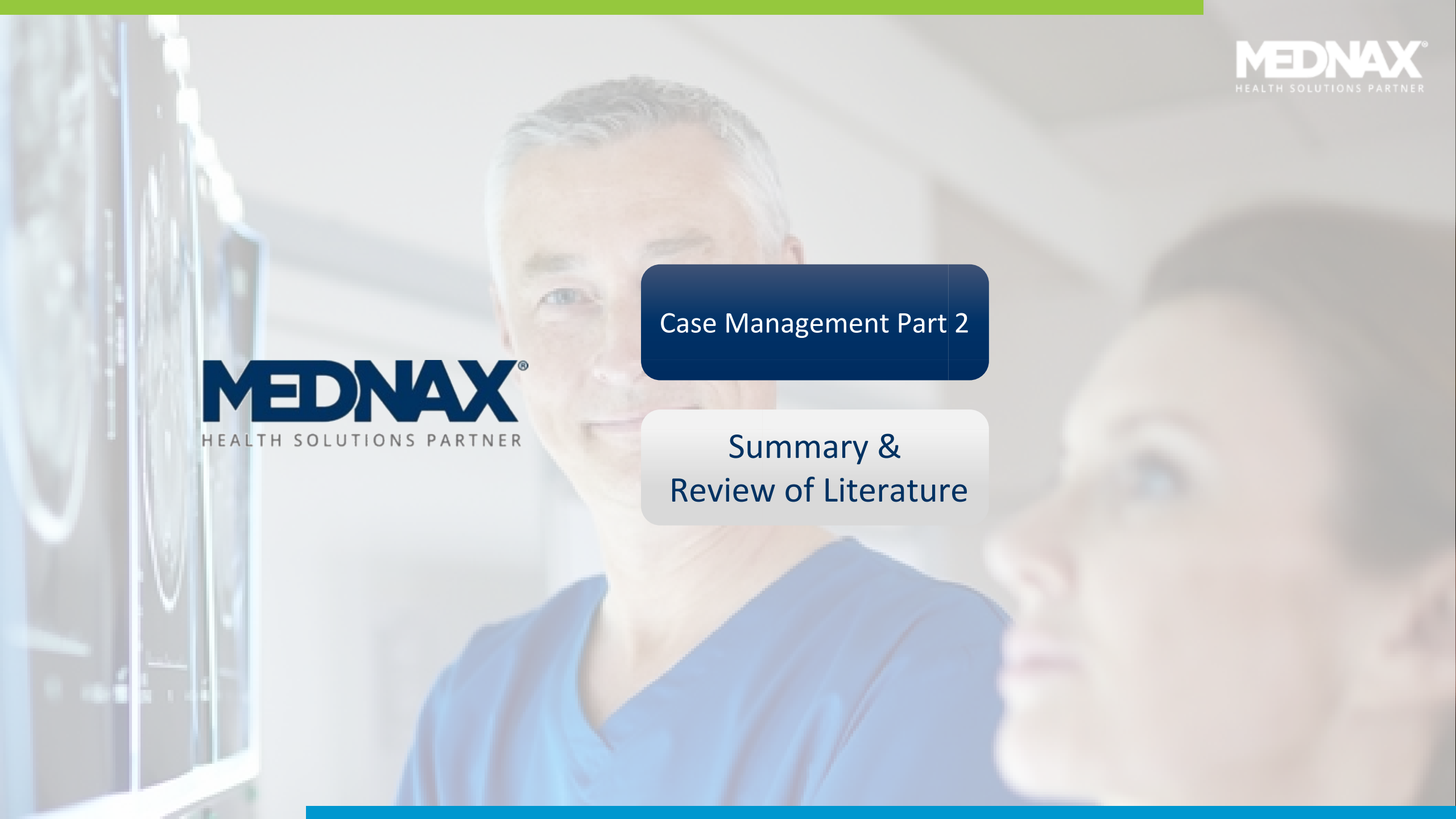


Case Management Part 2

Summary & Review of Literature

Part 2 – Case Management

Case Management:

The patient returned at 17 weeks 2 days gestation for planned selective fetocide of Twin B who carried the working diagnosis of sirenomelia with renal agenesis. Unfortunately, Twin A was found to have lagging femora and humeri (11 days), echogenic bowel, and a cardiac defect that could not be absolutely characterized. An evaluation of Twin A with amniocentesis along with the selective fetocide of Twin B was declined after extension discussion with the family; they elected termination of the entire pregnancy. This was accomplished the next day with misoprostol. Twin A was female, 111 grams, and died shortly after birth due to extreme prematurity; Twin B was of unknown gender, 84 grams, and also died shortly after birth. The classic appearance of sirenomelia with a fused lower extremity was noted. Parents declined perinatal autopsy and cytogenetics; placental pathology only confirmed the single umbilical artery for Twin B.


