



*an eu curriculum for  
visual disabilityEs  
RehabilitatOrs*



## Case study documents LT4y

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### VERSIONING DETAILS

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## **Instructions**

The attached documents are meant to support case analysis in multi-Disciplinary Team discussion simulation (see Lesson Plan LO3-D-E-1 "Promoting neuro-psychomotor development within the MDT)

The Functional clinical report layout was cut to ease the distribution of documents between involved students as the selection of attached professional reports. Together with the displayed materials some video resources are attached and listed in the last page of this document.

# MEDICAL HISTORY

L.T.                    gender M

Date of birth: 20/04/2018

## **ICD 9 Diagnosis Code**

36900- Profound impairment, both eyes, impairment level not further specified

## **ICD 10 Diagnosis Code**

F84 Pervasive developmental disorders

H54 Visual impairment including blindness

## **Notes**

Pervasive motor skill disorders, blindness bilateral. Suspected Leber amaurosis

# FUNCTIONAL CLINICAL REPORT

FUNCTIONAL CLINICAL REPORT 2022

L.T. 4Y

## Medical history and aspects

Severe hypo virus in a child with early-onset Leber congenital amaurosis. He is followed for the diagnostic aspects at the Mondino Hospital (Pavia).

Since being under our care since 2019, the child has been participating in neuropsychomotor training sessions and receiving speech therapy twice a week. He also undergoes an annual cycle of physiotherapy and visual rehabilitation.

He is in his first year of kindergarten at San Pietro school where he receives individualised teaching and support from an Educational Operator. He has been certified as having a serious handicap situation (paragraph 3) and civil disability, providing certain benefits.

During the psychiatric visit on 26/2, bilateral flat-valgus was noted particularly on the left, with consequent prescription of orthotics. In general, feedback of good motor skills, safer coastal path, the child seeks greater independence and likes climbing and overcoming obstacles.

The last ophthalmological evaluation done in October 2021, revealed no eye docking or fixation in both eyes, constant pendular nystagmus and dull pupillary reflexes.

## 1. RELATIONAL AND BEHAVIORAL ASPECTS

L.T. is a calm smiling and affectionate child who uses voice to recognise and discriminate between people familiar to him and strangers. He has good relationships with his therapists and appreciates body contact with his caregivers, often requiring attention and physical contact. Both the signs of Franceschetti, which include behaviours exhibited by children with severe visual impairment, such as pressing and crushing of the eyeballs with the fist or fingers, as well as antero-posterior swings of the trunk, persist. These behaviours typically subside after verbal prompting from the caregiver and are notably intensified during moments of reduced sensory input (hypoafferentation).

## 2. COGNITIVE/NEUROPSYCHOLOGICAL ASPECTS

L.T. demonstrates partial collaboration and, although there has been some lengthening of attention times in recent months, they still remain short and are not commensurate with their chronological age.

Regarding body scheme, L.T. demonstrates the ability to identify various body parts such as eyes, nose, mouth, ears, shoulders, belly, back, armpits, knees, heels, and feet. This indicates an understanding of topological references, particularly distinguishing between the front and back of the body (belly-front/back-back). L.T. also exhibits the capability to identify basic body parts on another person and, in emergency situations, on a doll.

Recently, there has been progress in L.T.'s ability to recognize and differentiate simple shapes, including circles and crosses.

### 3. COMMUNICATION ASPECTS

L.T. is a sociable and expressive child who prefers verbal communication. He demonstrates strong communicative intent and expresses himself through modulated facial expressions. He actively seeks attention from others through physical gestures and calling them by name. While he tends to be more passive in conversations, he has started to spontaneously comment on situations, although sometimes using stereotyped phrases

### 4. LINGUISTIC ASPECTS

L.T.'s language is continuously evolving in a positive manner, although sometimes can be repetitive and echolalic. L.T. faces challenges in generating/adapting expressions and phrases to the context, due to linguistic immaturity. With semantic content not always fully functional or relevant to the context. L.T. can answer simple questions about the "here and now," and is beginning to answer to questions that are not directly related to the immediate context. He often calls upon others to share or highlight events or sounds that they have heard in the moment

At a phonetic-phonological level, the inventory is still incomplete: systemic and structural processes are present, but they can be considered physiological for the age.

L.T. can recognise and name objects familiar to him even in different contexts. However, expansion of his vocabulary is being limited by his tactile experiences but is expanding steadily at a slow rate.

In terms of morphosyntactic skills, L.T. demonstrates the ability to construct subject-verb-object (SVO) sentences. However, there are frequent errors in the use of free morphology, and they encounter challenges in utilising linked morphology and employing personal pronouns correctly. While L.T. shows understanding of simple sentences, comprehension of sentences with medium difficulty in terms of morphosyntactic structure is still underdeveloped.

## 5. MOTOR-PRACTICAL ASPECTS

L.T. tends to walk in an indirect manner, with a slightly widened support base, and does not always use his upper limbs to explore or protect against obstacles. As a result, verbal cues from his caregivers are often necessary to avoid harm. However, within the neuro psychomotor room, L.T. can independently navigate routine movements such as going from the door to the chair, or vice versa. L.T. moves around the perimeter of the room while identifying objects such as tables, chairs, wardrobes, mirrors, radiators, etc. He is also capable of overcoming low-height obstacles and enjoys sensorimotor experiences such as swinging and jumping on the Bobath ball with the support of an adult at the front.

