Thalidomide Embryopathy

Orthopaedic Aspects, Degenerative Changes and Quality of Life at Age 45

Akademisk avhandling

Som för avläggande av medicine doktorsexamen vid Sahlgrenska akademin, Göteborgs universitet kommer att offentligen försvaras i Hörsal Arvid Carlsson, Academicum, Medicinaregatan 3, Göteborg, fredag den 9:e dec 2016, kl. 13.00

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Avhandlingen baseras på följande delarbeten

- I. Ghassemi Jahani S. A., Danielson B., Karlsson J., Danielsson A.J. Long-term follow-up of thalidomide embryopathy; malformations and development of osteoarthritis in the lower extremities and evaluation of upper extremity function. *J Child Orthop. 2014 Oct;8(5):423-33*
- II. Ghassemi Jahani S. A., Danielsson A.J., Ab-Fawaz R., Hebelka H., Danielson B., Brisby H. Degenerative changes in the cervical spine are more common in middle-aged individuals with thalidomide embryopathy than in healthy controls. *Plos One. Published: May 13, 2016*
- III. Ghassemi Jahani S.A., Karlsson J., Brisby H., Danielsson A.J.
 Health related quality of life and function in middle-aged individuals with thalidomide embryopathy. *Accepted. J Child Orthop.*
- IV. Ghassemi Jahani S.A., Danielsson A.J., Karlsson J., Brisby H. Middle-aged individuals with thalidomide embryopathy have undergone few surgical limb procedures and demonstrate a high degree of physical independence. *Submitted*

SAHLGRENSKA AKADEMIN INSTITUTIONEN FÖR KLINISKA VETENSKAPER



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Abstract

In the late 1950s and early 1960s, a sudden peak was seen among children with different, severe malformations, caused by the intake of the drug thalidomide during pregnancy. Among other featured malformations, so-called thalidomide embryopathy (TE), the most obvious were seen in the extremities. In this thesis, orthopaedic problems, physical function and quality of life in a cohort of survivors with TE in Sweden, around 45 years of age, were investigated. The study group of 31 individuals with TE underwent a clinical examination, computed tomography (CT) of the pelvis and lower limbs (Study I) and MRI of the cervical spine (Study II) and answered several questionnaires (Studies I, III, IV). For evaluations of function, the "Disability in Shoulder Arm and Hand" (DASH) and "Rheumatoid Arthritis Outcome Score" (RAOS) were used (Study I). Health-related quality of life was evaluated by the Short Form-36 (SF-36) and Euro Qol-5 Dimensions (EQ-5D) (Study III) and the level of independence was evaluated by a modified General Function Score (GFS) and WHO questionnaire (Study IV). The individuals with TE had a number of different malformations in the extremities. Five individuals were identified as having a proximal femoral focal deficiency (PFFD). The whole group had a high prevalence of moderate osteoarthritis in their hip and knee joints and reported greater disability in terms of upper limb function compared with the general population. The individuals with PFFD reported significantly poorer lower extremity function than the rest of the group (Study I). Individuals with TE demonstrated a high frequency of degenerative changes in the cervical spine compared with a control group (Study II). Individuals with TE reported a reduction in physical quality of life measured by both the SF-36, i.e. the Physical Composite Summary Score (PCS), and EQ-5D. Their mental quality of life was not affected, when measured by either the SF-36 Mental Composite Summary Score (MCS) or the EQ-5D. A relationship between low PCS and the number of extremities involved, as well as with the DASH score and the RAOS's pain subscore (Study III), was observed. Individuals with TE generally demonstrated a high level of independence, as the majority were employed and participated in physical activities (Study IV). In conclusion, middle-aged individuals with TE demonstrated an increased risk of degenerative changes in both larger joints and the cervical spine. Individuals with PFFD were most affected in terms of physical function, daily activities and quality of life. Despite a decrease in physical function in the overall group, their mental quality of life was not affected.

Keywords: Thalidomide, Thalidomide Embryopathy, Proximal Femoral Focal Deficiency, Osteoarthritis, Quality of Life, Malformation.

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