

Summary

Introduction: Congenital short femur, or proximal femoral focal deficiency (PFFD), is a rare complex deformity of the lower extremity with femoral dominance. The clinical findings cover wide range of variety, from femoral absence till inconspicuous shortening of the femur.

Aim of the study: 1. Molecular analysis of pseudoarthrosis tissue in congenital short femur with focusing on osteogenic and angiogenic gene expression in comparison with physiological bone. The differences in gene expression were expected. 2. Retrospective analysis of femoral healing after prolongation calculating the severity of affection, age, distance of elongation and complication. The extended healing according to severity type and age was expected compared to control group.

Material and methods: The RNA from piece of one was isolated and transcription profile of possible 113 genes of osteogenesis and angiogenesis was detected by biochip technology (SuperArray Bioscience Corporation). 10 samples analyses were performed (7 of PFFD, 3 controls). The data of 57 PFFD patients indicated for elongation of the femur with the types Pappas III, IV, VII, VIII and IX and 12 patients in control group were evaluated retrospectively and statistically by GLS method.

Results: The expected differences in gene expression in PFFD tissue compared with control were confirmed, especially in angiogenesis. In PFFD tissue were some genes over-expressed (calcitonin receptor gene, collagen XII, collagen II, IX, FGFR2, fibronectin, integrin), some of them under-expressed (gene for annexin A5, collagen XVIII, collagen I, cathepsin K, FGFR1, FGFR3, IGF2, VEGFB). The relationship between bone healing and severity of affection as so as age of the patient was not confirmed at clinical part of the project. Statistically significant difference in healing index (HI) during the second repetitive prolongation was noted. The significance of HI difference was found in the interval between repetitions of femur prolongation, when under 7 years between procedures the HI grows.

Conclusion: The hypothesis of the different gene expression was confirmed, but not so strong for generalize. The hypothesis of the relationship between severity of femoral defect and bone healing (HI) was not confirmed. The significant difference in bone healing after elongation in second repetition and interval between procedures less than 7 years was identified.

Key words: Proximal femoral focal deficiency, Gene Expression, Angiogenesis, Osteogenesis, Bone healing, Prolongation of the femur