0 (83.0)	42.0 (57.0)	0.140
Obliquus externus [%]	26.0 (31.0)	23.0 (25.0)	0.133
Concentric Phase of Sit-Up			
Rectus up [%]	65.5 (30.0)	57.0 (39.0)	0.334
Rectus low [%]	65.5 (53.0)	63.0 (51.0)	0.173
Obliquus internus [%]	67.5 (90.0)	59.0 (64.0)	0.093
Obliquus externus [%]	50.0 47.0)	53.5 (73.0)	0.733
Excentric Phase of Sit-Up			
Rectus up [%]	34.5 (21.0)	33.0 (20.0)	0.495
Rectus low [%]	43.0 (32.0)	38.0 (20.0)	0.608
Obliquus internus [%]	40.0 (66.0)	33.5 (38.0)	0.056
Obliquus externus [%]	33.5 (26.0)	34.5 (28.0)	0.910
Plank			
Rectus up [%]	38.5 (80.0)	24.5 (28.0)	0.071
Rectus low [%]	58.0 (82.0)	25.5 (32.0)	**0.019**
Obliquus internus [%]	25.5 (53.0)	37.5 (35.0)	0.937
Obliquus externus [%]	42.5 (52.0)	21.0 (15.0)	**0.015**
Straight leg Raise and Hold			
Rectus up [%]	45.5 (31.0)	32.0 (26.0)	**0.030**
Rectus low [%]	61.5 (64.0)	44.5 (38.0)	**0.016**
Obliquus internus [%]	53.0 (60.0)	42.0 (43.0)	0.152
Obliquus externus [%]	38.5 (51.0)	11.0 (17.0)	**0.001**

3.1.6 Stance and Gait Analysis

Postural measurements during quiet standing and during the Matthias Arm-Raising Test revealed no significant differences between groups. Centre-of-pressure variables, including sway path length, sway amplitude, mean sway velocity, and postural shift, were similar in AWD patients and controls across all tested conditions and showed no significant difference.

Gait assessment showed that for most spatiotemporal parameters there were no significant differences between AWD patients and healthy controls (**Table 8**). Cadence was slightly higher in the AWD group and stride time was marginally shorter. Both differences reached statistical significance. No significant group differences were observed for step length, walking speed, stride length, stance phase duration, or single-support time. No asymmetries or abnormal gait patterns were recorded in any participant.

Table 8: Stance and gait analyses of AWD patients vs. age-, sex-, and BMI-matched controls; all data are displayed as median and interquartile range; statistical comparisons were performed with the Mann-Whitney-U test (1). Adapted by the author.

	AWD [n=18]	**Controls [n=18]**	**p-value**
Stance Analyses			
Ordinary relaxed stand (ORS)			
Test duration [sec]	30.0 (4.5)	29.4 (1.97)	0.590
COP sway ellipse [cm^2]	2.1 (6.4)	4.6 (6.1)	0.483
total COP path [mm]	302.0 (308.8)	372.0 (386.8)	0.303
COP average speed [mm/s]	9.5 (9.3)	13.0 (13.0)	0.369
Matthias' Arm-Raising Test (MART)			
Test duration [sec]	29.2 (4.7)	28.9 (4.6)	0.732
COP sway ellipse [cm^2]	2.6 (6.0)	5.7 (6.1)	0.232
total COP path [mm]	299.0 (286.0)	369.0 (386.3)	0.184
COP average speed [mm/s]	10.0 (10.0)	13.5 (12.8)	0.153
Change in COP ORS / MART [cm^2]	27.0 (239.0)	-81.5 (227.0)	0.163
Gait Analysis			
Stance time [%]	63.1 (2.8)	63.2 (1.6)	0.481
Single support [%]	37.0 (2.8)	36.8 (1.6)	0.481
Gait line [mm]	209.5 (42.0)	208.5 (27.9)	0.462
Cadence [steps/min]	112.0 (19.0)	104.0 (11.8)	**0.031**
Gait speed [cm/s]	3.7 (0.7)	3. 5 (0.7)	0.389
Step length [cm]	55.0 (11.3)	55.5 (9.5)	0.770
Step time [ms]	535.5 (75.0)	592.5 (75.9)	0.086
Stride length [cm]	112.0 (15.0)	114.5 (28.0)	0.782
Stride time [ms]	1073.0 (151.0)	1154.0 (136.0)	**0.031**

COP…center of pressure

3.1.7 Ultrasound of the Abdominal Wall

Ultrasound measurements of abdominal wall morphology revealed no significant differences between AWD and control patients (**Table 9**). No systematic variation in muscle thickness was detected in relation to defect type or previous surgical interventions. No significant difference was found between the left and right side of the abdomen in muscle thickness (**Table 10**).

Table 9: Ultrasound of the abdominal wall of AWD patients vs. age-, sex-, and BMI-matched controls. All data are displayed as median and interquartile range; statistical comparisons were performed with the Mann-Whitney-U test and data displayed as median and (IQR) (1).

	AWD [n=18]	**Controls [n=18]**	**p-value**
I OE	3.4 (4.0)	2.5 (0.9)	0.135
II OE	2.9 (1.7)	2.2 (1.5)	0.211
III OE	3.8 (2.3)	3.1 (1.8)	0.203
IV OE	4.2 (2.5)	3.2 (1.8)	0.192
V OE	5.0 (3.9)	6.2 (2.0)	0.239
VI OE	5.5 (3.7)	6.5 (2.2)	0.696
I OI	3.1 (1.7)	3.8 (1.5)	0.057
II OI	3.5 (2.0)	3.2 (1.8)	0.279
III OI	5.1 (2.4)	4.7 (2.1)	0.839
IV OI	4.6 (2.9)	4.8 (2.6)	0.443
V OI	6.3 (3.5)	6.7 (3.0)	0.372
VI OI	7.1 (2.9)	6.1 (3.0)	0.521
I TA	2.5 (1.2)	2.6 (0.9)	0.287
II TA	2.6 (1.4)	2.7 (1.2)	0.660
III TA	2.2 (2.1)	2.6 (1.6)	0.152
IV TA	2.4 (1.6)	3.0 (1.1)	0.171
V TA	3.4 (2.2)	3.1 (1.6)	0.888
VI TA	3.3 (1.9)	3.4 (1.8)	0.963
B_1 RA right	8.0 (4.8)	8.3 (3.2)	0.782
B_1 RA left	8.7 (4.1)	7.7 (5.1)	0.546
B_2 RA right	8.6 (3.6)	8.6 (4.1)	0.839
B_2 RA left	9.6 (4.3)	8.8 (3.9)	0.462
C RA right	8.7 (3.6)	8.5 (4.7)	0.963
C RA left	8.9 (3.5)	8.6 (4.6)	0.815

OE…m. obliquus externus; OI…m. obliquus internus; TA…m. transversus abdominis; RA… m. rectus abdominis

Table 10: Ultrasound data of all AWD patients comparing thickness of left and right abdominal wall muscles. All data are displayed as mean ± standard deviation unless otherwise specified; statistical comparisons were performed with the Wilcoxon test(1). Licensed under CC BY 4.0. http://creativecommons.org/licenses/by/4.0/. No changes were made.

	Right [n=18]	Left [n=18]	p-value
I OE / II OE	2.7 (1.9)[c]	2.6 (1.5)[c]	0.270[b]
III OE / IV OE	3.5 (1.9)[c]	3.4 (2.1)[c]	0.654[b]
V OE / VI OE	5.8 (2.5)[c]	5.9 (3.3)[c]	0.561[b]
I OI / II OI	3.3 (2.0)[c]	3.2 (2.0)[c]	0.957[b]
III OI / IV OI	5.0 (2.0)[c]	4.7 (2.5)[c]	0.667[b]
V OI / VI OI	6.6 ± 1.7	6.6 ± 1.7	0.900[a]
I TA / II TA	2.8 ± 0.9	2.7 ± 0.8	0.578[a]
III TA / IV TA	2.5 (2.0)[c]	2.9 (1.5)[c]	0.471[b]
V TA / VI TA	3.5 ± 1.2	3.5 ± 1.0	0.163[a]
B_1 RA right / left	8.3 ± 2.4	8.5 ± 2.7	0.282[a]
B_2 RA right / left	8.8 ± 2.5	9.0 ± 2.5	0.278[a]
C RA right / left	9.0 ± 2.7	8.9 ± 2.6	0.339[a]

OE…m. obliquus externus; OI…m. obliquus internus; TA…m. transversus abdominis; RA… m. rectus abdominis

[a] paired t-test; [b] Wilcoxon test; [c] median (IQR)

3.1.8 Quality of Life and Cosmetic Satisfaction

Gastrointestinal quality of life, assessed using the GIQLI questionnaire, differed significantly between groups. Children with AWDs reached a mean GIQLI score of 137.2 ± 6.8, whereas healthy controls achieved 141.4 ± 4.9 (p=0.038, *unpaired two-sided t-test*). When comparing the two defect types, GS (137.5 ± 7.7) and OC (136.5 ± 5.2) showed no significant difference (p=0.749, *unpaired two-sided t-test*). The total number of abdominal surgeries did not correlate with GIQLI outcomes (r=0.154, p=0.542, *Spearman test*). Analysis of the individual GIQLI items revealed that AWD patients most frequently reported occasional abdominal discomfort and episodes of bloating.

Further questionnaire data indicated that gastro-esophageal reflux symptoms were present in a minority of AWD participants. Symptoms were recorded in two AWD patients (one monthly, one daily) and in one control (monthly). Statistical comparison revealed no significant group differences (p=0.500, F*isher exact test*). Six patients in the AWD group reported back pain (five monthly, one weekly) and in the control cohort, four children described similar symptoms (two monthly, two weekly). This distribution did not differ significantly between groups (p=0.354, *Fisher exact test*).

