Annals of Hematology & Oncology

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Special Article - Abdominal Wall Defects and Hernias

Abdominal Wall Defects in Prenatal Medicine

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Received: May 03, 2022; **Accepted:** June 18, 2022; **Published:** June 25, 2022

Keywords

Fetus; Embryo; Embryo Fetal Development; Gastroschisis; Omphalocele; Pentalogy of Cantrell; Body Stalk Anomaly; Prenatal Medicine; Prenatal Ultrasound; Prenatal Investigation; Umbilical Cord; Umbilical Hernia; Limb Body Wall Complex; Ectopia Cordis; Bladder Exstrophy; Ventral Body Wall Defects; Abdominal Wall Defects; Amniotic band.

Abbreviations

ABS: Amniotic Band Syndrome; AWD: Abdominal Wall Defects; BE: Bladder Extrophy; BSA: Body Stalk Anomaly; CE: Cloacal Extrophy; EC: Ectopia Cordis; EFD: Embryo Fetal Development; G: Gastroschisis; MRI: Magnetic Resonance Imaging; O: Omphalocele; OIES: Omphalocele Estropy Imperforatedanus Spinal Defect; PBS: Prune Belly Syndrome; PM: Prenatal Medicine; PCO: Pentalogy of Cantrell; UC: Umbilical Cord; UE: Umbilical Hernia; US: Ultrasound; VAWD/AWDs: Ventral Abdominal Wall Defects; 2D/3D: two/three dimensional US.

Short Communication

Abdominal Wall Defects (AWD) are anomalies of prenatal development which can manifest with different types of entities, also associated with malformations of the other fetal body districts, and are one of the main live birth defects; diagnosed in prenatal life, thanks to Prenatal Medicine (PM) monitoring protocols, those defects, with different anatomical characteristics, are likely caused by several multifactorial events that compromise the physiological development of the embryo – fetal anterior abdominal wall. The aim of our brief communication is to retrace the significant steps of the discovery and disclosure of this human intrauterine life pathology.

Total ADW have, at our time, a prevalence of about eight in 10.000 ([1]: European surveillance of congenital anomalies, Chart 1-A,B,C,D); the importance of the data called prevalence, used to plan health prevention and to understand the damage inducted by a specific pathogenic event in a population, should always be emphasized; it expresses the ratio between the number of affected people in a subjects of a specific territory, at a defined time, and the total number of individuals in the population, at the same period; hence, several international studies had given it in a lot value, as well as for epidemiological surveillance [2].

Figure 1: (from the original [2], totally identical to the original, with the permission of The Royal Society Publishing).

Figure 1.Tab.XVIII.

Explanation: the heart, blood-vessels, liver, as appeared when dissected; part of the ribs, sternum, thymus gland, lungs, have been removed, **A:** the heart, consisting in one auricle and one ventricle; **B:** a larg arterial trunk, arising from the ventricle; **C:** the aorta, arising from this trunk; **D:** the pulmonary artery, arising from the same trunk; **E:** the vena cava superior, descending on the left side; **F:** the pulmonary veins, entering into the auricle with the vena cava superior; **G:** the vena azygos, ascending on the left side; **H:** the diaphragm, adhering laterally to the margin of the chest, but deficient on the fore part, towards the sternum; **I:** the liver; **K:** the cavity on the upper surface of the liver, in which the heart was in part situated; **L:** the membranous covering turned downwards; **M:** the umbilical vein.

Figure 2.Tab.XVIII.

Explanation: represents the heart; the aorta and pulmonary artery are cut off near their origin, to shew the pulmonary veins, and vena cava superior, entering the auricle.

A: the auricle; **B:** the ventricle; **C:** the trunk from which the aorta and pulmonary artery arose; **D:** a large vessel entering the auricle, and receiving the blood from the pulmonary veins and vena cava superior; **E:** the trunk formed by the pulmonary veins and vena cava superior; **F:** the vena cava superior; **G:** the vena azygos; **H:** the right subclavian vein; **I:** the left subclavian vein.

