**Institutionen för klinisk forskning och undervisning,
Södersjukhuset, enheten för handkirurgi**.

Congenital Upper Limb Anomalies – Studies of Epidemiology and Hand Function

AKADEMISK AVHANDLING

som för avläggande av medicine doktorsexamen vid Karolinska Institutet offentligen försvaras i Södersjukhusets aula, plan 6, Sjukhusbacken 10, Stockholm

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ABSTRACT

Objectives: This thesis has three interrelated aims:
(1) To describe the epidemiology of congenital upper limb anomalies (CULA) in Stockholm County, Sweden, in order to augment the few existing population studies of CULA (paper I);
(2) To measure the incidence of different categories of CULA while using and evaluating a recently proposed new classification scheme (*Oberg, Manske and Tonkin (OMT) Classification*) based on more current knowledge of limb development than the previously used *International Federation of Societies for Surgery of the Hand (IFSSH) Classification* is based on (paper II); and
(3) To investigate the relationship between measurements of *body function and structure* with both *activity* and *participation* in children and adults with radial longitudinal deficiency (RLD) by using the *International Classification of Functioning and Health (ICF)* framework, in order to shed light on what aspects of physical limb function and structure actually affect individuals' daily life activity (papers III and IV).

Methods: 562 children born with a CULA were identified through registry studies. Incidence and relative frequency of different types of anomalies were calculated. Distribution of gender, affected side, associated non-hand anomalies and occurrence among relatives were investigated (paper I and II). In twenty children (paper III) and 20 adults (paper IV) with RLD, *Body function and structure* was evaluated by measures of range of motion, grip strength, key pinch, sensibility and radiographic parameters. *Activity* was evaluated by Box and Blocks test, Assisting Hand Assessment (AHA) and Sollerman Hand Function test and p*articipation* by Children Hand-use Experience Questionnaire (CHEQ), Quick-DASH and SF-12. Statistical correlations between assessments of *body function and structure*, *activity* and *participation* were examined.

Results: The incidence of CULA in Stockholm, Sweden, 1997 to 2007, was 21.5 per 10,000 live births (paper I). All CULA could be classified using the OMT classification. The largest main category was *Malformations* (429 cases), followed by *Deformations* (124 cases), *Dysplasias* (10 cases) and *Syndromes* (14 cases) (paper II).
In children with RLD (paper III), significant relationships were found between measurements of *activity* and range of motion of digits as well as between measurements of *participation* and range of motion of wrist. In adults with RLD (paper IV), significant relationships were found between measurements of *activity* and grip strength, key pinch and range of motion of elbow and digits. In adults, measurements of *participation* showed significant relationships with grip strength, forearm length and range of motion of elbow and digits. However, radiographic measurements of radial wrist deviation did not show a significant relationship with measurements of *activity* or *participation* in children or in adults with radial longitudinal deficiency.

Conclusions: The incidence of CULA in one Swedish region confirms the findings in the only previous comparable total population study. The OMT classification proved useful and accurate and with further refinements can replace the IFSSH classification. In children and adults with RLD, grip strength, key pinch, forearm length and elbow and digital motion seem to be more important for the individual´s levels of activity and participation than the radial angulation of the wrist. The current treatment principle of surgical correction of the angulated wrist could therefore be questioned.

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