20th World Congress in Fetal Medicine

Comparison of sonographic findings and postnatal phenotype in upper limb anomalies

Arduç A, van Dijk JBS, ten Cate FJ, van Doesburg MHM, Linskens IH, van Leeuwen E, van Maarle MC, Pajkrt E
Amsterdam UMC, Amsterdam, Netherlands

Objective

Evaluation of the current performance of prenatal sonography for different upper limb anomalies (reduction defect, polydactyly, syndactyly, and oligodactyly) by comparison of sonographic findings and postnatal phenotypes.

Methods

This is a retrospective cohort study of pregnant women undergoing sonography at the Fetal Medicine Unit of the Amsterdam University Medical Center (UMC), a tertiary referral center. Data from January 2007-December 2021 are evaluated. Ultrasound data on prenatally suspected cases of a upper limb anomaly, namely, a reduction defect, polydactyly, syndactyly, and oligodactyly, were included (prenatal group). Postnatal outcomes of the newborns were compared with the initial prenatal diagnosis. Furthermore, cases with a upper limb anomaly that were not identified with prenatal ultrasound were evaluated as a separate group (postnatal database). These children consulted the 'Congenital Hand Team', a multidisciplinary team of healthcare professionals that is specialized in congenital anomalies of the upper limb. Findings during physical examination and possible genetic associations were evaluated for each anomaly. Finally, a time trend analysis of a 3-years moving average of the ratio true positives (TP) /false negatives (FN) was calculated per year.

Results

An upper limb anomaly was sonographically suspected in 225 cases: 80 reduction defects, 124 polydactylies, 16 syndactylies, and five oligodactylies. Prenatal diagnosis was confirmed after birth in 72% of the cases: 91% for reduction defect, 60% for polydactyly, 80% for syndactyly, and 60% for oligodactyly. In total, 132 of the 225 (59%) cases were sonographically non-isolated and 93 (41%) were isolated. Pre- or postnatal genetic tests were performed in 133 (59%) of the 225 pregnancies. Genetic or chromosomal abnormalities were found in 81 pregnancies: aneuploidy in 49 (37%), other chromosomal disorders in 12 (9%), and monogenetic disorders in 20 (15%). Additionally, a postnatal group of 389 children with upper limb anomalies that were not identified on prenatal ultrasound was evaluated separately. All children consulted the 'Congenital hand team'. Of the 389 children, 102 (26 %) had non-isolated and 287 isolated (74%) defects on physical examination. Postnatal genetic tests were performed in 81 (20%) of the 389 cases. Genetic or chromosomal abnormalities were found in 20 children: aneuploidy in three (4%) pregnancies, other chromosomal disorders in 12 pregnancies (9%), and genetic disorders in five (6%) pregnancies. We estimated that a reduction defect, polydactyly, syndactyly, or oligodactyly was suspected in 0,06% of all pregnancies in the entire North-Holland region between January 2007 and December 2021. In this population, polydactyly had the highest estimated prevalence of 8 per 10.000 pregnancies, followed by 4 in 10.000 for reduction defects, 2 in 10.000 for syndactyly, and 0.15 in 10.000 for oligodactyly. During the study period 46% of all limb anomalies were sonographically recognized prenatally: 49% of the reduction defects, 31% of the cases with polydactyly, 19% with syndactyly, and 83% with oligodactyly. The time-trend analysis of the 3-years moving average of the TP/FN ratio showed no statistically significant differences for any of the anomalies (all $p>0.05$) during the study period.

Conclusion

Our findings suggest that the prenatal identification of upper limb anomalies has not changed over the last decades. The majority of the upper limbs anomalies were not recognized in the prenatal period. However, non-isolated defects were seen more often prenatally, as compared to the isolated cases which were predominantly discovered postnatally. This suggests that upper limb anomalies are better identified in the case other structural anomalies are present and the fetus undergoes detailed sonography. Due to the high incidence of chromosomal and genetic disorders in the prenatal period, a genetic consultation should always be considered as a standard workup to define the underlying pathology.