Stool habits were comparable between groups and show. In the AWD cohort, 16 of 17 respondents (94%) reported a normal stool frequency (“every other day to twice daily”), while one child (6%) indicated more frequent bowel movements. In the control group, 13 children (76%) described normal frequencies and four (24%) reported more frequent stools.

According to the Bristol Stool scale stool consistency were described by AWD as type 2 (n=1), type 3 (n=5), type 4 (n=11), and type 6 (n=1) and in contrast the control group described type 3 (n=12) and type 4 (n=6).

Assessment of scar characteristics showed a heterogeneous pattern. In nine patients (6 GS, 3 OC), the umbilicus represented the only visible scar following neonatal closure. Among the remaining participants, seven children (4 GS, 3 OC) had a horizontal abdominal scar, while one patient presented with a vertical midline scar. Six children (3 GS, 3 OC) had one or more additional abdominal scars and in twelve cases (10 GS, 2 OC) remnants of skin sutures were visible as fine linear marks.

The patient-rated overall impression averaged 3.9 ± 2.8 and the observer rating reached 2.7 ± 1.4 (lower values indicating better appearance). The median POSAS values were 16.7 ± 8.6 for the Patient Scale (PSAS) and 13.5 ± 5.7 for the Observer Scale (OSAS).

Correlation analysis demonstrated a significant positive association between OSAS scores and scar length (r=0.685, p=0.002, *Spearman test*). No associations were found between PSAS or OSAS scores and the number of surgeries, defect type or scar morphology beyond length. Data are shown in **Table 11**.

Table 11: Cosmetic satisfaction; all data are displayed as median and interquartile range unless otherwise specified(1). Licensed under CC BY 4.0. http://creativecommons.org/licenses/by/4.0/. Adapted by the author.

AWD [n=18]	PSAS	OSAS	p-value (PSAS / OSAS)
Cosmetic Satisfaction			
Umbilicus (yes=15, no=3)	15 (19), 14 (0)	12 (6), 21 (0)	0.824* / 0.076*
Hardening of the scar (yes=5, no=13)	18 (12), 14 (16)	12 (15), 13 (6)	0.503* / 0.775*
Additional scars (yes=6, no=12)	14.5 (3), 15.5 (23)	18 (9), 11 (4)	1.000* / **0.003***
Visible stiches (yes=12, no=6)	15.5 (14), 10 (16)	13.5 (7), 10.5 (11)	0.250*/ 0.250*
Correlation Coefficient			
Length of the scar (mm)	0.137	0.685	0.589† / **0.002**†
Width of the scar (mm)	-0.045	-0.123	0.861† / 0.626†
BMI (kg/m^2)	0.290	-0.016	0.243† / 0.951†
Number of surgeries	0.217	0.568	0.387† / 0.014†

* Mann-Whitney-U; † Spearman test

3.2 Subgroup Analysis

For subgroup analyses we separated AWD population in GS and OC, and for further analyses we compared uncomplicated and complicated GS as well as simple, and giant OC.

Comparison between defect types showed that scar width differed significantly, with GS patients having broader scars than those with OC (21.8 ± 9.2 mm vs. 11.5 ± 8.2 mm; p=0.034, *unpaired two-sided t-test*). In contrast, neither scar length nor subjective cosmetic ratings (PSAS, OSAS) differed between the two groups. Across all other comparisons, GS vs. OC, as well as uncomplicated vs. complicated GS and small vs. giant OCs, likewise showed no significant differences in any recorded parameters. Data are shown in **Table 12 - 14**.

Table 12: Anthropometric data, results of spirometry, CPET, DKT, and QoL of patients with GS and OC. All data are displayed as mean ± standard deviation unless otherwise specified (1). Licensed under CC BY 4.0. http://creativecommons.org/licenses/by/4.0/. No changes were made.

	Gastroschisis	**Omphalocele**	**p-value**
	[n=12]	**[n=6]**	
Age	11.9 ± 3.5	13.8 ± 3.4	0.287[a]
Anthropometry			
Height [m]	1.57 (0.28)[c]	1.69 (0.17)[c]	0.151[b]
Body Weight [kg]	45.7 ± 16.3	53.0 ± 18.1	0.403[a]
BMI [kg/Height²]	19.3 ± 4.7	19.5 ± 4.1	0.954[a]
Body Fat [%]	7.75 (19.0)[c]	17.5 (18.1)[c]	0.820[b]
Muscle Mass[d]	6.3 ± 1.3	6.2 ± 1.4	0.874[a]
Number of surgeries	6.3 ± 1.3	6.2 ± 1.4	0.588[a]
Physical Activity Level[f]	2 (0)[c]	2 (1)[c]	0.616[b]
Spirometry			
VC_{max} [%]	2.8 ± 1	3.0 ± 0.9	0.765[a]
Tiffeneau Index [%]	86.6 ± 7.6	85.9 ± 3,9	0.781[a]
CPET	**[n=8[e]]**	**[n=5[e]]**	
Relative Performance [%]	93.6 ± 14.9	104.6 ± 36.0	0.546[a]
RER	1.14 ± 0.08	1.19 ± 0.07	0.261[a]
peak VO_2 [ml/kg/min]	42.1 (11)[c]	38.4 (17)[c]	0.717[b]
O_2/HR [ml]	10.6 (5)[c]	12.7 (4)[c]	0.796[b]
EQO_2	22.3 ± 3.0	20.9 ± 3.3	0.450[a]
BR/FEV%	11.9 ± 10.2	8.4 ± 12.6	0.619[a]
DKT			
Mean DKT	3.6 (1.0)[c]	3.3 (1.0)[c]	0.820[b]
Lateral Jumping	4.0 (1.0)[c]	4.0 (2.0)[c]	1.000[b]
Sit and Reach	3.5 (2.0)[c]	4.5 (3.0)[c]	0.494[b]
Sit-Ups	4.0 (1.0)[c]	3.5 (2.0)[c]	0.291[b]
Long Stand Jump	5.0 (2.0)[c]	4.0 (1.0)[c]	0.385[b]
One-legged Stand	1.0 (3.0)[c]	2.0 (4.0) [c]	0.494[b]
Push-Ups	3.0 (1.0)[c]	2.0 (2.0)[c]	0.494[b]
QoL			
GIQLI	137.5 ± 7.7	136.5 ± 5.2	0.749[a]
Bristol Stool scale	3.5 ± 1.0[c]	4.0 ± 1.0[c]	0.053[a]
Cosmetic satisfaction	26.8 ± 5.8	28.0 ± 4.42	0.620[a]

Cosmetic Satisfaction			
PSAS	17.8 ± 8.7	14.7 ± 8.9	0.493
OSAS	12.9 ± 3.2	14.7 ± 9.4	0.673
Length of the scar [mm]	37.5(78)[c]	25.0(112)[c]	0.335[b]
Width of the scar [mm]	21.8 ± 9.2	11.5 ± 8.2	**0.034**

m…meter; kg…kilogram; VC_{max}…maximum vital capacity; RER…Respiratory Exchange Ratio; peak VO_2…peak oxygen uptake; O_2/HR…oxygen pulse; EQO_2…respiratory equivalent for oxygen, BR…breathing reserve, FEV…Forced expiratory volume; DKT…Dordel-Koch-Test; QoL…quality of life; GIQLI…Gastrointestinal Quality of Life Index

[a] unpaired t-test, [b] Mann-Whitney-U test, [c] median (IQR)

[d] n=14; it was not possible to detect muscle mass in 4 cases

[e] 5 patients were not able to perform CPET because they were too short for ergometry

[f] Physical Activity Level (once per month=0, once per week=1 several times a week=2, daily=3)

Table 13: Stance and gait analyses of GS vs. OC; all data are displayed as median and interquartile range; statistical comparisons were performed with the Mann-Whitney-U test.

	Gastroschisis [n=12]	Omphalocele [n=6]	p-value
Stance Analyses			
Ordinary relaxed stand (ORS)			
Test duration [sec]	30.1 (2.5)	28.7 (15.4)	0.591
COP sway ellipse [cm^2]	1.7 (2.2)	4.7 (13.6)	0.591
total COP path [mm]	269.0 (313.0)	286.5 (458.5)	0.884
COP average speed [mm/s]	9.0 (10.0)	9.5 (9.0)	0.733
Matthias' Arm-Raising Test (MART)			
Test duration [sec]	29.3 (7.1)	28.2 (3.8)	0.591
COP sway ellipse [cm^2]	2.1 (2.2)	6.4 (6.1)	0.122
total COP path [mm]	299.0 (317.0)	347.0 (270.3)	0.808
COP average speed [mm/s]	10.0 (12.0)	12.0 (7.8)	1.000
Change in COP ORS / MART [cm^2]	0.27 (2.1)	0.52 (7.7)	0.733
Gait Analysis			
Stance time [%]	62.2 (2.6)	63.8 (2.0)	0.067
Single support [%]	37.8 (2.6)	36.2 (2.0)	0.067
Gait line [mm]	209.5 (43.3)	196.8 (40.1)	0.750
Cadence [steps/min]	113.0 (14.5)	107.5 (26.0)	0.335
Gait speed [cm/s]	3.7 (0.9)	3. 8 (1.0)	0.494
Step length [cm]	54.5 (10.9)	56.5 (7.8)	0.679
Step time [ms]	530.8 (69.6)	570.0 (154.8)	0.371
Stride length [cm]	112.5 (29.0)	114.5 (18.0)	0.682
Stride time [ms]	1062.0 (119.0)	1118.0 (275.0)	0.291

COP…center of pressure

Table 14: Ultrasound data of GS and OC patients. All data are displayed as mean ± standard deviation unless otherwise specified.

