Introduction

Intrinsic errors of skeletal development are individually rare but of clinical importance because of their overall frequency and their impact on patients' lives. Conventionally they are divided into *malformations* – defects of single bone – and *dysplasias* – systemic defects of chondro-osseous tissue. Depending on the type of surveillance system, limb reduction defects, one major category of skeletal malformations, are recognized in 3.1–6.9 of 10,000 newborns (Eurocat 2002; Makhoul et al. 2003; McGuirk et al. 2001; Stoll et al. 2000). Due to spontaneous or induced fetal loss, the prevalence in fetuses is higher, up to 15.7 of 10,000 (A. Queisser 2003, personal communication). The overall prevalence of neonatally manifested skeletal dysplasias is about 2 out of 10,000, half of them lethal (Andersen 1989; Gobben et al. 1990; Gonnor et al. 1985; Rasmussen et al. 1996).

As sonography has become a routine component of prenatal care, many of these disorders are diagnosed prenatally confronting family and physician with the question of elective termination of pregnancy. Ideally, this question is discussed on the basis of a specific diagnosis. However, such a diagnosis is difficult to achieve by fetal sonography. Even under optimal conditions it is missed in at least 35% of cases (Schramm et al. 2009; Parilla 2003; Stoll et al. 2000) meaning that many abortions are performed on the basis of diagnostic suspicion.

Postnatally, the prenatal diagnosis has to be verified. To do this, fetal radiography becomes important. It is an effective, simple and economic way to establish a diagnosis or to narrow the number of diagnostic possibilities sufficiently to direct pathological, biochemical or molecular studies in their quest for a specific diagnosis. A specific diagnosis is required for various reasons. It permits sonographic quality control. It provides the clinical basis for research. More importantly, it is required for proper parental counseling. Parents who have gone through the termination of a pregnancy have a right to know all the available facts as to possibilities of recurrence.

This book has been written to assist in fetal postnatal radiological diagnosis. It is divided into three chapters:

1. *Development of the normal fetal* skeleton between gestational weeks 10 and 24. The chapter presents age-dependent standards with which to compare diagnostic films.
2. *Radiological differential diagnosis* of conspicuous defects of single bones such as pre- or postaxial limb deficiencies or vertebral segmentation defects. These malformations occur alone or combined in numerous conditions which are tabulated to assist in differential diagnosis. Starting from a given specific abnormality, malformation patterns can be recognized leading to a specific diagnosis or to a narrowing of the number of diagnostic possibilities.
3. *Fetal osteochondrodyspiasias.* Skeletal diseases caused by factors that continue to express themselves after the earliest stages of fetal development result in osteochondrodysplasias, systemic alterations of form, structure, and maturation of bone. Recognition of the expression pattern allows for a specific diagnosis with its prognostic, genetic, and often molecular information. In this chapter are found illustrations of the salient radiological manifestations of the most common lethal and nonlethal osteochondrodysplasias manifesting in fetal life.

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