In hystorical reviews, as early as 1734, even noticed by other scientists since the half of sixteenth century, Prochaska [3] reported an analysis of the abnormality noted in lower sternal area of the

Citation: Solerte ML and Cosmi E. Abdominal Wall Defects in Prenatal Medicine. Ann Hematol Oncol. 2022; 9(2): 1394.

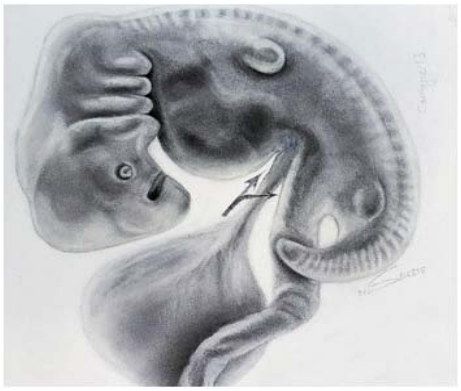

Figure 2: Scientific illustration shows a normal human embryo in Carnegie 13 morphological stage; the arrows indicate the areas involved in the anomalies of embrio-fetal development that lead to the VAWD/AWDs malformations; here, two stalks, yolk and body, develop as separate structures (credit Maria Laura Solerte).

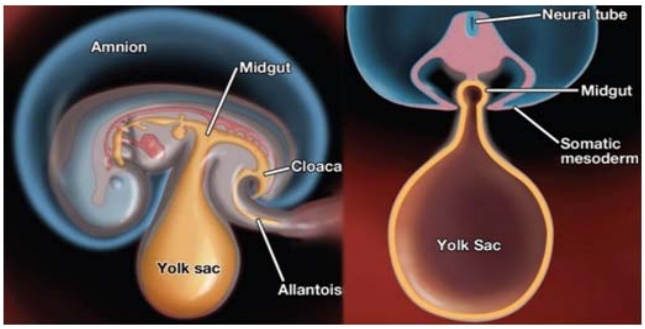

Figure 3: Normal embryonic development diagrams of an embryo shows craniocaudal folding.
Sagittal view1: the yolk sac is in direct continuity with the midgut; the allantois extends from the cloaca into the connecting stalk, which will ultimately develop into the umbilical cord.
Cross-sectional view 2: embryo shows lateral folding; the somatic mesoderm grows ventrally and medially, pinching off the yolk sac; this results in an outer tube (abdominal wall) surrounding an inner tube (gastrointestinal tract).
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newborn, with eventration of abdominal wall, more anencephaly; afterwards, in 1798 [4], Wilson issued an unusual formation of the human heart, describing, in detail, autopsy findings, also reproduced in an anatomic table (Figure 1).

Reports of similar anomalies followed, with multiple forms of phenotypic manifestations.

At later time, Cantrell and his group [5], described patients affected by a singular combination of congenital malformations, with supraumbilical abdominal wall, sternum, diaphragm and pericardium defects, assuming an embryological basis of those severe pathologies, result from the failure of embryo fold; he started to outline an embryo diagrammatic, cross section, reconstruction of the normal development at 16-17 days and his mesoderm layer, probably alterated at a very early period of the embryo life.

Carey and his collaborators [6], reported the Omphalocele (O) more Entropy more Imperforated anus more Spinal Defect complex syndrome (OIES) in several cases, already described previously, with a certain male predominance, due to an early arrest and/or alterated embryological growth, associated with chromosomal aneuploidies.

Ultrasound (US) examination in PM, thanks to advanced technologies, took over decades after decades, the main role in the diagnosis and management of AWD.

In 1986, Lockwood, Hobbins et al [7] detected the presence of an extreme abdominal and thoracic herniation probably due to umbilical cord and embryo body folding maldevelopment, included an amnio-chorion anomalous fusion.