	Gastroschisis [n=12]	Omphalocele [n=6]	p-value
I OE	3.9 ± 1.7	3.6 ± 2.3	0.806[a]
II OE	3.1 ± 1.6	3.6 ± 1.6	0.571[a]
III OE	3.8 (2.2)[c]	4.1 (3.5)[c]	0.892[b]
IV OE	4.0 ± 1.6	4.4 ± 1.6	0.639[a]
V OE	6.1 ± 2.4	5.5 ± 2.4	0.630[a]
VI OE	6.0 ± 2.1	5.8 ± 2.4	0.845[a]
I OI	3.1 ± 1.1	3.0 ± 1.1	0.987[a]
II OI	3.5 ± 1.0	4.3 ± 1.8	0.327[a]
III OI	4.7 ± 1.5	6.2 ± 2.2	0.151[a]
IV OI	4.3 ± 1.5	6.4 ± 2.1	0.061[a]
V OI	6.3 ± 1.7	6.4 ± 2.1	0.929[a]
VI OI	6.8 ± 2.0	6.8 ± 2.0	0.973[a]
I TA	2.9 ± 0.8	2.3 ± 0.9	0.146[a]
II TA	3.0 (1.5)[c]	2.6 (0.6)[c]	0.961[b]
III TA	2.0 (2.3)[c]	2.4 (2.5)[c]	0.750[b]
IV TA	2.3 (1.4)[c]	3.4 (2.2)[c]	0.180[b]
V TA	3.6 ± 1.8	3.6 ± 1.5	0.983[a]
VI TA	3.7 ± 1.1	3.0 ± 1.1	0.226[a]
B_1 RA right	8.6 ± 2.7	7.8 ± 3.6	0.637[a]
B_1 RA left	8.8 ± 2.7	8.8 ± 4.1	0.942[a]
B_2 RA right	9.0 ± 2.7	8.8 ± 3.1	0.884[a]
B_2 RA left	9.3 ± 2.8	9.5 ± 3.2	0.892[a]
C RA right	8.8 ± 2.7	9.3 ± 3.4	0.778[a]
C RA left	8.7 ± 2.6	8.9 ± 3.5	0.901[a]

[a] unpaired t-test; [b] Mann-Whitney-U test; [c] median (IQR)

4 Discussion

4.1 Answer to Research Questions and Summary of Results

The present study provides a broad overview of long-term functional outcomes in patients with congenital AWDs. In summary, the results reveal a heterogeneous pattern: while several objective measures were comparable to those of healthy controls, other domains showed significant differences. Motor abilities were the most affected area, with AWD patients performing less well in tasks requiring coordination and trunk involvement. Gait parameters and postural stability, in contrast, demonstrated only minor deviations.

Cardiopulmonary exercise testing yielded normal peak performance in almost all patients and no significant reduction compared with matched controls was identified. Resting pulmonary function was also largely preserved, with restrictive changes confined to isolated cases with a history of giant OC. Assessment of abdominal wall function using surface EMG and ultrasound did not show relevant group differences in activation patterns or muscle thickness.

Patient-reported outcomes indicated slightly lower gastrointestinal quality-of-life scores in the AWD cohort, although most specific gastrointestinal symptoms occurred at similar frequencies as in the control group. Cosmetic results were generally rated favorably by both patients and examiners, despite some variation in scar characteristics. A proportion of patients experienced abdominal wall–related complications or required re-operations during childhood, consistent with published long-term follow-up data (142, 143).

Overall, the findings demonstrate that long-term survivors of GS or OC show preserved cardiopulmonary and abdominal wall function, while mild motor impairments and subtle gastrointestinal limitations remain detectable in a part of the cohort.

Subgroup Analysis

Beside a significant difference in width of the scar, we could not find any significant differences in our outcome parameters comparing OC and GS, uncomplicated and complicated GS or small and giant OCs.

4.2 Anthropometry, Body Composition and Laboratory Parameters

In the present cohort, anthropometric parameters and body composition did not differ between patients with AWDs and matched healthy controls. Height, body weight, BMI, body fat percentage, and appendicular skeletal muscle mass were comparable between groups, indicating a valuable matching of the control group.

While infants with GS or OC are frequently born preterm and with reduced birth weight, longitudinal data suggest that most children achieve normal growth trajectories during childhood and adolescence. Persistent growth impairment has mainly been described in patients with complex GS or giant OC, short bowel syndrome or prolonged dependence on parenteral nutrition, conditions that were rare in the present cohort (61, 144-146).

Laboratory findings were largely unremarkable. Mildly elevated liver enzymes were observed in only one patient with a history of prolonged parenteral nutrition due to severe gastrointestinal morbidity. This isolated finding aligns with existing evidence that hepatobiliary complications are predominantly restricted to patients with extended neonatal intestinal failure and are uncommon in long-term survivors without ongoing nutritional dependency (147).

4.3 Pulmonary Function

In the present cohort, long-term pulmonary function was largely preserved in patients with AWDs when compared with matched healthy controls. Spirometry parameters, including VC_max, and FEV_1, were predominantly within age- and sex-adjusted reference ranges and no clinically relevant differences were observed between groups. These findings indicate that, despite early-life respiratory challenges, most patients achieve adequate pulmonary development into childhood and adolescence.

Two patients in our cohort showed restrictive ventilation disorders. Both suffered from giant OC and born with pulmonary hypoplasia. Reduced lung volumes and restrictive ventilatory patterns have been described in neonates and infants with AWDs, particularly in patients with giant OC due to associated pulmonary hypoplasia (113, 148). These results demonstrate the necessity of long-term follow-up including pulmonary function testing in patient with AWDs, especially if they were born with giant OC.

4.4 Cardiopulmonary Exercise Performance

Cardiopulmonary exercise performance is widely recognized as an important determinant of long-term health, functional capacity, and overall well-being in children and adolescents (149). Multiple systematic reviews confirm favorable associations between cardiorespiratory fitness and indicators of anthropometry, adiposity, cardiometabolic health, vascular health, and mental well-being (150, 151). In other conditions requiring major neonatal surgery, such as esophageal atresia or anorectal malformations, reduced exercise capacity has been reported, suggesting that early surgical interventions may be associated with persistent functional limitations despite successful anatomical correction (124, 125).

In comparison, data on cardiopulmonary exercise performance in patients with AWDs remain scarce. To date, only one study has specifically addressed this aspect. Zaccara et. al investigated a small cohort of patients with large AWDs who underwent treadmill-based exercise testing until exhaustion. They reported shorter exercise duration and lower maximal oxygen uptake compared with reference values from healthy children. However, the authors suggested that reduced physical fitness rather than disease-specific limitations might have contributed to these findings and emphasized the need for further studies (126).

The present study adds to the existing literature by providing a comprehensive evaluation of cardiopulmonary exercise performance using standardized cycle ergometry combined with breath-by-breath gas analysis. In contrast to the results reported by Zaccara et al., cardiopulmonary performance in the present cohort was largely preserved. None of the assessed parameters differed significantly from those observed in matched healthy control subjects.

Several factors may help explain these differing observations. Methodological differences between treadmill- and cycle-based exercise testing are known to influence performance outcomes, particularly in pediatric populations where familiarity with the testing modality plays an important role. In addition, the inclusion of a control group matched not only for age and sex but also for habitual physical activity reduces the potential confounding effect of general fitness levels. This methodological approach supports the interpretation that preserved exercise capacity in the present cohort reflects intact cardiopulmonary function rather than compensatory mechanisms.

Ventilatory responses during exercise further support this interpretation. Breathing reserve remained preserved at peak workload and ventilatory efficiency, as assessed by the ventilatory equivalent for oxygen, did not differ between groups. These findings indicate that exercise performance was not limited by ventilatory constraints and that pulmonary adaptation following neonatal abdominal wall repair is sufficient to meet increased physiological demands during exercising.

The present findings suggest that long-term survivors of GS or OC do not exhibit clinically relevant impairments in cardiopulmonary exercise performance. Nevertheless, given the heterogeneity of neonatal courses and defect severity, cardiopulmonary exercise testing remains a valuable tool in selected patients and might help identify individuals who could benefit from targeted interventions or structured physical activity programs.

4.5 Assessment of Motor Abilities

Motor performance represents an important component of functional outcome and everyday participation in childhood and adolescence (152). Standardized assessment of motor abilities therefore provides valuable information beyond organ-specific or anatomical outcome measures, particularly in patients with congenital conditions requiring major neonatal surgery.

In the present study, motor abilities were evaluated using the DKT, a well-established test battery for the assessment of basic motor skills in children and adolescents (128). The DKT has previously been applied in cohorts with congenital anomalies, including patients with anorectal malformations, in whom reduced locomotor performance compared with healthy peers has been reported (125).

Patients with AWDs achieved higher DKT scores than control subjects, reflecting reduced motor proficiency.. In particular, the “sit-up” and “lateral jumping” exercises showed significantly lower performance in the patient group. Both tasks impose substantial demands on dynamic trunk stabilization and coordinated abdominal muscle activation, suggesting that these functional domains may be particularly susceptible to long-term impairment in patients born with an AWDs.

While surface electromyography and ultrasound assessments did not reveal generalized abnormalities in abdominal wall muscle activation or thickness, the motor tasks included in the DKT rely on complex, dynamic interactions between multiple muscle groups and sensory feedback mechanisms. Standardized motor testing may uncover functional limitations that are not detectable through isolated or static assessments of muscle morphology or activation.

Importantly, reduced motor performance was observed despite preserved anthropometric parameters and intact cardiopulmonary capacity. This means that impaired motor abilities cannot be explained by reduced muscle mass, growth retardation or limited aerobic fitness alone. Instead, functional factors such as altered motor coordination, reduced trunk control or subtle deficits in neuromuscular integration might contribute to the observed findings.

In addition, secondary influences should be considered. Children born with AWD often experience prolonged neonatal hospitalization, delayed mobilization, recurrent abdominal symptoms, or multiple early surgeries , all factors known to influence neurodevelopmental and

motor development (153). Comparable findings have been described in other congenital conditions involving early intensive care, such as anorectal malformations, where motor deficits similarly persist (125, 154). The observed negative correlation between physical activity level and DKT score in the present cohort further supports the assumption that reduced habitual activity, rather than anatomical impairment alone, may play a role in diminished motor proficiency.