MEDNAX
HEALTH SOLUTIONS PARTNER
Summary &
Review of Literature

Summary & Review of Literature

Sirenomelia (mermaid syndrome) derives its name from Greek mythology wherein female “sirens” with their enchanted singing would lure Greek sailors to their small island where they would run aground on the rocky coast. Odysseus avoided death in his encounter with the sirens by having his crew (whose ears were plugged) tie him to the mast despite his insistence to be released. Sirenomelia was originally described by Rocheus in 1542 and Palfyn in 1553 (Kaygusuz et al. 2016). Such affected fetuses are often depicted with varying phenotypes, often with the lower body replaced by an aquatic tail among other depictions.

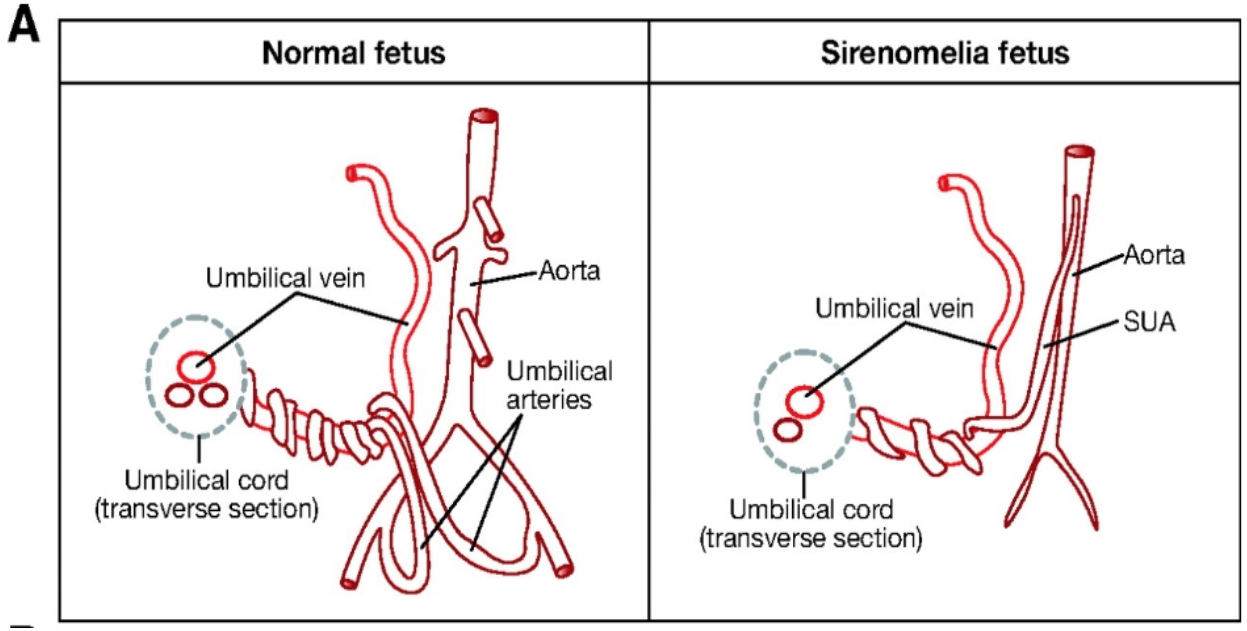
This syndrome is often included in the caudal regression sequence (CRS), representing the most severe form of that embryopathic group of anomalies. It is a rare malformation which is reported in approximately one out of every 24,000-67,000 births (Kaygusuz et al., 2016). There is much to support including sirenomelia as a severe form of CRS, including the association of other anomalies of the axial mesoderm. These would include cranial, esophageal, and cardiac defects, as well as anencephaly and alobar holoprosencephaly which have been reported in association with sirenomelia (Khoury, 2016). Interference with the formation of the notochord in the third gestational week can result in the maldevelopment of caudal structures as well as neural tube defects. At one end of this spectrum is simple coccygeal agenesis, progressing all the way to sirenomelia, with failure of lateralization and deficiency of the caudal eminence.

Summary & Review of Literature

The alternative pathophysiologic mechanism proposed for sirenomelia is vascular in etiology. The persistent vitelline artery directly from the descending aorta above the yet to develop renal arteries “steals” blood typically directed to the caudal end of the embryo and directs it to the placenta as the continuing single umbilical artery. The severe hypoperfusion of the caudal end of the embryo results in the severely malformed phenotype. Twickler and colleagues pointed out early on the distinct differences between CRS and sirenomelia, with the latter always having a single umbilical artery, single or fused lower limbs, renal agenesis, absent anus, and oligohydramnios (Twickler, Budorick, Pretorius, Grafe & Currarino, 1993).

Summary & Review of Literature

Vascular pattern in a normal versus sirenomelia fetus. (A) Schematic drawing of the fetal umbilical cord vasculature in a normal and a sirenomelia fetus. Note the abnormally high origin of the SUA in sirenomelia and the hypoplasia of the aorta caudal to its origin.



