ABSTRACT

Purpose: The purpose of this case report is to show one of the rarest syndromes such as that of Patterson – Lowry and to prove that with a wide variety of treatment procedures, physiotherapy and rehabilitation speciality could improve patients’ subjective complaints and restore the basic functions of the locomotor system in rare syndromes such as that of the two brothers represented in this report.

Materials and Methods: The methods of physiotherapy and rehabilitation offered by Pavel Banya MC were used, as well as the methods of kinesiotherapy.

Results: Improvement of patient’s subjective complaints.

Conclusion: Balneotherapy procedures have a beneficial effect on patients with rare diseases, not only through their analgesic action but also through the professionalism and the hope that experienced rehabilitators give patients and their families.

Keywords: Patterson-Lowry syndrome, rare syndromes, physiotherapy, limb shortening

BACKGROUND

These are two 15-year old patients – identical twins who received treatment at Pavel Banya Medical Center in July 2016, one of them (we will call Patient 1) reported that in Belgium he was diagnosed with an extremely rare disease – Patterson-Lowry syndrome.

The Department of Clinical Genetics at the ULB has offered to the parents of Patient 1 to make further genetic testing of the brothers and their parents by whole-exome sequencing, to seek for the genetic anomaly, manifested only in Patient 1. This could allow to find out the cause of this rare skeletal anomaly. Although an absolutely free of charge testing was offered, the mother refused, as she believed the testing would not help the child’s condition. If, however, the testing is to be made, the mode of inheritance, the cause of the disease, and the risk to the offspring of Patient 1 could be clarified.

The latest presented case of the twins with the Patterson-Lowry syndrome revealed a noteworthy fact - of the 6 cases described in the world to the present, 4 of the patients were from the Western Balkans, and for the other two patients, there is no clear indication of their origin [1].

AIM

The aim of this case report is to investigate and describe an extremely rare case of two twin brothers diagnosed with the Paterson-Lowry syndrome, as only 4 cases have been reported in the world, and to track the development of subjective complaints and the condition of patients during their rehabilitation at the Pavel Banya Medical Center.

CASE DESCRIPTION

Patient 1 and Patient 2 are two identical twins born on 12th July 2001 (15-year old). Since the age of 10, Patient 1 has complained of not being able to raise his arms at the shoulder joints, of pain in the muscles of the lower extremities and the back, as well as of flexion deformity of the distal phalanges of both 5th fingers without prior trauma. His brother, Patient 2, and their mother have also reported chronic back pain.

Fig. 1. Patient 1. Fig. 2. Patient 2.
After consultations and investigations at the Genetics Center of the Cliniques Universitaires Saint-Luc, Brussels, the Queen Fabiola Children’s University Hospital, a member of Université Libre de Bruxelles (ULB) and presentation of the case to a European skeletal dysplasia working group, the diagnosis was made - an extremely rare Patterson-Lowry rhizomelic dysplasia. The case of Patient 1 (Fig. 1.) is only the fourth such case in the world.

The syndrome is characterized by humerus shortening and humeral head dysplasia, but other abnormalities can also display short stature [2]; back pain and hip muscles pain; camptodactyly - the same as described in patient 1 (Fig. 1.). In comparison to his brother (Fig. 2.), Noted were also: short metacarpal, metatarsal bones and phalanges; coxa vara [2, 3].

The clinical and radiological signs of the syndrome may vary, but the shortening of both humeri, combined with humeral head dysplasia, makes the syndrome easily identifiable. The origin of the Patterson-Lowry rhizomelic dysplasia is currently unknown as well as its mode of inheritance. Women and men are thought to be equally affected.

The Patterson-Lowry syndrome was first described in 1975 by Caroline Patterson and R. Brian Lowry, describing a man of small stature with shortened limbs, most noticeably of the humeri and dysplasia of the shoulder joints [2, 4].

A second case was described in 1995 by Williams et al. who reported the case of a mentally normal 5-year-old boy with anomalous segmentation of the proximal humeral metaphyses, brachydactyly and brachymetacarpalia.

In 2004, a publication by Franceschini et al. in the American Journal of Medical Genetics reported two patients with many similar manifestations [4] – 1 Albanian and 1 Romanian with no connection between them. Both of them exhibited short humeri, coxa vara, short metacarpals, metatarsals and phalanges. The changes in the humerus, according to Franceschini, were characterized by striking flattening of the proximal epiphyses and lateral bulging of the proximal diaphyses, which was also established in Patient 1’s X-rays [5].

In a 2001 publication, Kamoda et al. reported this diagnosis in two sisters with humeral changes. However, these sisters were mentally retarded and therefore, should be treated as cases of another syndrome [6].