From a clinical perspective, reduced motor abilities may have relevant implications for participation in physical activity, engagement in sports, and long-term musculoskeletal health. Children with lower motor proficiency may be less motivated to participate in regular physical activity, potentially reinforcing functional limitations over time (155, 156). Although the observed impairments were generally mild, they were consistent and measurable, underscoring the importance of structured functional evaluation.

Patients with AWDs might benefit from long-term follow-up strategies that include assessment of motor development. Early identification of reduced motor performance could allow timely referral to targeted physiotherapy or structured exercise programs focusing on coordination, trunk stability, and functional strength.

4.6 Surface Electromyography of the Abdominal Wall

Surface electromyography provides insight into neuromuscular activation patterns of the abdominal wall and complements structural and functional assessments of trunk performance. Until now, it has not been used to evaluate patients born with AWD but has been applied in a broad range of settings, including neuromuscular, urodynamic, and laryngeal disorders before (157-159). In patients with congenital AWDs, concerns have been raised that early disruption of abdominal wall integrity, surgical reconstruction, and scar formation could lead to long-term alterations in muscle activation or coordination (99, 160).

In the present study, surface electromyography of the abdominal wall demonstrated largely comparable muscle activation patterns between patients with AWDs and healthy controls across a majority of tested exercises. No significant intergroup differences were observed for most muscles and tasks, indicating preserved global activation capacity of the rectus abdominis, external oblique, internal oblique, and transversus abdominis muscles during standardized core exercises. An exception was identified for the right internal oblique muscle during the “straight leg raise and hold” exercise, where a significant difference between AWD and control group was detected. This isolated finding did not extend to other muscles or tasks and was not associated with reduced task endurance, as the duration of both the “plank” and “straight leg raise and hold” exercises was comparable between patients and controls.

Subgroup analysis revealed significant side-to-side differences in muscle activation amplitudes in patients with AWDs during the “plank” and “straight leg raise and hold” exercises. These asymmetries were observed across multiple abdominal wall muscles and were less present in the control group. Previous studies have shown that symmetrical exercise tasks do not typically result in side-dependent activation patterns in healthy individuals (161, 162). In this context, the observed asymmetries may be indicative of altered neuromuscular coordination or compensatory activation strategies rather than generalized muscle weakness.

However, identification of side-to-side activation differences could be clinically relevant in patients presenting with reduced motor performance or musculoskeletal complaints and may help guide individualized physiotherapeutic interventions aimed at improving symmetry and coordinated trunk activation.

4.7 Stance and Gait Analysis

Assessment of stance and gait parameters provides valuable information on functional mobility and postural control and has been used for children with cerebral palsy before (163, 164). In patients with congenital AWDs, altered trunk mechanics or subtle impairments in core stability could theoretically influence balance and gait patterns in the long term.

In the present cohort, however, stance and gait parameters in patients with AWDs were largely comparable to those observed in matched healthy control subjects. These findings indicate that basic locomotor function is generally preserved and support the notion that potential functional consequences of AWDs are subtle and may become apparent primarily during tasks placing higher demands on coordination or trunk control.

4.8 Ultrasound of the Abdominal Wall

Ultrasonography represents a non-invasive and widely available method for assessing the morphology of the abdominal wall and its individual muscle layers (165-167). In patients with congenital AWDs, ultrasound allows evaluation of potential long-term structural alterations following neonatal surgical repair, including differences in muscle thickness, symmetry, and distribution.

In the present study, ultrasonographic assessment of the abdominal wall did not reveal relevant differences in muscle thickness between patients with AWDs and healthy controls. Measurements of the rectus abdominis as well as the lateral abdominal wall muscles, including the external oblique, internal oblique, and transversus abdominis, were comparable across groups at all predefined measurement points. These findings suggest preserved structural development of the abdominal wall musculature into childhood and adolescence.

The ultrasonographic findings align with the surface electromyography results of this study, which demonstrated preserved global activation of the abdominal wall muscles. Together, these complementary modalities suggest that neither muscle morphology nor basic voluntary activation capacity is substantially compromised in long-term survivors of AWDs.

Nevertheless, structural integrity assessed by ultrasound does not necessarily equate to full functional performance (168). While muscle thickness was comparable between groups, functional assessments revealed significant impairments in motor abilities. This discrepancy underscores the multifactorial nature of trunk function, which relies not only on muscle morphology but also on neuromuscular coordination, timing of muscle activation, and integration with postural control mechanisms.

From a clinical perspective routine long-term follow-up of asymptomatic survivors of AWD does not necessitate repeated imaging of the abdominal wall. Instead, ultrasonography might be informative in selected patients presenting with clinical signs suggestive of abdominal wall weakness, asymmetry, hernia development, or otherwise unexplained pain.

4.9 Quality of Life and Cosmetic Satisfaction

Beyond objective health measures, quality of life captures how individuals experience health, well-being, and participation in everyday life (169). Therefore, the assessment of QoL represents an essential component of long-term outcome evaluation in patients with congenital AWDs, as survival rates have improved substantially and functional as well as psychosocial aspects have gained increasing importance. In addition to physical health, gastrointestinal well-being, and cosmetic outcome might influence daily functioning, self-perception, and overall life satisfaction.

Previous investigations have evaluated QoL in patients with AWDs using generic pediatric instruments such as the Pediatric Quality of Life Inventory (PedQoL), with some studies reporting differences particularly in adolescent and young adult populations (74, 92, 170). Chronic abdominal symptoms have been described in a subset of these patients, affecting approximately one quarter of individuals and have been attributed in part to intra-abdominal adhesions (92). To date, disease-specific assessment of gastrointestinal quality of life in this population has been limited. In the present study, the GIQLI was applied for the first time in patients with AWDs (129). Patients demonstrated significantly lower GIQLI scores compared with controls, indicating a measurable impact of gastrointestinal aspects on daily life. However, no significant differences were observed between groups regarding stool frequency, stool consistency back pain, and signs of gastrointestinal reflux.

Cosmetic outcome and scar appearance represent other relevant aspects of long-term outcome, particularly during adolescence. In the present cohort, cosmetic outcomes were generally rated favorably by both patients and observers. Individual perception of aesthetic results, however, appears to be shaped by multiple factors, including scar location and size, patient expectations, and the broader psychosocial context (171).

Previous studies have identified dissatisfaction related to the absence or altered appearance of the umbilicus as a relevant concern, with individuals reporting associated psychological distress (99, 172). In this cohort, no significant association was identified between the absence of an umbilicus and cosmetic satisfaction. However, perception of cosmetic outcome may evolve over time, particularly during adolescence and young adulthood, when body image becomes increasingly important (173). In line with this, umbilical reconstruction is often considered at a later age in selected patients (99, 172, 174, 175).

A part of our patients experienced abdominal wall–related complications or required additional surgical procedures during childhood, including hernia repair or adhesiolysis. This observation is consistent with published long-term follow-up data and underscores that AWDs might be associated with ongoing surgical morbidity (142, 143). Importantly, in the present cohort, these events did not appear to have a substantial negative impact on overall quality of life.

4.10 Limitations

Some limitations of this study should be acknowledged. The sample size was relatively small, reflecting the rarity of congenital abdominal wall defects and limiting statistical power, particularly for subgroup analyses comparing gastroschisis and omphalocele or uncomplicated (n=10) and complicated (n=1) as well as simple (n=4) and giant (n=2) defects. As a result, subtle differences between subgroups may have remained undetected.

In addition, the study was conducted at a single tertiary care center. While this approach ensured standardized surgical management and follow-up protocols, the generalizability of the results to centers with differing treatment strategies or patient populations may be limited. Furthermore, the cross-sectional design provides a single time-point assessment of long-term outcomes and does not allow conclusions regarding functional trajectories over time. Longitudinal investigations would therefore be required to better understand the evolution of motor performance, pulmonary function, and quality of life from childhood into adulthood.

Although a comprehensive functional assessment was performed, certain outcomes relied on patient-reported questionnaires, which are inherently subjective and may be influenced by individual perception, coping mechanisms, and psychosocial context. Similarly, habitual physical activity was assessed using self-reported measures rather than objective monitoring, which may have introduced reporting bias.

Despite these limitations, the present study offers a detailed and multidimensional evaluation of long-term functional outcomes in patients with abdominal wall defects and contributes novel data to an area in which long-term evidence remains scarce. A particular strength of the study lies in the extended duration of follow-up, with a mean follow-up period of 13 years (range 7–18 years), combined with the inclusion of a control group carefully matched for age, sex, body mass index, and habitual physical activity.

5 Conclusion

In our cohort long-term survivors of GS and OC demonstrate preserved cardiopulmonary function and normal abdominal wall morphology. At the same time, relevant limitations were identified in motor performance and gastrointestinal quality of life, whereas overall cosmetic satisfaction was generally favorable. These findings emphasize the importance of structured, multidisciplinary long-term follow-up strategies that extend beyond anatomical and cardiopulmonary outcome measures. A focus on functional domains may facilitate early identification of emerging limitations and support timely initiation of targeted therapeutic interventions.

6 References

In this work, AI tools (ChatGPT, OpenAI) were used solely for language polishing. All AI-generated outputs were critically reviewed, and revised by me.

