Case series

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A proposal of rehabilitative approach in the rare disease "De Barsy Syndrome": case report

C. Celletti[1*], F. Camerota[1], T. Paolucci[2], L. Pezzi[2], D. De Meo[3], M. Castori[4], C. Villani[3], P. Persiani[3]

[1]Physical Medicine and Rehabilitation Division, Umberto I University Hospital of Rome; [2]Unit of Physical Medicine and Rehabilitation, G. D' Annunzio University of Chieti-Pescara, Department of Oral Medical Science and Biotechnology (DSMOB); [3] Department of Orthopaedic and Traumatology, Policlinico Umberto I Hospital-Sapienza University of Rome; [4]Divisione of Medical Genetics, Fondazione IRCCS-Casa Sollievo della Sofferenza, San Giovanni Rotondo, Foggia

Abstract

De Barsy syndrome is an autosomal recessive condition characterized by an progeroid appearance with distinctive facial features and cutis laxa. Ophthalmological, orthopedic, and neurological anomalies are generally also present. This syndrome is rare and the complex therapeutic management, from a surgical but also rehabilitative point of view, has not been recognized.

The aim of this paper is to describe a possible rehabilitative protocol, after an orthopedic surgical treatment, in a child with De Barsy Syndrome.

A 6-year-old boy was born with a congenital bilateral hip dysplasia associated with bilateral congenital foot deformity (vertical talus). Moreover, he showed stereotypic dyskinetic movements and psychomotor delay with cognitive impairment and absent language; the sitting position was maintained with orthoses to support the trunk control and the standing position was not acquired.

He was treated with pinstripe knee-highs for the foot and double nappy for the hips. At 19 months old, he underwent a two stage surgical approach for a bilateral pronated valgus foot with severe talonavicular subluxation. Satisfactory hip range of motion was achieved by conservative treatment alone. Afterwards, for the foot laxity and the flat-pronated foot corrective shoes were prescribed.

The main rehabilitative goals were: attention improvement, visual exploration for foot-eye and hand-eye coordination, encourage the essential prerequisites of language, controlling the upright position using support, improving hip-knee-foot relationship, improving load transfer between the right and left sides of the body, and bimanual coordination. The rehabilitation process lasted six months, three times a week, for a time from 30 minutes to 60 minutes per session.

The results were encouraging and the patient acquired the possibility of sitting with the indicated postural system, the possibility of assuming an upright position and taking a few steps with the aid of rollator with a postural stabilization system for the pelvis.***Clin Ter 2021; 172 (1):e4-7. doi: 10.7417/CT.2021.2273***

Key words: *cutis laxa, rare disease, autosomal recessive syndrome, multidisciplinary treatment*

Introduction

De Barsy syndrome (DBS) (MIM 219150) is a rare, autosomal recessive syndrome with cutis laxa (loose and anelastic skin). The young patients present lax skin with a defective elastic fiber with other secondary manifestation like the cornea cloudiness and developmental delay and cognitive impairments (1).

The DBS is a progeroid syndrome first described by De Barsy in 1968 (2) and successively described also by Karnes et al. (3) that observed aged appearance, dwarfism, mental retardation, right esotropia and myopia, lax skin, lax joints and elastic fiber deficiency. In addition, this syndrome has many orthopedics manifestations, including congenital hip dysplasia, joints hyperlaxity, scoliosis and severe foot deformities. (4) All these characteristics delay the acquisition of the sitting position and can also inhibit the standing position. In addition, the severe postural deficit and joint deformities are often aggravated by the presence of neurological disorders such as the presence of athetoid movements. This syndrome is rare and its complex therapeutic management, from a surgical but also rehabilitative point of view, has not been recognized (5-6).

These premises underline the importance of the rehabilitative team and a plan of treatment which, in childhood disability, represents a precondition that cannot be ignored: in this perspective, every professional figure (surgeon, neurologist, physiatrist, physiotherapist, psychologist) integrates and completes the work of other experts.

Then, this paper describes a possible rehabilitative protocol after orthopedic surgical treatment in a child with De Barsy Syndrome.

