

## Weaver syndrome associated with bilateral congenital hip and unilateral subtalar dislocation

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### Abstract

**Background:** Weaver syndrome is a congenital paediatric syndrome characterized by mental, respiratory and musculoskeletal manifestations. The coexisting deformities of the skull, the face, fingers and toes are typical. We report a case of a girl with Weaver syndrome associated with rare bilateral congenital dislocation of the hips associated with congenital hypoplastic talus and subtalar dislocation of her ankle joint.

**Case Report:** A 3-year old girl was admitted in our department with typical manifestations of Weaver syndrome, associated with congenital dislocation of bilateral hips, hypoplastic talus and subtalar dislocation of her right ankle. She was in pain while standing upright and incapable of independent walking. Both hips were treated operatively with open reduction and bilateral iliac osteotomy. Two years afterwards she had an open reduction of her talus and extraarticular arthrodesis of her subtalar joint in her right ankle. Six years postoperatively after the hip operations and four years after the ankle operation the girl is ambulant with a painless independent and unaided walking with a mild limp and full range of movements in all the operated joints.

**Conclusions:** We suggest that children with Weaver syndrome and disabling musculoskeletal deformities, particularly affecting their ability to stand up and walk should be treated early, before bone maturity, in order to achieve the best potential musculoskeletal as well as developmental outcome. Hippokratia 2010; 14 (3): 212-214

**Key words:** Weaver Syndrome, congenital dislocation, hip, ankle

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Weaver syndrome is a rare congenital paediatric syndrome characterized by mental, respiratory and musculoskeletal manifestations. It is characterized from skeletal overgrowth tendency and accelerated bone maturation. The syndrome was described first by Weaver et al in 1974<sup>1</sup> and since then only 33 cases have been described in the up to date literature.

We report a case of a girl with Weaver syndrome associated with rare bilateral congenital dislocation of the hips associated with congenital hypoplastic talus and subtalar dislocation of her ankle joint. To our knowledge, it is the first case reported in the literature with those particular lower limb manifestations.

### Case Report

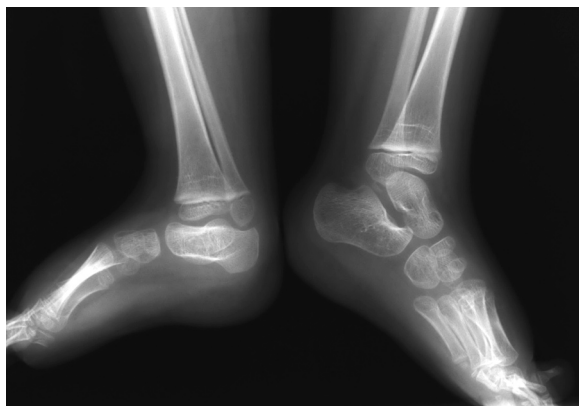
A 3 years old girl was referred to us by the paediatrician with probable Weaver syndrome and disabilities in walking. She was admitted in our department with the following clinical features: mental retardation, typical face appearance, respiratory problems and well as disability to stand upright and walk unaided. While trying to stand up she had pain in the right ankle, which was obviously in valgus deformity.

The child had a mild psychomotor retardation and generalized hypotonia. She had a typical face appearance with enlarged skull, broad forehead, flat occiput, wide

set eyes (hypertelorism), prominent philtrum as well as prominent ears. Her history included multiple chest infections and chronic bronchitis because of bilateral focal lung atelectasias. Her upper limbs had an overgrowth of the proximal phalanx in all the fingers, prominent finger pads, broad thumbs, clinodactyly and camptodactyly in bilateral hands. Regarding the lower limbs she had bilateral congenital dislocation of the hips (Figure 1) diagnosed directly after birth, broad distal femoral ends, hypoplastic vertical talus and congenital complete subta-



**Figure 1:** Bilateral congenital hip dislocation.



**Figure 2 & 3:** Congenital subtalar dislocation of the right foot.

lar dislocation with obvious valgus deformity of the right foot and ankle (Figure 2, 3), mild valgus deformity of her left hindfoot and bilateral overlapping small toes.

The child, Caucasian origin, was born full-term after an uneventful pregnancy with birth weight of 3.5 kg and height 54 cm. Her father had similar but milder expression of the same clinical appearance, however, he had no obvious disabling musculoskeletal disorders and he never had any problem with his walking ability.

