Objectives  The aim of this study is to represent the distribution of disorders resulting from neural tube defects (NTDs).

Materials and Methods  This study was conducted on 220 prenatally diagnosed cases with NTDs. Fetuses were evaluated by physical examination, anthropometric measurements, X-rays, and photographs after termination of pregnancy. Chromosome analysis and autopsy were performed for 37 fetuses (16.8%) with additional malformations.

Results  In 29 out of 37 fetuses (78.4%), additional malformations were detected by prenatal ultrasonography, whereas in eight cases postmortem evaluation produced additional findings that were not detected prenatally. Fourteen of 37 (37.8%) and 65 of 220 (29.5%) fetuses had clubfoot, which was mostly secondary to NTDs. There was no difference in sex distribution between isolated NTDs and the group with additional abnormalities and among the groups anencephaly and anencephaly + anomaly, encephalocele and encephalocele + anomaly, spina bifida and spina bifida + anomaly. There was only one case, a female fetus, with iniencephaly in this group. Anencephaly was more frequent in cases with isolated NTDs (48.1%) than in those with additional anomalies (27%). There was no difference for other groups of NTDs. The most frequent disorder was vertebral segmentation defects, which were detected in 11 out of 37 cases (29.7%).

Conclusions  Evaluation of associated malformations and confirmation of ultrasound findings can be performed by postmortem examination and simple X-ray studies for exact diagnosis, which strongly affects decisions on further pregnancies as well as genetic counseling. This method is straightforward, inexpensive and effective. Copyright 2006 John Wiley & Sons, Ltd.

KEY WORDS: neural tube defect; prenatal diagnosis; postmortem evaluation