Congenital Malformation Syndromes

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6.1 Introduction Congenital malformations are defects of the morphogenesis of organs or body regions identified during intrauterine development or at birth. They may be isolated and single, or multiple. Patients with multiple malformations can be classified as having syndromes, sequences, associations, or dysplasias. Syndromes are conditions where all the structural defects arise from a single etiological factor, which may be genetic or environmental. Malformation sequence, in which an intrinsic malformation exists in the embryo, resulting in certain other abnormalities (e.g., radial dysplasia). Deformation sequence, in which no intrinsic defect is found in the embryo; rather, an abnormal external mechanical or structural force results in secondary distortion or deformation (e.g., constriction bands). Although at least 112 recognized syndromes are described with hand anomalies as a part of their expression, this represents only 5% of congenital hand anomalies. [1]. The International Classification of Diseases (ICD) World Health Organization's classification used worldwide as the standard diagnostic tool for epidemiology, health management and clinical purposes. This includes the analysis of the general health situation of population groups. It is used to monitor the incidence and prevalence of diseases and other health problems. Within this classification "congenital malformations, deformations and chromosomal abnormalities" are (Q00-Q99) but excludes "inborn