1. Flucher C, Windhaber J, Gasparella P, Castellani C, Tschauner S, Mittl B, et al. Long-term motor activity, cardiopulmonary performance and quality of life in abdominal wall defect patients. Pediatr Res. 2024;95(4):1101-9.
2. Jones AM, Isenburg J, Salemi JL, Arnold KE, Mai CT, Aggarwal D, et al. Increasing Prevalence of Gastroschisis--14 States, 1995-2012. MMWR Morb Mortal Wkly Rep. 2016;65(2):23-6.
3. Prefumo F, Izzi C. Fetal abdominal wall defects. Best Pract Res Clin Obstet Gynaecol. 2014;28(3):391-402.
4. Calder J. Two examples of children with preternatural conformation of the guts. In: Edinburgh RSo, editor. Medical Essays and observations. Vol.1. Edinburgh1752. p. 203-6.
5. Curry JI, McKinney P, Thornton JG, Stringer MD. The aetiology of gastroschisis. BJOG. 2000;107(11):1339-46.
6. Torres US, Portela-Oliveira E, Braga Fdel C, Werner H, Jr., Daltro PA, Souza AS. When Closure Fails: What the Radiologist Needs to Know About the Embryology, Anatomy, and Prenatal Imaging of Ventral Body Wall Defects. Semin Ultrasound CT MR. 2015;36(6):522-36.
7. Schierz IAM, Pinello G, Giuffre M, Corsello G. An unusual association of left-sided gastroschisis and persistent right umbilical vein. Clin Case Rep. 2018;6(12):2511-2.
8. Mandelia A, Agarwala S, Sharma N, Solanki S, Panda S. Left-sided Gastroschisis: A Rare Congenital Anomaly. J Clin Diagn Res. 2013;7(10):2300-2.
9. Kirby RS, Marshall J, Tanner JP, Salemi JL, Feldkamp ML, Marengo L, et al. Prevalence and correlates of gastroschisis in 15 states, 1995 to 2005. Obstet Gynecol. 2013;122(2 Pt 1):275-81.
10. Li N, Chen YL, Li J, Li LL, Jiang CZ, Zhou C, et al. Decreasing prevalence and time trend of gastroschisis in 14 cities of Liaoning Province: 2006-2015. Sci Rep. 2016;6:33333.
11. Chuaire Noack L. New clues to understand gastroschisis. Embryology, pathogenesis and epidemiology. Colomb Med (Cali). 2021;52(3):e4004227.
12. Burgos CM, Irvine W, Vivanti A, Conner P, Machtejeviene E, Peters N, et al. European reference network for rare inherited congenital anomalies (ERNICA) evidence based guideline on the management of gastroschisis. Orphanet J Rare Dis. 2024;19(1):60.
13. ERNICA ERN. ERNICA – European Reference Network for rare inherited congenital anomalies [Available from: https://www.ern-ernica.eu.

14. Malhotra R, Malhotra B, Ramteke H. Enhancing Omphalocele Care: Navigating Complications and Innovative Treatment Approaches. Cureus. 2023;15(10):e47638.
15. Salemi JL, Pierre M, Tanner JP, Kornosky JL, Hauser KW, Kirby RS, et al. Maternal nativity as a risk factor for gastroschisis: a population-based study. Birth Defects Res A Clin Mol Teratol. 2009;85(11):890-6.
16. Baldacci S, Santoro M, Coi A, Mezzasalma L, Bianchi F, Pierini A. Lifestyle and sociodemographic risk factors for gastroschisis: a systematic review and meta-analysis. Arch Dis Child. 2020;105(8):756-64.
17. Torfs CP, Velie EM, Oechsli FW, Bateson TF, Curry CJ. A population-based study of gastroschisis: demographic, pregnancy, and lifestyle risk factors. Teratology. 1994;50(1):44-53.
18. Werler MM, Sheehan JE, Mitchell AA. Association of vasoconstrictive exposures with risks of gastroschisis and small intestinal atresia. Epidemiology. 2003;14(3):349-54.
19. Draper ES, Rankin J, Tonks AM, Abrams KR, Field DJ, Clarke M, et al. Recreational drug use: a major risk factor for gastroschisis? Am J Epidemiol. 2008;167(4):485-91.
20. Schmidt AI, Gluer S, Muhlhaus K, Ure BM. Family cases of gastroschisis. J Pediatr Surg. 2005;40(4):740-1.
21. Salinas-Torres VM, Salinas-Torres RA, Cerda-Flores RM, Martinez-de-Villarreal LE. Genetic variants conferring susceptibility to gastroschisis: a phenomenon restricted to the interaction with the environment? Pediatr Surg Int. 2018;34(5):505-14.
22. Marquart JP, Nie Q, Gonzalez T, Jelin AC, Broeckel U, Wagner AJ, et al. Genetics and Genomics of Gastroschisis, Elucidating a Potential Genetic Etiology for the Most Common Abdominal Defect: A Systematic Review. J Dev Biol. 2024;12(4).
23. Arnold MA, Chang DC, Nabaweesi R, Colombani PM, Bathurst MA, Mon KS, et al. Risk stratification of 4344 patients with gastroschisis into simple and complex categories. J Pediatr Surg. 2007;42(9):1520-5.
24. Mastroiacovo P, Lisi A, Castilla EE, Martinez-Frias ML, Bermejo E, Marengo L, et al. Gastroschisis and associated defects: an international study. Am J Med Genet A. 2007;143A(7):660-71.
25. Lausten-Thomsen U, Hedley PL, Conway KM, Lofberg KM, Johansen LS, Romitti PA, et al. Gastroschisis Prevalence and Co-occurring Malformations Among Danish Live Births During 1994-2021: A Nationwide Register-Based Study. J Pediatr Surg. 2024;59(12):161931.
26. Bergholz R, Boettcher M, Reinshagen K, Wenke K. Complex gastroschisis is a different entity to simple gastroschisis affecting morbidity and mortality-a systematic review and meta-analysis. J Pediatr Surg. 2014;49(10):1527-32.
27. Bhat V, Moront M, Bhandari V. Gastroschisis: A State-of-the-Art Review. Children (Basel). 2020;7(12).

28. David AL, Tan A, Curry J. Gastroschisis: sonographic diagnosis, associations, management and outcome. Prenat Diagn. 2008;28(7):633-44.
29. Oakes MC, Porto M, Chung JH. Advances in prenatal and perinatal diagnosis and management of gastroschisis. Semin Pediatr Surg. 2018;27(5):289-99.
30. Lepigeon K, Van Mieghem T, Vasseur Maurer S, Giannoni E, Baud D. Gastroschisis--what should be told to parents? Prenat Diagn. 2014;34(4):316-26.
31. Schmidt CN, Wen T, Friedman AM, D'Alton ME, Andrikopoulou M. Trends in Attempted Vaginal Delivery among Pregnancies Complicated by Gastroschisis, 2014 to 2020. Am J Perinatol. 2024;41(5):543-7.
32. Kirollos DW, Abdel-Latif ME. Mode of delivery and outcomes of infants with gastroschisis: a meta-analysis of observational studies. Arch Dis Child Fetal Neonatal Ed. 2018;103(4):F355-F63.
33. Martins BMR, Abreu I, Meio MDB, Moreira MEL. Gastroschisis in the neonatal period: A prospective case-series in a Brazilian referral center. J Pediatr Surg. 2020;55(8):1546-51.
34. Behram M, Oglak SC, Ozaydin S, Caypinar SS, Gonen I, Tunc S, et al. What is the main factor in predicting the morbidity and mortality in patients with gastroschisis: delivery time, delivery mode, closure method, or the type of gastroschisis (simple or complex)? Turk J Med Sci. 2021;51(3):1587-95.
35. Haddock C, Al Maawali AG, Ting J, Bedford J, Afshar K, Skarsgard ED. Impact of Multidisciplinary Standardization of Care for Gastroschisis: Treatment, Outcomes, and Cost. J Pediatr Surg. 2018;53(5):892-7.
36. Zalles-Vidal C, Penarrieta-Daher A, Bracho-Blanchet E, Ibarra-Rios D, Davila-Perez R, Villegas-Silva R, et al. A Gastroschisis bundle: effects of a quality improvement protocol on morbidity and mortality. J Pediatr Surg. 2018;53(11):2117-22.
37. Baerg J, McAteer J, Miniati D, Somme S, Slidell M, American Pediatric Surgical Association Outcomes Evidence-based Practice C. Improving outcomes for uncomplicated gastroschisis: clinical practice guidelines from the American Pediatric Surgical Association Outcomes and Evidence-based Practice Committee. Pediatr Surg Int. 2024;40(1):246.
38. Rentea RM, Gupta V. Gastroschisis. StatPearls. Treasure Island (FL)2025.
39. Rosen O, Angert RM. Gastroschisis Simulation Model: Pre-surgical Management Technical Report. Cureus. 2017;9(3):e1109.
40. Jansen LA, Safavi A, Lin Y, MacNab YC, Skarsgard ED, Canadian Pediatric Surgery N. Preclosure fluid resuscitation influences outcome in gastroschisis. Am J Perinatol. 2012;29(4):307-12.
41. Al Maawali A, Skarsgard ED. The medical and surgical management of gastroschisis. Early Hum Dev. 2021;162:105459.
42. Puri P, Höllwarth M. Pediatric Surgery. 1 ed. Berlin: Springer-Verlag; 2006.

