PO-19

Horizontal Gaze Palsy and progressive scoliosis syndrome-case report

Anja Vidmar, Silvija Delfin

Očesna klinika, Univerzitetni klinični center (UKC) Ljubljana, Slovenia

The purpose of the article is to present 2 cases with horizontal gaze palsy and progressive scoliosis syndrome. Only individual cases of the syndrome are described in literature. 2 children (siblings) with limited abduction and adduction were examined at the Eye Clinic in Ljubljana.
After additional examinations the diagnose of horizontal gaze palsy and progressive scoliosis syndrome was determined. It is a rare autosomal recessive disease caused by a mutation of the ROBO 3 gene on chromosome 11, which controls the crossing of brainstem neurons, neurite outgrowth, growth cone guidance and axon fasciculation.
Of the ophthalmological signs, there is complete limitation of abduction and adduction beyond the primary position with appropriate elevation and depression. Limited bulbomotor palsy can lead to diplopia with subsequent possible amblyopia. Radiological changes on MRI are characteristic: loss of the facial colliculus due to the absence of the abducens nerve nucleus, split pons sign, butterfly appearance and hypoplasia of the medulla oblongata, anomalies of the 4th ventricle. Radiological examinations of the spine show scoliosis. Therapy of ophthalmological symptoms is individualized and may include the prescription of prism lenses in the case of diplopia. Most often, it is the prescription of prisms of lower powers (up to 20PD). Strabismus surgery is also considered, most often due to esodeviation up to 20 PD. Developmental delay has also been described in children, which improved after the diagnosis and understanding of specific ophthalmological limitations by therapists. Learning everyday strategies to overcome a narrower functional visual field is also crucial. Early recognition of the syndrome of limited horizontal bulbomotor activity and progressive scoliosis allows for rapid intervention and appropriate multidisciplinary treatment.

Sindrom omejene horizontalne bulbomotorike in progresivne skolioze-prikaz primera

Anja Vidmar, Silvija Delfin

Očesna klinika, Univerzitetni klinični center (UKC) Ljubljana, Slovenia

Namen prispevka je predstaviti 2 primera s sindromom omejene horizontalne bulbomotorike in progresivne skolioze (angl. Horizontal Gaze Palsy and progressive scoliosis syndrome). V strokovni literaturi so opisani le posamezni primeri sindroma. Na Očesni Kliniki v Ljubljani sta bila pregledana 2 otroka (sorojenca) z omejeno abdukcijo in addukcijo. Po opravljenih dodatnih preiskavah je bilo ugotovljeno, da gre za sindrom omejene horizontalne bulbomotorike in progresivne skolioze. Gre za redko avtosomno recesivno bolezen, povzročeno z mutacijo ROBO 3 gena na kromosomu 11, ki nadzoruje križanje nevronov možganskega debla oz izraščanje nevrita, vodenje rastnega stožca in fascikulacijo aksonov. Od oftalmoloških znakov je prisotna popolna omejenost abdukcije in addukcije čez primarni položaj ob ustrezni elevaciji in depresiji. Omejena bulbomotorika lahko privede do diplopije s posledično možno ambliopijo. Značilne so radiološke spremembe na MRI: izguba facialnega kolikulusa zaradi odsotnosti jedra n. abducensa, znak razcepljenega ponsa, metuljni izgled in hipolazija medulle oblongate, anomalije 4. ventrikla. Radiološke preiskave hrbtenice pokažejo skoliozo. Terapija oftalmoloških simptomov je individualizirana in lahko vključuje predpis prizemskih leč v primeru diplopije. Najbolj pogosto gre za predpis prizem nižjih jakosti (do 20PD). V poštev pride tudi operacija strabizma, najbolj pogosto zaradi esodeviacije do 20 PD. Pri otrocih je opisan tudi razvojni zaostanek, ki se je pa izboljšal po postavitvi diagnoze in razumevanja specifičnih oftalmoloških omejitev s strani terapevtov. Ključno je tudi učenje vsakodnevnih strategij za premagovanje ožjega funkcionalnega vidnega polja. Zgodnje prepoznavanje sindroma omejene horizontalne bulbomotorike in progresivne skolioze omogoča hitro ukrepanje in ustrezno multidisciplinarno zdravljenje.