Shoulder dysplasia in longitudinal deficiencies of the upper extremity

1. Summary:

- *Study purpose:* Clinical and radiological assessment of the shoulder in children with congenital radial and ulnar longitudinal deficiencies
- *Design:* Prospective study with one-time clinico-radiological assessment
- *Study duration/follow-up:* one time assessment, no follow-up needed
- *Main outcome parameters:*
    i. Radiologic shoulder evaluation (e.g. dysplasia of the glenoid, acromion, humeral head) acc. to Waters classification; arm length
    ii. Shoulder range of motion and stability
- *Secondary outcome parameters:*
    iii. Pediatric outcomes data collection instruments (PODCI)
    iv. Pediatric/Adolescent shoulder survey (PASS)
    v. Pain score (VAS, visual analogue scale)
- *Radiologic assessment:* X-ray of the affected and contralateral shoulder and upper extremity (a.p.+ Bernageau + Humerus).
- *Expected study participants*: approx. 20.

2. Authors:

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3. Introduction:

The congenital radial longitudinal deficiency (RLD), though very rare, is the more common longitudinal reduction defect of the upper extremity, with an incidence of about 2,4:10.000.[1] In this disorder, the radius is malformed, shortened and bowed; in certain cases, no radius is evident at all. It is bilateral in 50% of cases and affects males more commonly than females (1,5:1). It is commonly associated with other syndromes such as TAR, Holt-Oram-syndrome or Fanconi anaemia. Main clinical features include radial wrist deviation, a foreshortened forearm and malformations (hypoplasia) of the thumb. Besides the affected bony structure, soft tissue is malformed as well, mainly involving tight radial-sided muscles and aberrant tendons/muscles. The ulnar dyplasia is 10 times less common and usually affects only the distal part of the ulna. The ring and small finger may not be formed at all, and moreover, the thumb may be affected as well. In the literature, just few anecdotic cases of associated growth defects in the shoulder were mentioned. However, the only two reports published to date lack a clear and concise description and discussion of the potentially concomitant shoulder deformity.[2,3]

4. Purpose and Clinical Relevance:

This study aims to systematically assess the shoulder of children with these 2 entities with regards to malformation of the anatomic structures, functional deficits and patient-reported outcome scores. To shed more light on this rare feature, precise measurements of the humerus and forearm will reveal the amount of expected length deficiencies.

The clinical relevance of this work is to highlight 1) any growth- and length-related abnormalities of the upper extremities in these disorders, which might affect patients´ future and functional abilities; 2) to provide evidence of potentially clinically relevant shoulder dysplasia which might lead to functional impairment (e.g. instability). Due to the very scarce literature on this topic we will expect relevant further insight into this area.

5. Study design:

This study will be initially a monocentric prospective evaluation of patients with the afore-mentioned disorder. Data will be compared with the contralateral healthy side (if not as well affected). The final patient count depends on the amount of patients documented in the hospital records of the Pediatric orthopedic department in the Orthopedic Hospital Speising, matching the requested inclusion criteria and being available for this study. Approximately 20 patients will be expected to meet all criteria and be available for this study. Future collaborations with other centers to increase patient count are likely and expected.

Expected Duration: 12 months

Minimum age for inclusion: 5 years

Ethics committee (IRB): approved by the Ethics committee of the Vinzenzgruppe (AUT) on 28.6.2021 (EK 19/2021)

Material and methods: The shoulder joint will be assessed in the 1) a.p. view, 2) Bernageau view (for glenoid dysplasia), 3) Humerus a.p. with calibration. Function will be documented using ROM, stability, and the following outcome scores: PODCI, PASS, VAS.

Evaluation: The study parameters will be documented in the appropriate Case Report Form (CRF). After completion of data collection, the parameters will be compared and analyzed with the contralateral side.

6. Patient selection:

Participants:
All participants must fulfill the inclusion criteria and must not fulfill any exclusion criterion. All cases will be exclusively be recruited from our hospital using charts from 1/2000 to 12/2020. Patients will be contacted in the following ways:

1. First written invitation (siehe Amendment1)
2. Second written invitation after 6 weeks
3. Telephone invitation

Inclusion criteria:
Congenital radial longitudinal deficiency or congenital ulnar longitudinal deficiency.

Exclusion criteria:

Age < 5 years; pregnancy; previous shoulder trauma; habitual shoulder instability

7. Patient information:

Patients and their parents/caregivers will be informed about this study, any potential risks and benefits prior to inclusion into this research project. Females with potential pregnancy (e.g. >14 yrs) will be requested a pregnancy test. They will be informed that a third person might have data access for needs of statistic evaluation and/or data work. A written informed consent is to be signed by the patient (if over 14yrs) and the parents.

8. Quality control:

Ethics: This study will follow the declaration of Helsinki (2004). An appropriate IRB approval is already granted as mentioned above (Dekl. Von Helsinki B.13).

Informed consent: Acc. to the declaration von Helsinki (B.20-26) the patient will be informed by the PI about the study purpose, duration, and data protection efforts (B.21). The PI will assure that the patient has full understanding of everything discussed, and a written confirmation will be obtained.

Side effects: They are not expected for this study; however, any such effects will be documented on the CRF.

Documentation: To assure patient privacy, the CRF will not include names or addresses and will be anonymized.

Data analysis: Data will be analyzed using a descriptive analysis, including patient characteristics (demographics, glenoid dysplasia, scores…). The results will be highlighted using tables, box-plots and histograms. Nominal and ordinal variables will be shown with percentage. Continuous variables will be presented by means, Minimum-Maximum, median and SD. For quantitative variables (e.g. age, BMI) mean, SD, quartiles, Minimum and Maximum will be calculated. Depending on the data, further statistical comparisons and/or correlations might be added.

9. References:

1. Abzug JM, Kozin SH. Radial longitudinal deficiency. J Hand Surg Am. 2014 Jun;39(6):1180-2. doi: 10.1016/j.jhsa.2014.03.036. Epub 2014 May 5. PMID: 24810938.
2. Goldfarb CA, Manske PR, Busa R, Mills J, Carter P, Ezaki M. Upper-extremity phocomelia reexamined: a longitudinal dysplasia. J Bone Joint Surg Am. 2005 Dec;87(12):2639-2648. doi: 10.2106/JBJS.D.02011. PMID: 16322613.
3. Tytherleigh-Strong G, Hooper G. The classification of phocomelia. J Hand Surg Br. 2003 Jun;28(3):215-7. doi: 10.1016/s0266-7681(02)00392-3. PMID: 12809650.