Four years later, Jauniaux, Campbell and their group [8], presented an ultrasonographic concrete suspicious diagnosis, based on first observation of a severe kyphoscoliosis at the lower spine and a serious anterior abdominal wall defect, in direct contact with the placenta, in a 18th week of gestation fetus (menstrual age); the couple, informed of the serious fetal conditions, decided to terminate the pregnancy: autopsy findings confirmed the severity of this malformation, with a very short Umbilical Cord (UC), O, many grave organ defects, absence of diaphragm central part and pulmonary hypoplasia; they then studied the abortion findings and the X-ray examination of the fetus at delivery , to acquire specific pathologies details.

Various forms were therefore classified, according also to Victoria et al [9] and Revels et al [10]: they can be summarized as follow: Gastroschisis (G) and O which are the most frequent ventral AWD (VAWD/AWDs), followed by Bladder Exstrophy (BE) and Ectopia Cordis (EC) [14], causing varying degrees of severe pathological entities, mainly represented by the phenotype expression of the umbilical hernias (UE) evidences. Furthermore, Pentalogy of Cantrell (PCO) [5], Prune Belly Syndrome (PBS), Limb Body Wall Complex (LBWC) (Figure 6) [20], Body Stalk Anomaly (BSA) (Figure 5), Amniotic Band Syndrome (ABS), OIES , Cloacal Extrophy (CC), complete the constellation of the pathological panel extensively described in literature.

All the pathological findings described in the above list, had led to several studies focused on estimating the incidence and prevalence of those anomalies attributable to ADW/AWDs, in the various states, worldwide; numerous risk factors related to the development of malformations were therefore identified. Moreover, of fundamental importance, for the purpose of pregnancy management and evaluation of prognostic factors discussed during counseling, it is seen to be the identification of embryo/fetal associated anomalies; there is a wide range of adjunct malformative conditions with varying degrees of gravity as literature reports.

Many scientific papers have focused on the G and O, the two main VAWD/AWDs forms.

G, or complex G when associated to at least one bowel complications such as Artesia, stenosis and/or ischemia necrosis and perforation, has been well described as an isolated small anomaly of the abdominal wall containing bowel and situated to the right of the umbilical cord insertion site; is an intestinal herniation, without membrane cover, within the primordial umbilical ring, precisely [27]; there is no significant association with genetic syndromes; associated cardiac defects are reported to be rare.