During the recent period, there has been improvement in tactile exploration skills, although it still remains at an immature and imprecise level. The rate of exploration has decreased compared to previous assessments. L.T. demonstrates the ability to differentiate various textures, including smooth, rough, striped, and soft textures.

L.T. is capable of grasping objects that are placed in his hand and can search for lost objects for short periods of time. He can also get objects from containers, and then correctly place them back in place, close the container lids or replace screw caps.

L.T. demonstrates that he can recognise certain everyday objects through appropriate use, indicating a significant understanding of object functionality.

L.T. displays a high level of attentiveness to sound stimuli, as demonstrated by his ability to accurately orient himself towards the source of the sound. He can also recognise familiar sounds.

## 6. ASPECTS of the GAME

The ludic baggage is still predominantly in the sensorimotor phase. L.T. enjoys dropping, throwing, and shaking objects to hear the accompanying noises. He has started to engage in functional play and the early stages of symbolic play, such as feeding a baby or combing hair, but requires support from an adult to do so.

## 7. ASPECTS of LEARNING

Not developmentally relevant.

## 8. ASPECTS of AUTONOMY, ORIENTATION AND MOBILITY

With regard to the autonomy of food and clothing, L.T. cooperates in being dressed, when it comes to food and clothing autonomy, L.T. cooperates with getting dressed, he can drink from a cup independently, attempts to use a spoon while eating. He can remove his overcoat, socks, and shoes when undressing.

L.T. is learning how to explore the internal environment, including recognising furnishings and objects within it. Additionally, he is developing the ability to walk along walls and independently navigate short routes using sound and tactile sources for orientation. L.T. is encouraged to walk independently, use his motor skills which relate to posture, balance, and coordination of movements.

L.T. has not developed an adequate sense of danger. He tends to walk without taking precautions or utilising protective techniques. He does not always walk slowly, and when navigating stairs, he tends to rely on the support of accompanying individuals.

## OVERALL AND PERSPECTIVE

### **Verification of the objectives at the end of the enabling project:**

Objectives achieved: building a positive relationship with therapists, motor autonomy; acquisition of the main parts of the body on oneself.

Objectives partially achieved: stimulate and support communicative competence; stabilize communication routines; reduce echolalic language and encourage contextual use of language verbal; monitor phonemic inventory; enrich the input and output vocabulary; favour the correct sentence structuring and increasing it; lengthen attention times within the playful context; favour the exploration of objects through a more mature and accurate way; hold up L.T. within the ludic context through the proposal of functional games and the first games symbolic and fictional; propose playful activities that develop the first cognitive and learning tasks categorization; encourage games related to the body scheme (recognition of the parts of the body on oneself and on the other, identification of the spatial relationships between the parts of the body); stimulate motor pathways and sensorimotor experiences.

Based off what has been highlighted, a revision of the ongoing Habilitation project is needed. However, the speech therapy and neuro psychomotor interventions should be kept the same with a twice-weekly frequency and an annual cycle of physiotherapy. It is necessary for the mobility and orientation intervention to be conducted outside the facility to provide didactic guidance and promote motor autonomy in the school context. Additionally, a cycle of home psychoeducational intervention is needed to support the generalisation of acquired skills in the domestic setting.

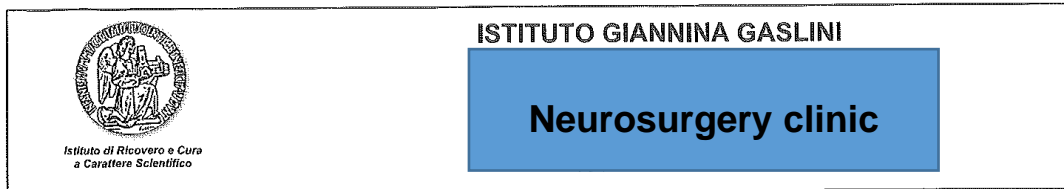
The team of the Outpatient Rehabilitation Centre

15/04/2022



# PROFESSIONAL OBSERVATIONS AND REPORTS

Simulation defined at November 2022



Patient: L.T.

Cod.

Gender: M

Date of birth: 20/04/2018

Age: 0

**Reason for access and therapy in progress:** evaluation of anterior cranial fontanel closure in low vision patient (hypermetropia +8). Negative MRI for intracranial problems

**Medical objective examination:** severe low vision with evident eye fixation deficit. Harmonic shape of the head with a punctiform anterior fontanelle. Head circumference between the 25th and 50th percentiles.

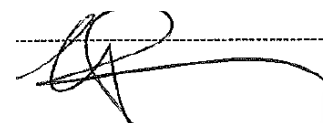
**Pain assessment:** no pain referred

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**Diagnostic and therapeutic conclusions:** no neurosurgical indications, continue according to ophthalmological and genetic indications. Requested Child Neuropsychiatry (NPI) developmental evaluation. We remain available for any need.