Clinical appearance as well as chromosomal profile established the diagnosis of Weaver syndrome and based on father's clinical appearance it is claimed that our patient is an autosomal dominant inherited case.

The congenital dislocations of the hips were treated by open reduction and «Dega» pelvic innominate osteotomy. The subtalar dislocation and valgus deformity of the right hind foot was treated with extra-articular subtalar arthrodesis using the Grice Green operative technique.

The patient postoperatively, 6 years after the hip operations (Figure 4) and 4 years after the right foot operation (Figure 5), has a painless unaided walking with a mild limp and full range of motion in all the operated joints, although we were not satisfied from the clinical and x-ray outcome of the right foot because the joint remains sub-dislocated and there is a slight instability in valgus position.



**Figure 4:** The hips 6 years post-operatively.



### Discussion

Weaver syndrome was described first by Weaver et al in 1974<sup>1</sup> as a syndrome of accelerated growth and osseous maturation, unusual craniofacial appearance, hoarse and low pitched cry, and hypertonia with camptodactyly. Thumbs are broad, some fingers are permanently bent (camptodactyly) and the pads of the fingers are prominent. Toes are malformed in the form of clinodactyly, and the foot develops varus (clubfoot) or valgus (pes talipes) deformity. The craniofacial deformities include flat head, flat occiput, broad forehead, extremely wide set eyes (hypertelorism) and large ears.

Since 1974, 33 cases are reported in the literature. The first female patient reported in the literature was in 1983. Ozkan and Bekeret<sup>2</sup> in 2000 reported another female patient 2 year- old and they stated that females have usually milder expression of the syndrome, a hypothesis that is not confirmed in our case.

Weaver syndrome is clinical co-existence of some particular manifestations. Because often the clinical appearance is less typical with generally milder clinical expression the syndrome is often called as Weaver – like or other times Weaver – Smith syndrome.



**Figure 5:** Four years after the right foot operation.

Although initially Weaver syndrome was attributed to endocrine disorders, endocrine studies were proved to be normal in most studies. Kondo et al<sup>3</sup> reporting another case suggest that a genetic mutation might be the reason of the syndrome, the same in Japanese as other ethnic groups, and that probably the syndrome is inherited, an autosomal dominant disorder with variable clinical expressions.

Fryer et al<sup>4</sup> reported a family with affected father and daughter claiming that this provides evidence for autosomal dominant inheritance. Dumic et al<sup>5</sup> reported a pair of twins, brother and sister with the complete form of Weaver syndrome, and their mildly affected mother, suggesting autosomal dominant inheritance as well. In our patient it seems that her father was mildly affected because he had similar characteristics (large skull, broad forehead, flat occiput, mild mental retardation, wide set eyes, joint laxity, broad distal metaphyses).

A number of coexisting characteristics have been reported in the literature. Coulter et al<sup>6</sup> reporting a case of Weaver syndrome with neuroblastoma, stated that overgrowth syndromes have been reported to have an increased risk of tumorigenesis. This was supported by the hypothesis that a factor (s) capable of augmenting somatic growth plays a role in the pathogenesis of the tumour. Clinical, laboratory and radiological investigations showed no tumour in our case. Cardiovascular disorders that coexist with Weaver syndrome are reported by Sarigul et al<sup>7</sup>. Upper extremities and spine disorders are reported by Thompson et al<sup>8</sup>. Farrell and Hughes<sup>9</sup> reported a female case with bilateral pes cavus. In our case bilateral congenital dislocations of the hips and congenital dislocation of the ankle joint coexisted, a combination of deformities which has never been reported in the literature.

Although the patient had a chronic bronchitis and at-

electasy, there were no problems during the anaesthesia. Crawford and Rohan<sup>10</sup> reported that due to relative micrognathia, short neck and an anterior to cephalad position of the larynx there could exist difficulty in tracheal intubation, which was not observed in our case.

### Conclusion

We suggest that children with Weaver syndrome and disabling musculoskeletal deformities, particularly affecting their ability to stand up and walk should be treated early, before bone maturity, in order to achieve the best potential musculoskeletal and as extension to that developmental outcome.

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