Physical examination: Obvious shortening of both humeri. Limited abduction of both shoulder joints. 5th finger camptodactyly of the upper limb, bilaterally. Light scoliosis of the thoracolumbar transitional region with the rigidity of the paravertebral muscles on the left. Hypermobility in the lumbar region. Preserved muscle strength and sensitivity of both upper limbs (Fig. 3.). Abnormal flexion of both shoulder joints in Patient 1 with 5th finger camp to dactyly (Fig. 4.). Slight scoliosis of the twins with higher left shoulders (Fig. 5.). Patient 2 – no scoliosis (Fig. 6.). Obvious shortening of both humeri in Patient 1 (Fig. 7, 8.).

Fig. 3. Obvious shortening of both humeri.
Fig. 4. Abnormal flexion of both shoulder joints in Patient 1 with 5th finger camp to dactyly.
**Fig. 5.** Slight scoliosis of the twins with higher left shoulders.

**Fig. 6.** Patient 2 – no scoliosis.

**Fig. 7.** Obvious shortening of both humeri in Patient 1 (front).
Fig. 8. Obvious shortening of both humeri in Patient 1 (back).

X-Ray: Investigations were conducted at Pavel Banya Medical Center Ltd.

To Patient 1 we conducted X-rays radiographs of humerus and shoulder joints (Fig. 12, 13, 14, 15), frontal and lateral cervical region X-rays; frontal and lateral thoracic region X-rays (Fig. 11.); frontal lumbar region X-rays; X-ray of palms bill. (Fig. 16, 17.), electromyography and electroneurography.

To Patient 2, we conducted frontal and lateral thoracic region X-rays. X-rays of Patient 2 – left convex scoliosis from Th5 to L4 with enhanced lumbar lordosis (Fig. 9., 10.); electromyography and electroneurography.

Fig. 9. X-rays of Patient 2 – left convex scoliosis from Th5 to L4.

Fig. 10. X-rays of Patient 2 – enhanced lumbar lordosis.
**Fig. 11.** First featured X-rays of Patient 1, made in Belgium.

**Fig. 12.** X-rays of Patient 1’s both humeri – congenital malformation of both humeral heads with lateral bulging of the proximal diaphyses and shortening of the two bones.

**Fig. 13.** X-rays of Patient 1’s both humeri – congenital malformation of both humeral heads with lateral bulging of the proximal diaphyses and shortening of the two bones.

**Fig. 14.** X-rays of Patient 1’s both humeri – congenital malformation of both humeral heads with lateral bulging of the proximal diaphyses and shortening of the two bones.
Fig. 15. X-rays of Patient 1’s both humeri – congenital malformation of both humeral heads with lateral bulging of the proximal diaphyses and shortening of the two bones.

Fig. 16. X-rays of Patient 1 – 5th finger camptodactyly, bilaterally.

Fig. 17. X-rays of Patient 1 - 5th finger camptodactyly, bilaterally.

Therapy course: The following therapy courses were carried out:
• Electrotherapy;
• Magnetic and brine therapy;
• Ultrasound therapy
• Underwater hydro massage with aromatherapy;
• Underwater exercises for the upper limbs;
• Dry massages

RESULTS:
This case report was represented to show that there was an improvement of patient’s subjective complaints. This report investigated and described an extremely rare case of two twin brothers diagnosed with the Paterson-Lowry syndrome, as only 4 cases have been reported in the world, and followed the development of subjective complaints and the condition of patients during their rehabilitation at the Pavel Banya Medical Center – patients’ subjective complaints and restore the basic functions of the locomotor system in rare syndromes such as that of Patterson-Lowry was improved.

DISCUSSION:
Physiotherapy and rehabilitation is a multi-discipline covering all medical specialities and serving a wide range of patients with various diseases and rare syndromes, as described in this report. With a wide variety of treatment procedures, this speciality could improve patients’ subjective complaints and restore the basic functions of the locomotor system in rare syndromes such as that of Patterson-Lowry [1, 5].

Balneotherapy procedures have a beneficial effect on patients with rare diseases, not only through their analgesic action but also through the professionalism and the hope that experienced rehabilitators give patients and their families [3].
CONCLUSION:
Balneotherapy procedures have a beneficial effect on patients with rare diseases, not only through their analgesic action, but also through the professionalism and the hope that experienced rehabilitators give patients and their families.

REFERENCES:

Please cite this article as: Gecheva-Fermendzhieva G, Radev R, Marinov M. Balneotherapy in an extremely rare condition – Patterson-Lowry syndrome. J of IMAB. 2019 Oct-Dec;25(4):2805-2811. DOI: https://doi.org/10.5272/jimab.2019254.2805

Received: 28/02/2019; Published online: 02/12/2019

Address for correspondence:
Dr. Gergana Gecheva – Fermendzhieva
Medical centar “Pavel banya” Ltd,
2, Osvobojdenie str., Pavel banya, Bulgaria,
E-mail: dr.ggecheva@abv.bg,