43. Tullie LG, Bough GM, Shalaby A, Kiely EM, Curry JI, Pierro A, et al. Umbilical hernia following gastroschisis closure: a common event? Pediatr Surg Int. 2016;32(8):811-4.
44. Schlatter M, Norris K, Uitvlugt N, DeCou J, Connors R. Improved outcomes in the treatment of gastroschisis using a preformed silo and delayed repair approach. J Pediatr Surg. 2003;38(3):459-64; discussion -64.
45. Pastor AC, Phillips JD, Fenton SJ, Meyers RL, Lamm AW, Raval MV, et al. Routine use of a SILASTIC spring-loaded silo for infants with gastroschisis: a multicenter randomized controlled trial. J Pediatr Surg. 2008;43(10):1807-12.
46. Sandler A, Lawrence J, Meehan J, Phearman L, Soper R. A "plastic" sutureless abdominal wall closure in gastroschisis. J Pediatr Surg. 2004;39(5):738-41.
47. Machida M, Takamizawa S, Yoshizawa K. Umbilical cord inverting technique: a simple method to utilize the umbilical cord as a biologic dressing for sutureless gastroschisis closure. Pediatr Surg Int. 2011;27(1):95-7.
48. Diyaolu M, Wood LS, Bruzoni M. Sutureless closure for the management of gastroschisis. Transl Gastroenterol Hepatol. 2021;6:31.
49. Fraser JD, Deans KJ, Fallat ME, Helmrath MA, Kabre R, Leys CM, et al. Sutureless vs sutured abdominal wall closure for gastroschisis: Operative characteristics and early outcomes from the Midwest Pediatric Surgery Consortium. J Pediatr Surg. 2020;55(11):2284-8.
50. Baird R, Eeson G, Safavi A, Puligandla P, Laberge JM, Skarsgard ED, et al. Institutional practice and outcome variation in the management of congenital diaphragmatic hernia and gastroschisis in Canada: a report from the Canadian Pediatric Surgery Network. J Pediatr Surg. 2011;46(5):801-7.
51. Williams SL, Tkach JA, Rattan MS, South AP, Wessel J, Kingma PS. Feeding Tolerance, Intestinal Motility, and Superior Mesenteric Artery Blood Flow in Infants with Gastroschisis. Neonatology. 2020;117(1):95-101.
52. Embleton ND, van den Akker CHP, Johnson M. Parenteral nutrition for preterm infants: benefits and risks in 2025. Semin Fetal Neonatal Med. 2025;30(2):101635.
53. Groh-Wargo S, Barr SM. Parenteral Nutrition. Clin Perinatol. 2022;49(2):355-79.
54. Wallace MW, Danko ME, Zamora IJ, Morris EA, Li J, Froehlich M, et al. Infectious Complications and Antibiotic Use in Gastroschisis. Surg Infect (Larchmt). 2023;24(5):405-13.
55. Baird R, Puligandla P, Skarsgard E, Laberge JM, Canadian Pediatric Surgical N. Infectious complications in the management of gastroschisis. Pediatr Surg Int. 2012;28(4):399-404.
56. Uribe-Leitz M, McCracken CE, Heiss KF, Wulkan ML, Raval MV. The Influence of Infectious Complications in Gastroschisis on Costs and Length of Stay. Am J Perinatol. 2017;34(1):62-9.

57. Bence CM, Landisch RM, Wu R, Szabo A, McElhinney K, Austin MT, et al. Risk factors for perioperative hypothermia and infectious outcomes in gastroschisis patients. J Pediatr Surg. 2021;56(7):1107-12.
58. Palatnik A, Loichinger M, Wagner A, Peterson E. The association between gestational age at delivery, closure type and perinatal outcomes in neonates with isolated gastroschisis. J Matern Fetal Neonatal Med. 2020;33(8):1393-9.
59. Gurien LA, Dassinger MS, Burford JM, Saylors ME, Smith SD. Does timing of gastroschisis repair matter? A comparison using the ACS NSQIP pediatric database. J Pediatr Surg. 2017;52(11):1751-4.
60. Sangkhathat S, Patrapinyokul S, Chiengkriwate P, Chanvitan P, Janjindamai W, Dissaneevate S. Infectious complications in infants with gastroschisis: an 11-year review from a referral hospital in southern Thailand. J Pediatr Surg. 2008;43(3):473-8.
61. De Bie F, Swaminathan V, Johnson G, Monos S, Adzick NS, Laje P. Long-term core outcomes of patients with simple gastroschisis. J Pediatr Surg. 2021;56(8):1365-9.
62. Suominen J, Rintala R. Medium and long-term outcomes of gastroschisis. Semin Pediatr Surg. 2018;27(5):327-9.
63. Baerg JE, Munoz AN. Long term complications and outcomes in omphalocele. Semin Pediatr Surg. 2019;28(2):118-21.
64. Caldeman C, Fogelstrom A, Wester T, Mesas Burgos C, Lof Granstrom A. Long-term gastrointestinal morbidity in patients born with gastroschisis: A national register-based cohort study. J Pediatr Gastroenterol Nutr. 2024;79(5):983-90.
65. Arnold HE, Baxter KJ, Short HL, Travers C, Bhatia A, Durham MM, et al. Short-term and family-reported long-term outcomes of simple versus complicated gastroschisis. J Surg Res. 2018;224:79-88.
66. Rundell MR, Bailey RA, Wagner AJ, Warner BB, Miller LE. Long-Term Neurodevelopmental Outcomes in Children with Gastroschisis: A Review of the Literature. Am J Perinatol. 2025;42(2):147-63.
67. Gorra AS, Needelman H, Azarow KS, Roberts HJ, Jackson BJ, Cusick RA. Long-term neurodevelopmental outcomes in children born with gastroschisis: the tiebreaker. J Pediatr Surg. 2012;47(1):125-9.
68. Raitio A, Syvanen J, Tauriainen A, Hyvarinen A, Sankilampi U, Gissler M, et al. Congenital abdominal wall defects and cryptorchidism: a population-based study. Pediatr Surg Int. 2021;37(7):837-41.
69. Ceccanti S, Migliara G, De Vito C, Cozzi DA. Prevalence, management, and outcome of cryptorchidism associated with gastroschisis: A systematic review and meta-analysis. J Pediatr Surg. 2022;57(7):1414-22.

70. Lawson A, de La Hunt MN. Gastroschisis and undescended testis. J Pediatr Surg. 2001;36(2):366-7.
71. Umeda S, Takayama K, Takase K, Kim K, Yamamichi T, Tayama A, et al. Clinical factors related to undescended testis in infants with gastroschisis. Pediatr Int. 2022;64(1):e15054.
72. Wiebe ME, Kattini C, Larocca V, Thorburn C, Hayawi L, Bijelic V, et al. Quality of Life Outcomes in Children Following Surgical Management of Gastroschisis: A Systematic Review and Meta-analysis. J Pediatr Surg. 2025;60(4):162222.
73. Kaiser MM, Kahl F, von Schwabe C, Halsband H. [Omphalocele and gastroschisis. Outcome--complications--follow-up--quality of life]. Chirurg. 2000;71(10):1256-62.
74. Snoep MC, de Heus R, Manten GTR, Lap C, Snoeker BAM, Lindeboom MYA. Gastro-intestinal function and quality of life are favorable in adolescent and adult gastroschisis patients. Early Hum Dev. 2020;141:104936.
75. Corey KM, Hornik CP, Laughon MM, McHutchison K, Clark RH, Smith PB. Frequency of anomalies and hospital outcomes in infants with gastroschisis and omphalocele. Early Hum Dev. 2014;90(8):421-4.
76. Springett A, Draper ES, Rankin J, Rounding C, Tucker D, Stoianova S, et al. Birth prevalence and survival of exomphalos in england and wales: 2005 to 2011. Birth Defects Res A Clin Mol Teratol. 2014;100(9):721-5.
77. Stallings EB, Isenburg JL, Short TD, Heinke D, Kirby RS, Romitti PA, et al. Population-based birth defects data in the United States, 2012-2016: A focus on abdominal wall defects. Birth Defects Res. 2019;111(18):1436-47.
78. Nembhard WN, Bergman JEH, Politis MD, Arteaga-Vazquez J, Bermejo-Sanchez E, Canfield MA, et al. A multi-country study of prevalence and early childhood mortality among children with omphalocele. Birth Defects Res. 2020;112(20):1787-801.
79. Khan FA, Hashmi A, Islam S. Insights into embryology and development of omphalocele. Semin Pediatr Surg. 2019;28(2):80-3.
80. Kluth D, Lambrecht W. The pathogenesis of omphalocele and gastroschisis : An unsolved problem. Pediatr Surg Int. 1996;11(2-3):62-6.
81. Sadler TW. The embryologic origin of ventral body wall defects. Semin Pediatr Surg. 2010;19(3):209-14.
82. Khan FA, Raymond SL, Hashmi A, Islam S. Anatomy and embryology of abdominal wall defects. Semin Pediatr Surg. 2022;31(6):151230.
83. Gilbert WM, Nicolaides KH. Fetal omphalocele: associated malformations and chromosomal defects. Obstet Gynecol. 1987;70(4):633-5.
84. Frolov P, Alali J, Klein MD. Clinical risk factors for gastroschisis and omphalocele in humans: a review of the literature. Pediatr Surg Int. 2010;26(12):1135-48.

85. Weksberg R, Shuman C, Beckwith JB. Beckwith-Wiedemann syndrome. Eur J Hum Genet. 2010;18(1):8-14.
86. Brioude F, Kalish JM, Mussa A, Foster AC, Bliek J, Ferrero GB, et al. Expert consensus document: Clinical and molecular diagnosis, screening and management of Beckwith-Wiedemann syndrome: an international consensus statement. Nat Rev Endocrinol. 2018;14(4):229-49.
87. Watkins ML, Rasmussen SA, Honein MA, Botto LD, Moore CA. Maternal obesity and risk for birth defects. Pediatrics. 2003;111(5 Pt 2):1152-8.
88. Mac Bird T, Robbins JM, Druschel C, Cleves MA, Yang S, Hobbs CA, et al. Demographic and environmental risk factors for gastroschisis and omphalocele in the National Birth Defects Prevention Study. J Pediatr Surg. 2009;44(8):1546-51.
89. Raymond SL, Downard CD, St Peter SD, Baerg J, Qureshi FG, Bruch SW, et al. Outcomes in omphalocele correlate with size of defect. J Pediatr Surg. 2019;54(8):1546-50.
90. Akinkuotu AC, Sheikh F, Olutoye OO, Lee TC, Fernandes CJ, Welty SE, et al. Giant omphaloceles: surgical management and perinatal outcomes. J Surg Res. 2015;198(2):388-92.
91. Nolan HR, Wagner ML, Jenkins T, Lim FY. Outcomes in the giant omphalocele population: A single center comprehensive experience. J Pediatr Surg. 2020;55(9):1866-71.
92. van Eijck FC, Wijnen RM, van Goor H. The incidence and morbidity of adhesions after treatment of neonates with gastroschisis and omphalocele: a 30-year review. J Pediatr Surg. 2008;43(3):479-83.
93. Stoll C, Alembik Y, Dott B, Roth MP. Omphalocele and gastroschisis and associated malformations. Am J Med Genet A. 2008;146A(10):1280-5.
94. Shi X, Tang H, Lu J, Yang X, Ding H, Wu J. Prenatal genetic diagnosis of omphalocele by karyotyping, chromosomal microarray analysis and exome sequencing. Ann Med. 2021;53(1):1285-91.
95. Cantrell JR, Haller JA, Ravitch MM. A syndrome of congenital defects involving the abdominal wall, sternum, diaphragm, pericardium, and heart. Surg Gynecol Obstet. 1958;107(5):602-14.
96. Khoummane N, Abakka S. OEIS complex. Pan Afr Med J. 2013;16:129.
97. Tobin M, Gunaji R, Walsh JC, Grice GP. A review of genetic factors underlying craniorachischisis and omphalocele: Inspired by a unique trisomy 18 case. Am J Med Genet A. 2019;179(8):1642-51.
98. Pyle AK, George TN, Cummings JJ, Laventhal NT, Committee on B, Section on Neonatal-Perinatal M, et al. Guidance for Caring for Infants and Children With Trisomy 13 and Trisomy 18: Clinical Report. Pediatrics. 2025;156(2).