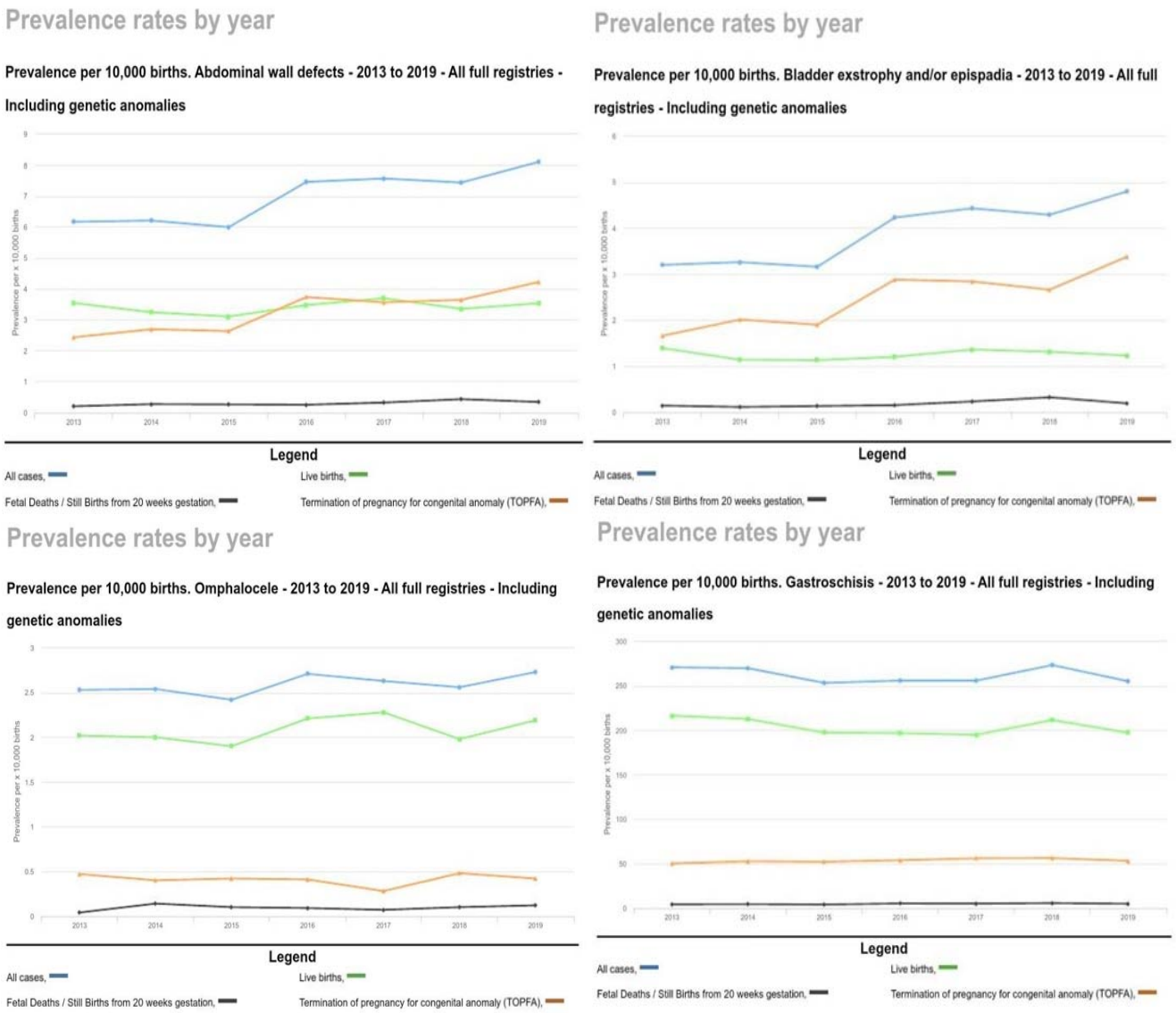

Chart 1: A=ADWs, **B=**BE, **C=**O, **D=**G; from the original [1], with the EU permission by the license CC BY 4.0 International.

Differently, O, is an entity morphogenetic ally different from G, found at the midline of the base of the umbilical cord insertion, also classified in central/epigastria/hypogastria, according to the location on the abdominal wall; O is covered by a three layered sac (peritoneum, Wharton's jelly, amnion) may contain liver, and may associated with chromosomal abnormalities, usually trisomies 21, 18 and 13 in 30-40% of cases [22], depending on maternal and gestational age at the time of diagnosis [16]; moreover, cardiac and central nervous system malformations may be seen in 14–47% and 3-33%, respectively, of affected fetuses [22] that an increased association probably by the errors during the incorporation of the heart and pericardium into the cest by the lateral folds [10].

Regards aneuploidies, already Nicolaides and his group [11], from 1983 to 1991, made the blood karyotyping in 235 fetuses affected by VAWD/AWDs and gastro-intestinal tract defects: the karyotype was abnormal in 42 (36%) of the 116 fetuses with O, in none of the 26 with G.

Later, Rankin et al studied 296 VAWD/AWDs fetuses in the 11-year period 1986–1996, calculated a G and O prevalences of 2.98 and 2.11 in 10. 000 births, respectively; almost two-thirds of O were associated with other anomalies, and half of these had also a chromosomal anomaly; moreover, they observed sensitivity of PM by US of fetal abnormality in isolated G and O in 75% and 64.5% respectively; in a range from 61.8 to 64.5% the VAWD/AWDs was correctly diagnosed [12].