Correspondence: Claudia Celletti MD, PhD, Physical Medicine and Rehabilitation Division, Umberto I University Hospital of Rome, Italy
e-mail: clacelletti@gmail.com, Phone +39 328 6269632; ORCID: https://orcid.org/0000-0001-7154-5882

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Case presentation

The male infant was born at 34 weeks of gestation by a cesarean delivery because the fetus presented severe intrauterine growth retardation. At 25-27 weeks of gestation, his mother presented a threat of abortion. At birth, the baby showed auxological parameters lower than 3° percentile (length 33 cm, head circumference 25 cm, weight 1.100 gr) and fetal suffering (Apgar: 5 at the first minute and 7 at the fifth minute). He showed bilateral buphthalmos (left more severe than right), gastroesophageal reflux, hydronephrosis with the left megaureter, bilateral cryptorchidism and bilateral congenital hip dysplasia associated with talo-valgus feet. The gastroesophageal reflux was pharmacologically treated, and the ocular problem was brought back to a cataract.

At clinical evaluation at 6 years he, showed a weight of 13 Kg (severely less than 3° centile) associated to 105 cm of length (less than 3° percentile) and a head circumference of 49 cm (3° percentile). He showed an important generalized joint hypermobility at the interphalangeal hand joints and at the ankles. Hypermobility was associated with severe hypotonia, cutis laxa, progeroid aspect and venous network. (7) He showed also stereotypic dyskinetic movements and psychomotor delay with cognitive impairment and absent language. (8) The sitting position was maintained with the need for orthoses to support the trunk control, but the standing position was not acquired.

The genetic diagnosis was made with the identification of the mutation c.349G>T and c.540+1G>A in the PYCR1 gene in a compound heterozygous state.

Orthopedic treatment

The baby was born with a congenital bilateral hip dysplasia associated with bilateral congenital foot deformity (vertical talus). He was immediately treated with pinstripe knee-highs for the foot and double nappy for the hips. At the age of 11 months, he was treated with brace positioned in the abduction and extra rotation of the hips. A radiograph examination obtained 1 week into bracing treatment showed the hip had been reduced. After 40 days the bracing was removed, and radiographic examinations were done after one and three months (Fig. 1).

During all the period he underwent rehabilitation therapy for the foot deformity consisting in manipulation and application of casts followed by extensive soft-tissue releases. (9-11) At the age of 19 months, he presented a good range of motion of the hips but shows a bilateral pronated valgus foot with severe talonavicular subluxation (type vertical talus) that needs a surgical approach (Fig. 2A). A two stage surgical approach was performed: the first stage was a Z-lengthening of the Achilles tendon combined wih a leghtening of peroneal muscles tendons; in a second stage a reduction of the talus, stabilized with a Kirschner wire, was associated to a capsuloplasty and tenodesis. A rigid brace was applied for the realignment of the pelvis and trunk (Fig. 2B).

Kirschner wire was removed after one month and the brace after 50 days from the surgery; an orthosis like Bebax shoes were placed. (12) Then, the satisfactory outcomes allowed a rehabilitative approach with gait training using corrective shoes and walking aids.

The patient underwent periodic orthopedic examination. At the age of six years, he showed a joint laxity localized to knees and shoulders with spontaneous joint luxation. For the foot laxity and the flat and pronated foot corrective shows were prescribed.

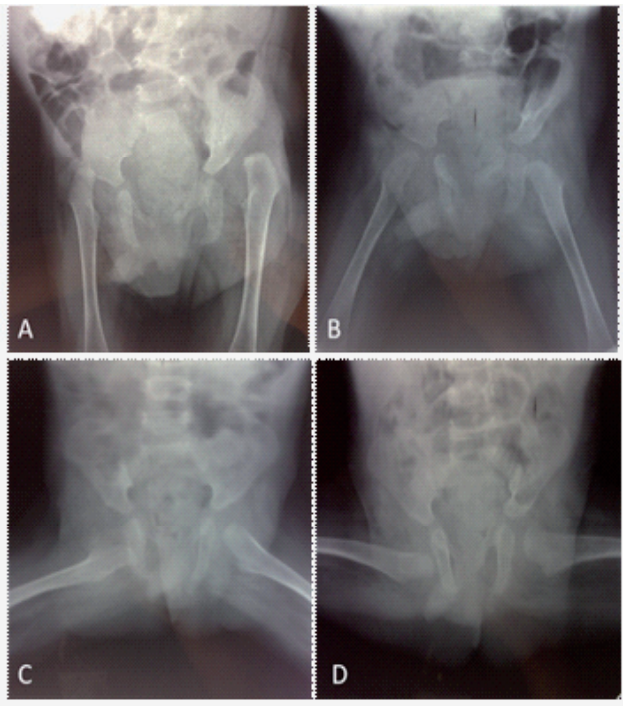

Fig. 1. Radiographic examination before (A) and one week (B) after the bracing treatment; radiographic control at one month (C) and three months (D) from the brace discharge.