99. Gamba P, Midrio P. Abdominal wall defects: prenatal diagnosis, newborn management, and long-term outcomes. Semin Pediatr Surg. 2014;23(5):283-90.
100. Bence CM, Wagner AJ. Abdominal wall defects. Transl Pediatr. 2021;10(5):1461-9.
101. Akinkuotu AC, Sheikh F, Cass DL, Zamora IJ, Lee TC, Cassady CI, et al. Are all pulmonary hypoplasias the same? A comparison of pulmonary outcomes in neonates with congenital diaphragmatic hernia, omphalocele and congenital lung malformation. J Pediatr Surg. 2015;50(1):55-9.
102. Danzer E, Hedrick HL, Rintoul NE, Siegle J, Adzick NS, Panitch HB. Assessment of early pulmonary function abnormalities in giant omphalocele survivors. J Pediatr Surg. 2012;47(10):1811-20.
103. Fawley JA, Peterson EL, Christensen MA, Rein L, Wagner AJ. Can omphalocele ratio predict postnatal outcomes? J Pediatr Surg. 2016;51(1):62-6.
104. Ayub SS, Taylor JA. Cardiac anomalies associated with omphalocele. Semin Pediatr Surg. 2019;28(2):111-4.
105. Roux N, Jakubowicz D, Salomon L, Grange G, Giuseppi A, Rousseau V, et al. Early surgical management for giant omphalocele: Results and prognostic factors. J Pediatr Surg. 2018;53(10):1908-13.
106. Gonzalez KW, Chandler NM. Ruptured omphalocele: Diagnosis and management. Semin Pediatr Surg. 2019;28(2):101-5.
107. Lestari PM, Bernolian N, Mirani P, Martadiansyah A, Latifah ME, Sutrisno MAF, et al. Prenatal diagnosis of omphalocele with extracorporeal liver. Radiol Case Rep. 2024;19(12):5896-9.
108. Bauman B, Stephens D, Gershone H, Bongiorno C, Osterholm E, Acton R, et al. Management of giant omphaloceles: A systematic review of methods of staged surgical vs. nonoperative delayed closure. J Pediatr Surg. 2016;51(10):1725-30.
109. Kethman WC, Sinclair TJC, Abrajano CT, Chao S, Wall JK. Case Report: Rapid staged abdominal closure using Gore-Tex® mesh as a bridge to primary omphalocele sac closure. Journal of Pediatric Surgery Case Reports. 2016;9:37-9.
110. Schuster SR. A new method for the staged repair of large omphaloceles. Surg Gynecol Obstet. 1967;125(4):837-50.
111. Kondra K, Jimenez C, Stanton E, Chen K, Shin CE, Hammoudeh JA. Meeting in the middle: pediatric abdominal wall reconstruction for omphalocele. Pediatr Surg Int. 2022;38(12):1981-7.
112. Skarsgard ED. Immediate versus staged repair of omphaloceles. Semin Pediatr Surg. 2019;28(2):89-94.
113. Duggan E, Puligandla PS. Respiratory disorders in patients with omphalocele. Semin Pediatr Surg. 2019;28(2):115-7.

114. Danzer E, Victoria T, Bebbington MW, Siegle J, Rintoul NE, Johnson MP, et al. Fetal MRI-calculated total lung volumes in the prediction of short-term outcome in giant omphalocele: preliminary findings. Fetal Diagn Ther. 2012;31(4):248-53.
115. Baerg JE, Thirumoorthi A, Carlton W, Haug S, Hopper AO, Goff D, et al. Late onset of pulmonary hypertension and sepsis in omphalocele infants. Journal of Pediatric Surgery Case Reports. 2016;15:14-8.
116. Edwards EA, Broome S, Green S, Douglas C, McCall E, Nuthall G, et al. Long-term respiratory support in children with giant omphalocele. Anaesth Intensive Care. 2007;35(1):94-8.
117. Fawley JA, Abdelhafeez AH, Schultz JA, Ertl A, Cassidy LD, Peter SS, et al. The risk of midgut volvulus in patients with abdominal wall defects: A multi-institutional study. J Pediatr Surg. 2017;52(1):26-9.
118. Abdelhafeez AH, Schultz JA, Ertl A, Cassidy LD, Wagner AJ. The risk of volvulus in abdominal wall defects. J Pediatr Surg. 2015;50(4):570-2.
119. Danzer E, Gerdes M, D'Agostino JA, Bernbaum J, Hoffman C, Rintoul NE, et al. Patient characteristics are important determinants of neurodevelopmental outcome during infancy in giant omphalocele. Early Hum Dev. 2015;91(3):187-93.
120. van Eijck FC, Hoogeveen YL, van Weel C, Rieu PN, Wijnen RM. Minor and giant omphalocele: long-term outcomes and quality of life. J Pediatr Surg. 2009;44(7):1355-9.
121. Thomas E, De Benedetti L, Parente G, Di Mitri M, Cravano SM, D'Antonio S, et al. Long-Term Assessment of Aesthetic Results in Omphalocele Repair with POSAS Scale. Aesthetic Plast Surg. 2024;48(23):5171-9.
122. Ghattaura H, Ross A, Aldeiri B, Mutanen A, Saxena A. Managing giant omphalocele: A systematic review of surgical techniques and outcomes. Acta Paediatr. 2024;113(11):2459-65.
123. Harris EL, Minutillo C, Hart S, Warner TM, Ravikumara M, Nathan EA, et al. The long term physical consequences of gastroschisis. J Pediatr Surg. 2014;49(10):1466-70.
124. Arneitz C, Windhaber J, Castellani C, Kienesberger B, Klymiuk I, Fasching G, et al. Cardiorespiratory performance capacity and airway microbiome in patients following primary repair of esophageal atresia. Pediatr Res. 2021;90(1):66-73.
125. Arneitz C, Windhaber J, Flucher C, Gasparella P, Amerstorfer E, Huber-Zeyringer A, et al. Cardiorespiratory performance and locomotor function of patients with anorectal malformations. Sci Rep. 2021;11(1):18919.
126. Zaccara A, Iacobelli BD, Calzolari A, Turchetta A, Orazi C, Schingo P, et al. Cardiopulmonary performances in young children and adolescents born with large abdominal wall defects. J Pediatr Surg. 2003;38(3):478-81; discussion -81.

127. Windhaber J, Steinbauer M, Holter M, Wieland A, Kogler K, Riedl R, et al. Bicycle spiroergometry: comparison of standardized examination protocols for adolescents: is it necessary to define own standard values for each protocol? Eur J Appl Physiol. 2021;121(6):1783-94.
128. Jouck S, Koch B, Graf C, Predel HG, Dordel S. Dordel-Koch-Test (DKT) – zur Erfassung der motorischen Basisfunktionen im Kindes- und Jugendalter; erste Ergebnisse der Normierung. Aktuelle Ernährungsmedizin. 2006;31(05):P27.
129. Eypasch E, Williams JI, Wood-Dauphinee S, Ure BM, Schmulling C, Neugebauer E, et al. Gastrointestinal Quality of Life Index: development, validation and application of a new instrument. Br J Surg. 1995;82(2):216-22.
130. Laje P, Fraga MV, Peranteau WH, Hedrick HL, Khalek N, Gebb JS, et al. Complex gastroschisis: Clinical spectrum and neonatal outcomes at a referral center. J Pediatr Surg. 2018;53(10):1904-7.
131. Skrabal F, Pichler GP, Penatzer M, Steinbichl J, Hanserl AK, Leis A, et al. The Combyn ECG: Adding haemodynamic and fluid leads for the ECG. Part II: Prediction of total body water (TBW), extracellular fluid (ECF), ECF overload, fat mass (FM) and "dry" appendicular muscle mass (AppMM). Med Eng Phys. 2017;44:44-52.
132. Jat KR. Spirometry in children. Prim Care Respir J. 2013;22(2):221-9.
133. Fletcher GF, Ades PA, Kligfield P, Arena R, Balady GJ, Bittner VA, et al. Exercise standards for testing and training: a scientific statement from the American Heart Association. Circulation. 2013;128(8):873-934.
134. Mezzani A, Corra U, Bosimini E, Giordano A, Giannuzzi P. Contribution of peak respiratory exchange ratio to peak VO2 prognostic reliability in patients with chronic heart failure and severely reduced exercise capacity. Am Heart J. 2003;145(6):1102-7.
135. Swedenhammar E, Wahlstrom O, Brandt JD, Strigard K, Hager C, Stark B, et al. Reliability and validity of surface EMG assessments combined with isometric muscle strength testing in patients with abdominal rectus diastasis and asymptomatic controls. Hernia. 2024;28(4):1413-26.
136. McGill S, Juker D, Kropf P. Appropriately placed surface EMG electrodes reflect deep muscle activity (psoas, quadratus lumborum, abdominal wall) in the lumbar spine. J Biomech. 1996;29(11):1503-7.
137. Ng JK, Kippers V, Parnianpour M, Richardson CA. EMG activity normalization for trunk muscles in subjects with and without back pain. Med Sci Sports Exerc. 2002;34(7):1082-6.
138. Lewis SJ, Heaton KW. Stool form scale as a useful guide to intestinal transit time. Scand J Gastroenterol. 1997;32(9):920-4.

