ROLE OF SPEECH THERAPY TRAINING IN A YOUNG PATIENT AFFECTED BY SEVERE SENSORINEURAL HEARING LOSS AND CONCOMITANT VISUAL IMPAIRMENT

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ABSTRACT

The article aims to describe in a detailed way the logopedic intervention made on a young patient affected by Peroxisome Biogenesis Disorders (PBD). In particular, the focus is the relationship between the two affected senses: sight and hearing. Our patient is a 10-year-old female with an early onset Retinitis Pigmentosa (RP), Sensorineural Hearing Loss (SNHL) and a mild cognitive impairment. The child has been precociously prothesized with a good prosthetic outcome and underwent both speech therapy and neuro-psychomotor therapy. A stability in her hearing abilities was detected, but the growing visual impairment was accompanied by a deficiency of attention abilities, of visual perception abilities and of visual-motor integration.

INTRODUCTION

One of the main characteristics of living beings is the ability to interact with their environment. This ability depends on their capability to extract useful information from the environment and elaborate adaptive responses; this allows living beings to survive and evolve both individually and as members of a given species. An organism's perception of their environment depends on their sensory functions.

The senses allow us to communicate with the external world: through the senses we explore, understand, interpret, and interiorize the world we live in. Having a knowledge of oneself and being able to get to know the world through the sensations our body receives, thanks to our perceptive organs, is fundamental: we are constantly receiving visual, auditory, tactile, smell and taste related information in everyday situations. When we talk about the senses, we talk about the body's functions and structures. The body's structures are the "apparati", the different parts of our body. These are known to function only if they are complete and if they are supported by the body's functions, that is the systems' physiological functions. Our senses (seen as structures and functions which can function or not) guide our daily life in every moment. If everything is working correctly, sight, smell, taste, hearing, and touch work synergically and constantly to allow us to understand the world around us, to relate with it and to regulate our activity. What if one or more of these fail?

Maybe the senses we fear of losing the most are sight, which is the most used one, and hearing.

CLINICAL CASE PRESENTATION

Mutations of the PEX1 gene are the most common cause of Peroxisome Biogenesis Disorder (PBD) with mild to severe phenotypes.

Here, we present a 10-year-old female with an early onset RP, SNHL and mild cognitive impairment, who resulted positive to pathogenic mutations in PEX1 gene.

The clinical history of the patient begins at 3 months of age, with the onset of moderate to severe bilateral sensorineural hearing loss confirmed by the Auditory Brainstem Responses; at 6 months of age the patient was treated with behind-the-ear hearing aids and started weekly speech therapy.

The neuro-psychiatric assessment was done again in December 2016 at the age of 8. The child showed to be collaborative and available towards the examiner from the beginning and performed the requested tasks, although occasionally displaying inadequate levels of motivation and a delay in performing such tasks.

The clinical evaluation showed the following characteristics:

- An impairment of the intellectual functions associated with a substantial difficulty in the adaptive functionality. These two elements together allowed to diagnose a mild intellectual disability.
- The verbal comprehension abilities, the speed of elaboration and the working memory are sensibly compromised. There was a drastic drop in the working memory.
- The child's attention levels were inadequate; recalls to the current task and reinforcement were often required.
- Speech was fluent enough, but still immature and affected by phono-articulatory defects.

Annual audiological follow ups showed a sloping audiometric threshold (Sloping Audiogram with PTA of about 70 dB without prothesis and of about 30 dB with prothesis) that seems to be not progressive over time.

At the age of 8, the patient experienced night blindness and a reduction in visual acuity and, later, a specific genetic investigation highlighted the presence of a mutation in the PEX1 gene.

Speech therapy was increasingly focused on improving auditory abilities and cognitive maturation.
The speech therapist explained the importance of letting the child discover the world of sounds and of words in the following ways:

- have her listening to music and let her grasp the alternation between sound and silence
- accustom the child to take part in the world of sounds
- have her use touch to discover sound vibrations.

The speech therapist then proceeded to work on strengthening the auditory abilities through:

- detection
- identification of the sound source
- listening to herself: focusing and discovering her own voice
- identification (closed list)
- speech tracking (appropriate to the age)
- acknowledgement of the same material with and without clue (age 2 and up)
- common questions with and without clue (age 2 and up).

Cognitive therapy evolved through the following 6 phases:

- Stimulation and development of the following activities, which are essential to allow a good cognitive maturation: interest, attention, perception, memory, metacognition
- Development of vocabulary
- Similarities/differences and contrasts
- Classification and knowledge of semantic fields
- Acquisition of grammar elements and the morphosyntactic structure of the sentence
- Discovery of the meaning of simple sentences and, in time, more complex sentences, through early reading of new words

In June 2018, the child received the phoniatric evaluation. This evaluation showed that the child was collaborative with the given tasks, she tolerated the waiting times and respected the alternation of turns, although with discontinued attention times. No difficulty in establishing a relationship with the examiner was observed. A good level of motivation while carrying out tasks was detected. The formal linguistic abilities and the narrative abilities were found to be discrete.
Moreover, the child was given repeated free-field tonal audiometry tests, which highlighted a good prosthetic outcome. The child did not experience a decline in the auditory situation, but there were also no improvements in the last years. In the same period, the child also experienced a visual impairment.
People affected by deafness have a significantly modified view of the surrounding reality compared to able bodied people, because they must compensate for the hearing loss with an emphasized use of sight. The eyes turn into a critical instrument to compensate for the auditory impairment with regards to communicative strategies, such as picking up and deciphering facial expressions rapidly, lip reading and the interlocutors' gestures.
With regards to the patient in exam, whose auditory impairment remained unvaried, since the visual impairment worsened and affected language acquisition, we detected a deficiency of attention abilities, of perceptive abilities and of visual-motor integration.

