SCA11 - Rehabilitation in spinocelebrallar ataxia: a case report

Chiara Mastantuono [1], Luca Labianca [2], Carlo Brogioni [3], Mirco Fava [1], Luigina Petrucci [1], Elena D'Agostino [1], Chiara Marcheggiani [1]

1 Officina dei Sensi, Ascoli Piceno Italy - **2** Sapienza University, Roma Italy - 3 Ospedale Area Vasta 5 Ascoli Piceno Italy

Case description

24 year old girl affected by mutazione de novo c.1287-1288delGAS mutation in TTBK2 gene, compatibly with SCA11, epilexia in childhood with generalised crisis treated till the age of 13, since 2010 double vision appearance with associated correction intervention. The patient relates an enlarged instable walking , associated with nystagmus with prismatic correction. She shows uncertainty in addition to majour right soft bilateral dismetry, lack of static and dynamic balance and instability in the change of directions.

Case diagnosis

The spinocelebral ataxia type 11 (SCA11)which it is reckoned represents the 2% of ADCA type 3 cases , is characterised by progressive celebral ataxia , walking and balance difficulties and abnormal ocular signs, jerkily pursuits and nystagmus , pyramidal features , peripheral neuropathia with numbness, weakness at the edges and in other parts of the body and dysarthia.

Discussion

Since November 2019 the girl performs hydrotherapy activitie, ortoptic exercices and and psychological counselling lasting 40 and 60 minutes . The motor activities of hydrotherapy , followed by specialised staff and at the temperature of 34 degrees, were made to reinforce the balance skills with and checked transfert the load from a limb to the other, and pelvis rotation activity, keeping her stem, shoulders and head steady. The integration of the hydrotherapy and psychological intervention has led to the following results: items E of the GMFM an increase of the percentage from 66.7 to 70.8 and in the A B C D items a stationary tendency of 100%, 96.7% , 8.1%.

Conclusions

This case confirms, with the collection of data, how usefull it is to those patients affected by forms of ataxia , even if declined, to obtain unexpected results in terms of increase of the quality of life , security and authonomy.

OFFICINA dei sensi
CENTRO POLIFUNZIONALE