



Successful timely ambulation with orthosis and physiotherapy in 2 cases of femoral hypoplasia-unusual facies syndrome

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Introduction: Femoral hypoplasia-unusual facies syndrom or femoral facial syndrome (FFS) is a rare skeletal malformation characterized by a spectrum of anomalies including femoral hypoplasia and craniofacial dysmorphism. The reported patients have no cognitive impairments¹. Etiopathogenesis remains unknown¹ but a strong association with maternal insulin-dependent diabetes mellitus has been documented².

Materials: 1 male child and 1 unrelated female child, both 2 years old, both with FFS. The male child exhibited bilateral femoral hypoplasia, leg length discrepancy, bilateral hip dysplasia, talipes equinovarus and limited ROM in the knees and elbow. Similarly, the female child presented with bilateral femoral hypoplasia, leg length discrepancy, bilateral hip dysplasia, limited ROM in both elbows and spinal curvature but also with visceral manifestations.



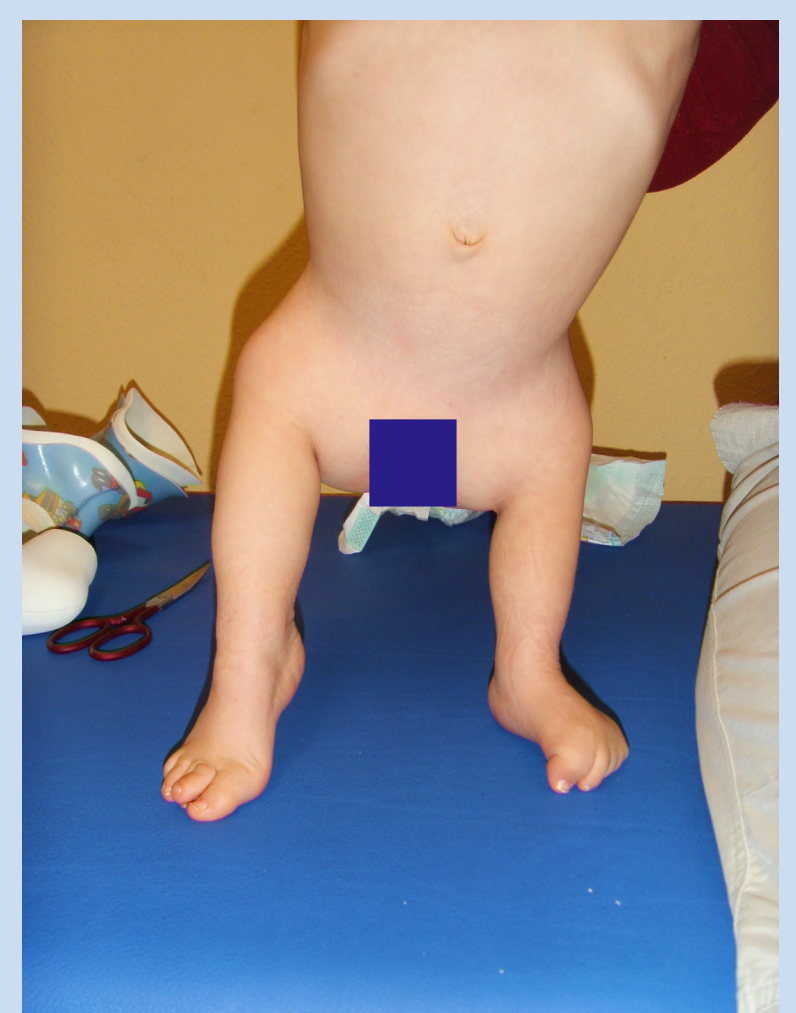
Methods: During several inpatient hospitalizations, both children were individually-fitted with orthoses and underwent extensive physical and occupational therapy. Their progress was documented.

Results: Following the treatment plan mentioned above, both children were successfully able to ambulate, timely according to their age. Further progression of contractures could be prevented.

Discussion

This syndrome encompasses femoral hypoplasia, hip dysplasia, facial dysmorphism with cleft palate, micrognathia, a short, broad nose and thin upper lip. Other manifestations involving cardiovascular, ear, nose and throat and genitourinary have been documented³. Both cases mentioned above are consistent with established clinical findings in FFS.

Many children diagnosed with FFS fail to develop a free, stable gait. This can depend on many factors, from extraskeletal complications to inadequate consultation and physiotherapy. This should be carefully monitored, as the child's healthy development and independence are influenced significantly by the development of free, stable ambulation. We are able to demonstrate that with individually-fitted orthoses and extensive physical and occupational therapy, the children mentioned above were able to ambulate successfully and timely after 3-6 weeks treatment. In order to prevent progressive contractures, children with FFS should begin therapy early, before walking is initiated.



References

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