About heart diseases, Gibbin et al [13] performed a retrospective study from 1992 to 2000, following previous papers; subjects were considered to have congenital heart and or cardiovascular disease when there was the evidence of either structural defects or non-structural defects as persistent pulmonary hypertension of the newborn, pericardial effusions and significant fetal and/or neonatal dysrhythmias; VAWD/AWDs, in particular O, seemed to have an increased risk of above mentioned cardiac issues.

In 2012, it has been presents a rare abnormality in a twin pregnancy, where just one twin presenting EC and exencephaly, without prior case reported (Gibbs et al; [14]); the patient has referred for US at 16 weeks and genetic counseling; at 19 weeks demonstrated a spontaneous demise for the malformed twin and normal findings for the normal twin, until the end of a gestation: authors also reviewed a detailed embryo-fetal growth process leading to this lethal anomalies [15].

In clinical practice, once suspected and therefore diagnosed VAWD/AWDs, by Ultrasound (US) examination, considering the primary identification of UC insertion, the accurate definition of pregnancy time (less or more 12 weeks) and of the complete fetal anatomy is necessary for the management and taking care in a high risk center of reference [23].

US monitoring, weekly biweekly or monthly, thanks to advanced transvaginal and transabdominal technologies, in PM, take a central role in diagnosis and management of VAWD /AWDs, as well as magnetic resonance imaging (MRI); on the basis of the known processes of embryonic and fetal development, in systems and apparatuse, it is possible, with the US method, to examine the structure at the embryo time and later, fetus time, due to find the early signs of AWDs, within the first trimester of pregnancy, and, further on, to get more details by MRI [9], in order to acquire most of the information's regarding the size of the defects, the anatomical relationships the gradually develop with US checks necessary to evaluate the conditions of the fetus, the amniotic fluid and the placenta, both maternal and fetal side. MRI would be useful to predict associated pulmonary hypoplasia, by the measurement of the lung/torax transverse area ratio in giant O [22]. It's widely established that the study of the embryogenetic process [15,25,26,27] of the abdominal wall, allows to understand the events of failure in one of it's phases. Biometric parameters have been recently studied [17], during autopsies of fetuses affected by G and O, to look for a probable correlation between the number of herniated organs and the size of the defect.

In a recent review, both detailed 3D illustrations and images relating to the physiological development of the abdominal wall and cavity and their complex defects, have been decribed; moreover, an algorithm has been elaborated as a methodology for diagnostic procedures [10].

In PM, an algorithmic scan approach is essential for an accurate definition of the fetal growth, and also to locate the correct position of the defects in relation to the umbilical cord insertion, based on US early monitoring and close programming for the patients with the risk factors. It will be necessary to articulate further investigations towards epidemiological and prognostic factors, mainly to reduce pulmonary complications and intestinal damage of fetuses affected by VAWD/AWDs, and any associated anomalies, compatible with survival. Clinically relevant studies, underline the importance of multidisciplinary management of these fetal pathologies, in referral centers for high-risk pregnancies [23].

Following the VAWD/AWDs diagnosis and in depth counseling, supported by multidisciplinary skills, the pregnancy time and fetal conditions are of substantial relevance for the planning and delivery methods.