SIGHT-HEARING LINKS

The close association between visual and auditory information was discovered by professor Proverbio's research group that demonstrated that viewing a photograph associated with a sound can activate the associative auditory cortex. High density ERPs have been recorded in 15 participants while viewing hundreds of images which were associated or not with sounds. Auditory stimuli have been distinguished from non-auditory stimuli at as little as 110ms. SwLORETA's reconstructions have highlighted the common activation of areas of the ventral auditory processing stream in both types of stimuli and of the associative temporal cortex, in the initial phase, only for the auditory stimuli. The primary auditory cortex (BA41) was activated by auditory images after about 200ms.
The visual stimuli can change the listener's auditory perception in subtle ways, as in McGurk's illusion. Here, the perception of a phoneme's auditory identity is modified by a simultaneous video of a mouth while it articulates a different phoneme. With McGurk's effect we can demonstrate that the neural representations in the auditory cortex are related to the visual stimulus of the mouth's articulations, which guide the illusory subjective auditory perception, compared to the auditory stimulus.

CONCLUSIONS

Ultimately, I had the opportunity to deepen my knowledge of this rare disease by collaborating with the Phoniatric and Audiology Divison of the Department of Mental and Physical Health and Preventive Medicine of the University of Campania "Luigi Vanvitelli" (Napoli), with the Ophthalmic Clinic of the Multidisciplinary Department of Medical, Surgical and Dental Sciences of the University of Campania "Luigi Vanvitelli" (Napoli) and with the Medical Genetics Laboratory of the Department of Precision Medicine of the University of Campania "Luigi Vanvitelli" (Napoli).
We presented the case of a 10-year-old female with an early onset of RP, SNHL and mild cognitive impairment. The child was precociously prothesized with a good prosthetic outcome and underwent speech therapy and neuro-psychomotor therapy. At the age of 8, she experienced night blindness and a decline in visual acuity. Stability in the auditory abilities was detected. The increase of the visual impairment was also associated with an inadequacy of the attention abilities, of visual perception abilities and of visual-motor integration.
This article has also analyzed the interaction between the two affected senses (sight and hearing) through research studies.
The interaction between the senses in the field of perception is well known and when the information coming from a sense is ambiguous, another sense can compensate and make the information clearer. Most of us know that sight can influence hearing, but we tend to believe that "we see what we see" and "we hear what we hear". Thanks to these research studies, we were able to demonstrate that the impact of the visual information on the auditory system is significant.
I would like to end by quoting a Lebanese poet called Khalil Gibran:
"*You are blind, and I am deaf and dumb, so let us touch hands and understand*."

REFERENCES

1. Adriana De Filippis, Il Nuovo Manuale di Logopedia, Erickson, 1998
2. Adriana De Filippis, L'impianto cocleare-Manuale operativo, Elsevler-Masson, 2002
3. Alajmo E., Otorinolaringoiatria, Piccin- Nuova Libreria 1995
4. Anne M. Gilroy, Brian R. MacPherson, Lawrence M. Ross, Prometheus- Atlante diAnatomia, Edises, 2013
5. Dott. med. Hanno Bolz / Prof. dott. med. Andreas Gal, Istituto di genetica umana, Clinica oculistica universitaria di Amburgo-Eppendorf, "Genetica della sindrome di Usher", Giornale Retina Suisse, 2-3 settembre 2003, n°88/89.
6. Eldra P. Solomon, Linda R. Berg, Diana W. Martin, Biologia, Edises 2017
7. Elliot Smith,Scott Duede, Sara Hanrahan, Tyler Davis, Paul House, Bradley Greger , Vedereper credere: rappresentazioni neurali di stimoli visivi nella corteccia uditiva umana si collegano conpercezioni uditive illusorie Pubblicato: 4 settembre 2013;/https://doi.org/10.1371/journal.pone.0073148/
8. G. Binelli, D,Ghisotti, Genetica , Edises 2017 https://www.ncbi.nlm.nih.gov/books/NBK1265/
9. James Edge ed Akiko Matsumato, Consulente: Steven D. Collins, Gallaudet University, Redazione: Giorgio Aloisio, "Introduzione alla Sindrome di Usher", Edizione Kappa, 2007-2009.
10. Keats BJB, Lentz J. Usher Syndrome Type I. GeneTests. University of Initial Posting:December 10, 1999; Last Update: May 2016. Washington; Seattle:
11. Luciano Liuzzi, Franco Bartoli, Manuale di oftalmologia- Testo atlante, Minerva medica,2002
12. Marco Piccolino, Anacleto Navangione, I sistemi sensoriali "accordano" gli organismi col loro ambiente (19.1), "Un sistema sensoriale ad alte prestazioni, La retina dei vertebrati"
13. Martini F.H.. Timmons M.J., Tallitsch R.B., Anatomia Umana, Napoli, Edises, 2016
14. NIDCD Fact Sheet: "Usher Syndrome", u.s. department of health & human services nationalinstitutes of health national institute on deafness and other communication disorders, Publication No. 98-4291, Updated February 2008.
15. Proverbio AM, D'Aniello GE, Adorni R, Zani A (2011). When a photograph can be heard: vision activates the auditory cortex within 110 msec. www.Nature.com/ScientificReports. 1:54.DOI:10.1038/srep00054.
16. S. Prosser, A. Martini; Argomenti di Audiologia, Omega, 2013