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Limb Body Wall Complex and Sirenomelia in Twins

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Abstract. The coexistence of Limb body wall complex (LBWC) and Sirenomelia sequence (SS) is present in only two studies in the literature on the topic. Tang et al (1991) and Martinez-Frias et al (1992) examined single fetuses and pointed out that the association of these two phenomena is extremely rare. The present study further contributes to the investigation and description of this particular association in a pair of twins. Both twins presented abdominal defects, urogenital anomalies and persistence of the primitive cloaca, but did not show craniofacial (CF) defects or amniotic band related defects. One twin had one leg only. In a previous study on LBWC we identified two morphologically and pathogenetically different phenotypes which, among other characteristics, are identifiable by the presence or absence of CF defects. (Russo et al. 1993; Russo et al. in press). Our twin pair belongs to the LBWC group without CF defects. This group, like SS cases, is usually characterized by the presence of lower limb anomalies. Aside from this, they also share a similar pattern of anomalies involving the urogenital tract and the cloaca. Analysis of the anomalies present in the twin pair leads us to suggest that the Sirenomelia pattern might be at the extreme end in a spectrum of lower limb defects within the LBWC group that does not present CF defects. In brief, we suggest that the coexistence of this LBWC type and SS is not related to the occurrence of two different defects but is the consequence of a common maldevelopmental process occurring during blastogenesis.

(Poster presentation)

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