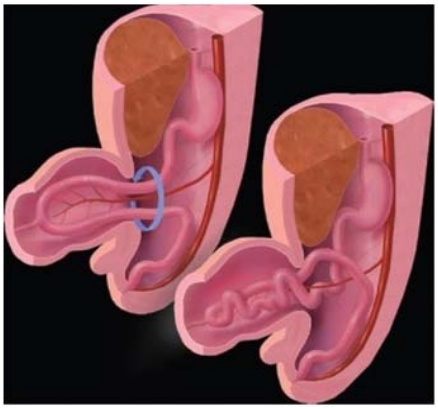

Figure 4: Normal anatomic structure and embryologic features. Schematic of physiologic bowel herniation that occurs during weeks 6-12 of development. Return of bowel into the abdominal cavity is accompanied by a 270° counterclockwise rotation.
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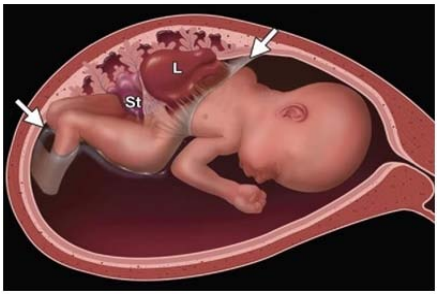

Figure 5: BSA scientific illustration; fetal organs, stomac (St) and liver (L) are situated in extra-amniotic part and are adherent to placenta.

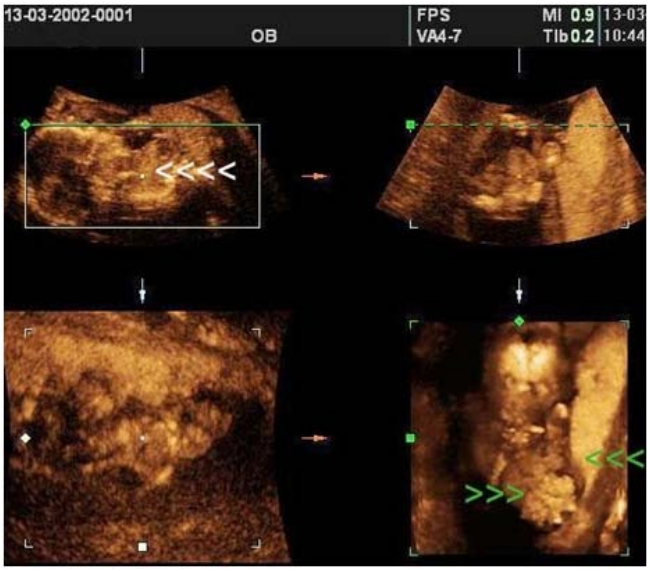

Figure 6: (Credit Solerte ML scan operator).
3D US examination shows the relationship between placenta ad fetal abdomen; an adequate 2D point reference position (white arrows) permitted to get a significative 3D construction of the relative 3D image; severe defect of the fetal abdominal wall close/attached to placenta (green arrows) [20].

The deepening of the fascinating process of embryogenesis and it's pathophysiology, Saldler [15,26,27] Moore et al [25], has allowed to understand the phases in which it's likely to alter the embryo intra (or somatic) and extra (or chorion cavity) coeloma, the embryo body folding, with relative formation of the thoracic, abdominal and pelvic cavities (Figure 2,3) and the rotation-reentry of the bowel (Figure 4). At the same time, the analysis of the etiopathogenetic processes, has seen the commitment of scientists in the description of the embryonic growth, in which the failure of the embryonic development phases probably occurs already from the third week of gestation, calculated by conceptional age : regarding the latter aspect, many specialists from various medical disciplines use to calculate the gestational age, referring to the first day of the last menstrual period, or the conceptional time, both translating the length of pregnancy in weeks and days.

It's widely established that the study of the embryo genetic process [15,25,26,27] of the abdominal wall, allows to understand the events of failure in one of its phases. Biometric parameters have been recently studied [17], during autopsies of fetuses affected by G and O, to look for a probable correlation between the number of herniated organs and the size of the defect.

We that's have performed an updated historical review, which will be exposed in a separate section, of the various stages of the normal and malformated fetuses and the prenatal therapy which could be published with a view to using the current state of the art, to acquire further insights.

Acknowledgment

Thanks for the precious collaborations to Mrs. Milena Giacometti of the civic Library of the Municipality of Varese and to Mr. Causarano Lorenzo of biological Pole BFM "Ferdinando Rossi", University of Torino.

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