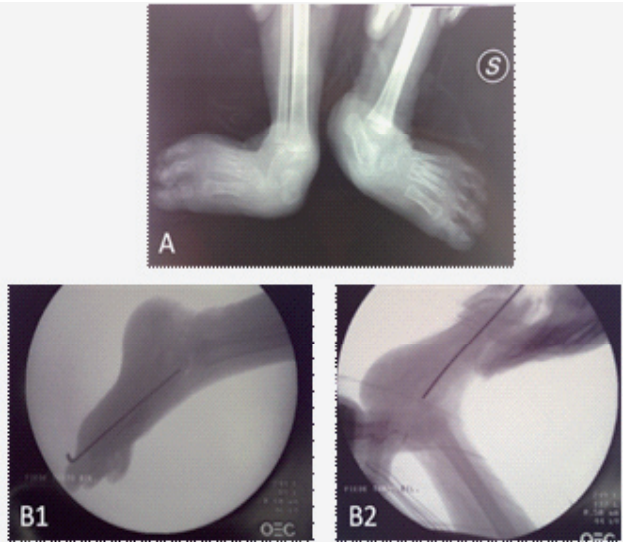

Fig. 2. Foot deformity before treatment (A) and after surgical correction for the left (B1) and right (B2) foot.

Rehabilitative treatment

The more significant rehabilitative aspects treated were the joint laxity and the postural alterations of the pelvis and trunk. The upper limbs were characterized by a flexed finger with a bilateral reduction of the fifth finger range of motion. He used a global socket and was not able to singularize the finger motion and the flexion-extension of the wrist was limited. The trunk was anterior flexed, and the head was down positioned and right inclined.

A specific postural device was prescribed to maintain a correct postural alignment for maintaining a better sitting position. The postural system was characterized by an adjustable width/depth seat, variable leg length and platform tilt, reclining, elevating, and tilting platform with support for trunk and head. The correct alignment of the trunk and head has also allowed improving his visual control of the peri-personal space, allowing him a better interaction with the family context. The main rehabilitative goals were: the attention improvement within a complex treatment, visual exploration for foot-eye and hand-eye coordination exploiting the play context and appropriate for the cognitive age of the child, encourage and accompany the essential prerequisites of language, control of the upright position using support, improve the hip-knee-foot relationship, improve the load transfer between the right and left sides of the body, bimanual coordination.

To stimulate attention and facilitate the representation of the movement required during rehabilitation sessions, many mirror exercises have been proposed, sometimes even two mirrors placed rear and front.

Discussion

The results have been encouraging. At the end of the rehabilitation process which lasted for six months three times a week for a time from 30 minutes to 60 minutes per session (taking into account the participation and cognitive fatigue of the child) the patient had acquired: the possibility of sitting with the indicated postural system, the possibility of assuming an upright position and taking a few steps with the aid of rollator with a postural stabilization system for the pelvis. Parents were instructed to perform rehabilitation treatment three times a week with a physiotherapist specializing in childhood rehabilitation. Periodic checks were performed by the physiatrist and child neuropsychiatrist every six months. In addition to motor rehabilitation, indications were given, in agreement with the child neuropsychiatrist, to integrate the treatment with a specific path of cognitive and psychomotor rehabilitation.

Taking care of the child with a chronic disabling pathology necessarily requires multidisciplinary and interdisciplinary teamwork and a punctual and personalized rehabilitation plan that always includes parental figures. The need for personalized surgical treatments has been followed by a non-intensive rehabilitation protocol which was based on the "modifiability" of the functional conditions of our young patient with DBS. Our rehabilitation process has tried to answer mainly these questions: What can be changed? in which direction? to what extent? when and for how long? Mostly in rare diseases such as DBS, the rehabilitation process, should aim to guide the child with impairment to face the problems that growth gradually offers him. Taking charge of rehabilitation must consider a treatment path where healing is not possible that must integrate with the times, needs and desires of the life of the child and their family: "not life made of rehabilitation but rehabilitation to improve the life". The improvements reported in our young patient are a result of a close collaboration between the surgeon and the rehabilitators and are encouraging so that specific rehabilitation pathways must be structured in children with DBS and in all those forms of rare diseases where, to date, there are no specific rehabilitation protocols.

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Consent

Written informed consent was obtained from the patient's parents for publication of this case report and accompanying images.

Declaration of competing interest

The authors declare that they have no conflict of interest.

Conceptualization: Villani C.; Persiani P.
Data curation: De Meo D.
Investigation: Castori M.
Writing – original draft: Celletti C.
Writing – review and editing: Celletti C., Camerota F., Paolucci T., Pezzi L.

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