

Proximal focal femoral deficiency

Also known as: **congenital hip joint of the femur, pseudoarthrosis femoris congenita, dysgenesis of the proximal femur, aplasia femoris congenita, congenitally short femur, coxa vara congenita** (smaller degree of aplasia femoris congenita).

Proximal focal femoral deficiency (PFFD) represents a diverse and severe clinical entity. It is a non-hereditary, rare disorder affecting the pelvis and the proximal part of the femur.

The proximal part of the femur is hypoplastic or aplastic, fibula dysplasia is present in 69% . The patella is also often missing.

The disability can be unilateral or bilateral.

Etiology

The cause is teratogens acting during pregnancy (thalidomide , see Congenital developmental defects , Congenital limb defects), radiation and bacterial or viral infections.

Classification according to Pappas

(according to localization and severity, most often Pappas III or IV occur in our country):

- **I.** - congenital aplasia (absence, i.e. the femur is completely missing, aplasia of the acetabula).
- **II.** - proximal femoral and pelvic deficiency (aplasia), femur shortened by 70-90% (Aitken D).
- **III.** - proximal femoral defect, where the bony connection between the diaphysis and the femoral head is missing (hip joint formed, femoral head hypoplastic), shortening of the femur by 45-80% (Aitken C).
- **IV.** - proximal femoral defect with disorganized fibrous connection between diaphysis and femoral head, femur shortened by 40-60% (Aitken B).
- **V.** - defect of the femur in the diaphysis, rare (defect of the middle area of the femur with hypoplasia of the proximal and distal ends).
- **VI.** - distally defective femur, missing functional knee joint (distal femoral deficiency).
- **VII.** - hypoplastic femur with coxa vara and sclerosis of the diaphysis (Aitken A).
- **VIII.** - hypoplastic femur with coxa valga.
- **IX.** - proportionally hypoplastic femur.

Classification according to Aitken

(according to the radiological findings):

- **A** - femoral head present, possible varus deformities.
- **B** - femoral head present, but ossification delayed, possible varus deformities and occlusion.
- **C** - femoral head missing, severe dysplasia of the acetabulum, severe shortening of the femur.
- **D** - femoral head missing, severe dysplasia of the acetabulum, severe shortening of the femur.

Clinical picture and treatment

The tactics of treatment differ according to the extent of the disability of the femur - whether it is a simple **shunt** or a right **hip joint**. In more serious defects, the lower limb is excluded from the load-bearing function, there are defects of the knee joint, hip joint, limb shunt.

- with the aim of healing the joint, returning the load-bearing function of the limb and equalizing its length;
- used various techniques **of external fixation** for stabilization and multiplicity of distraction (Ilizarov's apparatus);
- **180° rotational osteotomy of the lower leg: we create ideal conditions for replacing the function of the knee with the ankle joint;**
- if a sufficiently long section of the femur is preserved and the knee joint is at least partially functional, a tiered prosthesis can be used.

Links

References

- SOSNA, A., P. VAVŘÍK and M. KRBEČ, et al. *Basics of orthopedics*. 1st edition. Prague: Triton, 2001. ISBN 80-7254-202-8 .
- DUNGLE, P., et al. *Orthopedics*. 1st edition. Prague: Grada Publishing, 2005. ISBN 80-247-0550-8 .

- KOUDELA, K., et al. *Orthopedics*. 1st edition. Prague: Karolinum, 2004. ISBN 80-246-0654-2 .
- WIKIPEDIA EN,. *Proximal femoral focal deficiency* [online]. ©2005. Last revision 2009, [cit. 2009]. <https://en.wikipedia.org/wiki/Proximal_femoral_focal_deficiency >.

Related articles

- Congenital limb defects
- Developmental hip dysplasia
- Pes equinovarus congenitus

External links

- PFFD.org (<http://www.pffd.org/>) (overview and patient stories)
- eMedicine (<https://emedicine.medscape.com/article/1248323-overview>) (overview)
- Proximal femoral focal deficiency (http://gait.aidi.udel.edu/res695/homepage/pd_ortho/educate/clincase/pffd.htm) (article from the Department of Pediatric Orthopedics, the Al duPont Hospital, EN)

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