Summary & Review of Literature

As many as 22% of CRS infants are born to diabetic mothers (Boulas, 2009), as opposed to only 2% of sirenomelic infants (Twickler et al., 1993). Most authors believe that even at 2%, this association argues in favor of sirenomelia belonging to the severe end of the CRS phenotype with the presumed hyperglycemic effects on the developing embryo (possible homeobox gene expression) inducing the whole range of CRS phenotypes.

Sonographic clues relate specifically to the unique findings in sirenomelia, although by the second trimester oligohydramnios limits visualization. The single umbilical artery and renal agenesis (with absent or hypoplastic renal arteries) in a setting of marked oligohydramnios should put sirenomelia at the top of the differential diagnosis list irrespective of what can be delineated about the status of the lower extremities. The exact atretic extent of the lower extremities have prompted a classification of sirenomelia from I through VII, of dubious clinical value (Fig 1). The cystic area found in the caudal end of our fetal patient could not be confirmed after birth as no autopsy was permitted; it likely represented some colonic obstruction ending blindly in the cloacal atretic area.

Figure 1

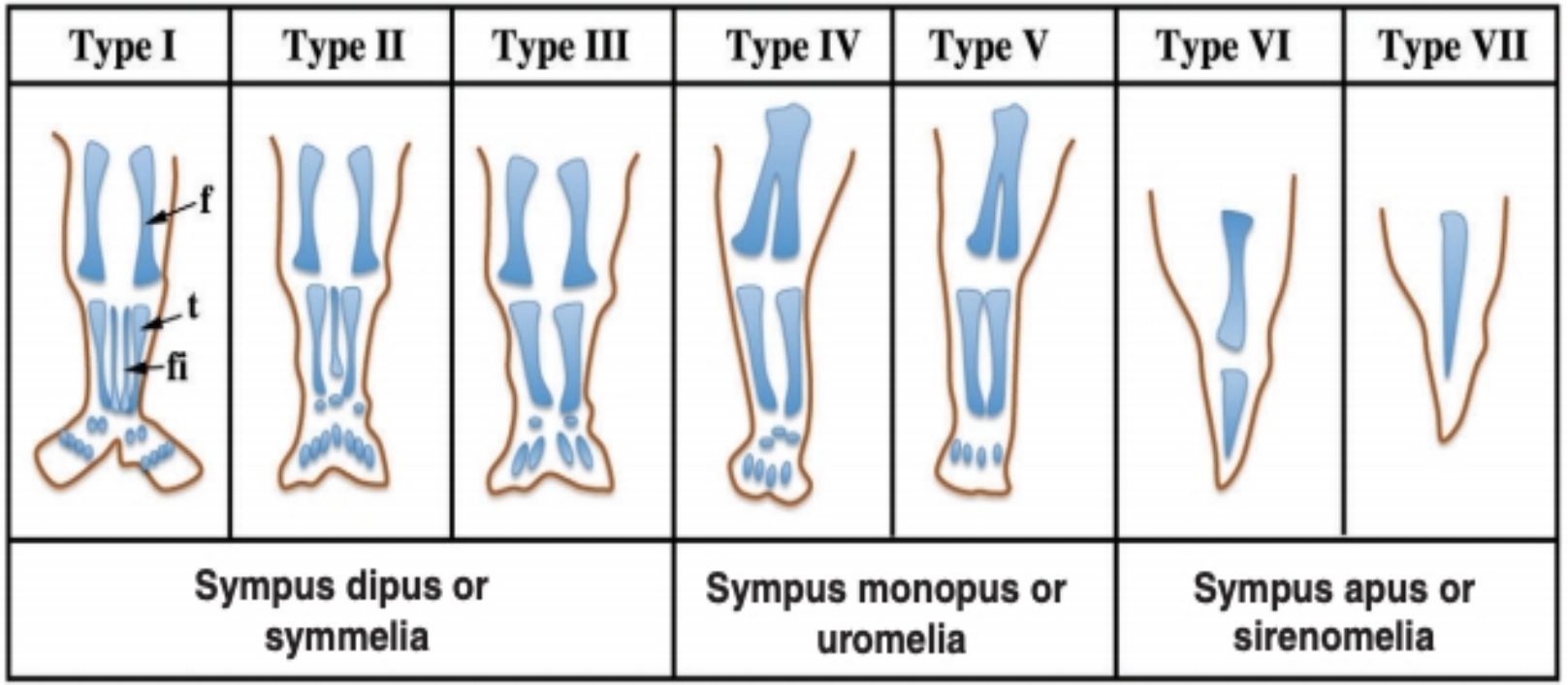


Fig. 1. Classification of sirenomelia according to Stocker and Heifetz.
(Perspect Pediatr Path 10:7-50, 1987)