139. Van de Kar AL, Corion LU, Smeulders MJ, Draaijers LJ, Van der Horst CM, Van Zuijlen PP. Reliable and feasible evaluation of linear scars by the Patient and Observer Scar Assessment Scale. Plast Reconstr Surg. 2005;116(2):514-22.
140. Draaijers LJ, Tempelman FR, Botman YA, Tuinebreijer WE, Middelkoop E, Kreis RW, et al. The patient and observer scar assessment scale: a reliable and feasible tool for scar evaluation. Plast Reconstr Surg. 2004;113(7):1960-5; discussion 6-7.
141. Kemme FM, Mieras A, Ket JCF, Meij-de Vries A, van Zuijlen PPM, Pijpe A. How to Assess Scar Quality in Pediatric Burn Patients: A Systematic Review on the Type and Content of Outcome Measurement Instruments. J Burn Care Res. 2025;46(6):1294-301.
142. Raitio A, Kalliokoski N, Syvanen J, Harju S, Tauriainen A, Hyvarinen A, et al. High incidence of inguinal hernias among patients with congenital abdominal wall defects: a population-based case-control study. Eur J Pediatr. 2021;180(8):2693-8.
143. Raitio A, Syvanen J, Tauriainen A, Hyvarinen A, Sankilampi U, Gissler M, et al. Long-term hospital admissions and surgical treatment of children with congenital abdominal wall defects: a population-based study. Eur J Pediatr. 2021;180(7):2193-8.
144. Hijkoop A, Peters NCJ, Lechner RL, van Bever Y, van Gils-Frijters A, Tibboel D, et al. Omphalocele: from diagnosis to growth and development at 2 years of age. Arch Dis Child Fetal Neonatal Ed. 2019;104(1):F18-F23.
145. Nair N, Merhar S, Wessel J, Hall E, Kingma PS. Factors that Influence Longitudinal Growth from Birth to 18 Months of Age in Infants with Gastroschisis. Am J Perinatol. 2020;37(14):1438-45.
146. Martins BMR, Souza NCO, Meio M, Rebelo F, Ribeiro CTM. Neurodevelopment of children born with gastroschisis: a scoping review. Pediatr Res. 2025.
147. Btaiche IF, Khalidi N. Parenteral nutrition-associated liver complications in children. Pharmacotherapy. 2002;22(2):188-211.
148. Gipsman AI, Danzer E, Aarthun A, Mathew L, Flohr SJ, Avitabile CM, et al. Long-Term Pulmonary Function Outcomes in Children with Pulmonary Hypoplasia. J Pediatr. 2025;285:114671.
149. Ortega FB, Ruiz JR, Castillo MJ, Sjostrom M. Physical fitness in childhood and adolescence: a powerful marker of health. Int J Obes (Lond). 2008;32(1):1-11.
150. Demchenko I, Prince SA, Merucci K, Cadenas-Sanchez C, Chaput JP, Fraser BJ, et al. Cardiorespiratory fitness and health in children and adolescents: an overview of systematic reviews with meta-analyses representing over 125 000 observations covering 33 health-related outcomes. Br J Sports Med. 2025;59(12):856-65.
151. Garcia-Hermoso A, Ramirez-Velez R, Garcia-Alonso Y, Alonso-Martinez AM, Izquierdo M. Association of Cardiorespiratory Fitness Levels During Youth With Health Risk Later in Life: A Systematic Review and Meta-analysis. JAMA Pediatr. 2020;174(10):952-60.

152. Burton AM, Cowburn I, Thompson F, Eisenmann JC, Nicholson B, Till K. Associations Between Motor Competence and Physical Activity, Physical Fitness and Psychosocial Characteristics in Adolescents: A Systematic Review and Meta-analysis. Sports Med. 2023;53(11):2191-256.
153. Roorda D, Konigs M, Eeftinck Schattenkerk L, van der Steeg L, van Heurn E, Oosterlaan J. Neurodevelopmental outcome of patients with congenital gastrointestinal malformations: a systematic review and meta-analysis. Arch Dis Child Fetal Neonatal Ed. 2021;106(6):635-42.
154. van den Hondel D, Sloots CE, Gischler SJ, Meeussen CJ, Wijnen RM, Jsselstijn HI, et al. Prospective long-term follow up of children with anorectal malformation: growth and development until 5years of age. J Pediatr Surg. 2013;48(4):818-25.
155. Wrotniak BH, Epstein LH, Dorn JM, Jones KE, Kondilis VA. The relationship between motor proficiency and physical activity in children. Pediatrics. 2006;118(6):e1758-65.
156. Adank AM, Van Kann DHH, Hoeboer JJAA, de Vries SI, Kremers SPJ, Vos SB. Investigating Motor Competence in Association with Sedentary Behavior and Physical Activity in 7- to 11-Year-Old Children. Int J Environ Res Public Health. 2018;15(11).
157. Higashihara M, Sonoo M, Ishiyama A, Nagashima Y, Matsumoto K, Uesugi H, et al. Quantitative Analysis of Surface Electromyography for Pediatric Neuromuscular Disorders. Muscle Nerve. 2018;58(6):824-7.
158. Maizels M, Firlit CF. Pediatric urodynamics: a clinical comparison of surface versus needle pelvic floor/external sphincter electromyography. J Urol. 1979;122(4):518-22.
159. Aragon-Ramos P, Garcia-Lopez I, Santiago S, Martinez A, Gavilan J. Laryngeal electromyography, a useful tool in difficult cases of pediatric laryngeal mobility disorders. Int J Pediatr Otorhinolaryngol. 2022;161:111264.
160. Staab V. Management of Abdominal Wall Defects. Surg Clin North Am. 2022;102(5):809-20.
161. Mannion AF, Pulkovski N, Toma V, Sprott H. Abdominal muscle size and symmetry at rest and during abdominal hollowing exercises in healthy control subjects. J Anat. 2008;213(2):173-82.
162. Lehman GJ, McGill SM. Quantification of the differences in electromyographic activity magnitude between the upper and lower portions of the rectus abdominis muscle during selected trunk exercises. Phys Ther. 2001;81(5):1096-101.
163. Armand S, Decoulon G, Bonnefoy-Mazure A. Gait analysis in children with cerebral palsy. EFORT Open Rev. 2016;1(12):448-60.
164. Khouri N, Desailly E. Contribution of clinical gait analysis to single-event multi-level surgery in children with cerebral palsy. Orthop Traumatol Surg Res. 2017;103(1S):S105-S11.

165. Johnson AW, Adams L, Kho JB, Green DM, Pace NB, Mitchell UH. Extended field-of-view ultrasound imaging is reliable for measuring Transversus Abdominis muscle size at rest and during contraction. BMC Musculoskelet Disord. 2021;22(1):282.
166. Plumb AA, Windsor ACJ, Ross D. Contemporary imaging of rectus diastasis and the abdominal wall. Hernia. 2021;25(4):921-7.
167. Pirri C, Todros S, Fede C, Pianigiani S, Fan C, Foti C, et al. Inter-rater reliability and variability of ultrasound measurements of abdominal muscles and fasciae thickness. Clin Anat. 2019;32(7):948-60.
168. ShahAli S, Shanbehzadeh S, ShahAli S, Ebrahimi Takamjani I. Application of Ultrasonography in the Assessment of Abdominal and Lumbar Trunk Muscle Activity in Participants With and Without Low Back Pain: A Systematic Review. J Manipulative Physiol Ther. 2019;42(7):541-50.
169. Phyo AZZ, Freak-Poli R, Craig H, Gasevic D, Stocks NP, Gonzalez-Chica DA, et al. Quality of life and mortality in the general population: a systematic review and meta-analysis. BMC Public Health. 2020;20(1):1596.
170. Hijkoop A, Rietman AB, Wijnen RMH, Tibboel D, Cohen-Overbeek TE, van Rosmalen J, et al. Gastroschisis at school age: what do parents report? Eur J Pediatr. 2019;178(9):1405-12.
171. Reindl N, Seitz S, Schleier M, Besendorfer M, Diez S. Long-term follow-up of scar quality and satisfaction after surgical closure of congenital abdominal wall defects: a single center perspective. Pediatr Surg Int. 2025;42(1):57.
172. Bongini M, Tanini S, Messineo A, Facchini F, Ghionzoli M. Umbilical reconstruction in children: a simplified operative technique. Aesthetic Plast Surg. 2015;39(3):414-7.
173. Imren C, Jsselstijn HI, Vermeulen MJ, Wijnen RHM, Rietman AB, Keyzer-Dekker CMG. Scar Perception in School-aged Children After Major Surgery in Infancy. J Pediatr Surg. 2024;59(11):161659.
174. Gardani M, Palli D, Simonacci F, Grieco MP, Bertozzi N, Raposio E. Umbilical reconstruction: different techniques, a single aim. Acta Biomed. 2019;90(4):504-9.
175. Lee Y, Lee SH, Woo KV. Umbilical reconstruction using a modified inverted C-V flap with conjoint flaps. J Plast Surg Hand Surg. 2013;47(4):334-6.