Date: 6/03/2019

Signature





Fondazione "Istituto Neurologico Nazionale Casimiro Mondino"  
Via C. Mondino 2, 27100 Pavia

## Developmental age neurophthalmology center

Date: 13/04/2022

Patient: L.T.

Date of birth: 20/04/2018

### Outpatient report

**Reason for access:** L.T. arrives is 3y 11months old for an NPI assessment, due to congenital amaurosis of Leber. Also performed a neuropsychomotor and multidisciplinary sensory aspects observation by NPI and neuropsychomotor therapist specialists.

### Anamnesis:

L.T. was in good general health during the last year, since last check-up (April 2021) with the exception of some infectious episodes of the upper respiratory tract.

On 21/05/20 underwent genetic counselling at our facility for suspected amaurosis of Leber. The molecular analysis of 25 genes associated with this disease has allowed us to document the heterozygous variants c.180+1G>A, c.2991+1665 A>G in the CEP290 gene. The molecular analysis was extended to the parents: the father was not a carrier of any variant while the mother was a carrier of the c.2991+1665 A>G variant. In November 2021 L.T. underwent ultrasound of the upper and lower abdomen, blood tests, urinalysis, as recommended during the last check-up at our Institute. These tests showed normal findings.

In February 2022 L.T. performed a control brain MRI (the previous one in January 2019) in which no substantial changes in the neuroradiological findings were found. Only a linear signal alteration characterized by FLAIR hyperintensity is documented in the subcortical white matter of the posterior aspect of the right superior temporal gyrus of current non-specific significance. Small FLAIR hyperintensities of non-specific gliosis significance is appreciated in the right frontal periventricular white matter and ipsilateral peritrigonal.

In conclusion substantially stable finds; necessary correlation with genetic data and clinically evaluate the need and timing of a further check.

In October 2021 eye examination at the Chiossone Institute: OO transparent cornea, deep anterior chamber, transparent lens. OOM: pendular nystagmus without obvious blocking position. Defense reaction indifferent to alternating occlusion. Visual Function OO absent visual hook, locates objects exclusively by hearing or touch. Light perception present. OO: numb direct and consensual photomotor reflex. OOF: hypermetropic small papilla, crowded. In cycloplegia high hyperopia (+9.00)

Last PEV and ERG carried out at the Gaslini hospital, on an annual basis, made in 2019 with irregular morphology.

He continued his rehabilitation at the Chiossone where he carried out two speech therapy sessions and two psychomotricity sessions a week. The cycle of physiotherapy ended in December 2021; in the same period, he started weekly spatial orientation sessions, working in the school environment.

From a motor point of view, the mother reports good orientation and movement skills in the home, independently. He searches the auditory channel a lot for communicative purposes, while less the tactile one, although more used than in the past. The family turned to a sports association for children with disabilities, where L.T. Since October 2021, he has been taking a bicycle and football course once a week, activities in which the child enjoys participating. The family would like to enrol him in a group swimming course in the near future, given L.'s excellent relationship with water.

From the point of view of autonomy L. brings the cutlery to his mouth but needs the help of the adult to remove the food from the plate with the cutlery, rides a bike with support wheels.

The language is defined as fluid, with a rich vocabulary, with good communicative intentionality.

Regarding visual behaviour, the mother reports that L. keeps his eyes more open than in the past and his head less bowed.

He currently attends nursery school three days a week with good investment and is assisted by a support teacher throughout the school hours. The relationship with the other children and inclusion in the class group is good, although the mother reports some episodes in which he would have shown himself to be spiteful with the other children. Described as a curious, lively and sociable child, able to adapt to changes. He gets along well with his older brother, they play a lot together. He loves listening to music.

Regarding the physiological anamnesis, the diet reportedly regular, adequate in quality and quantity. Sleep rhythm regular wakefulness, sleeps in his bed, in the room with his parents, falls asleep in the presence of his mother, sometimes looking for the pacifier, which he does not use at other times of the day.

### **Neurological examination:**


NPI and neuropsychomotor evaluation: under the current control, neuropsychomotor development appears overall to be in a positive evolution: L. approaches the context, less echolalic, at times also narrating his gaming experiences at home and in kindergarten: the reiterated behaviors that are still observable in moments of greater emotional stresses are more contextualized and modulated when involved by the examiner in meaningful activities; the rocking movements express excitability while the moments in which the eyeballs are pressed are observed more easily when the child "listens" or becomes alert in processing the environmental information: he prefers movement games to those in which use the hands to manipulate or discriminate with touch, which instead require more support within the enabling and educational context in order to make touch more functional for haptic purposes: typological and spatial concepts as well as action planning they still need to be promoted and supported in order to improve movement in space, also improving orientation and autonomy in movement.

With regard to the visual aspects, the occasional ability to locate a luminous target close to the face is confirmed, more easily in a semi-dark environment, the gaze is characterized by anomalous eye movements, less evident than in the previous check, misalignment of the visual axes in exotropia. The measurement of visual acuity cannot be tested even with behavioral methods (Teller Acuity Cards). Bilateral enophthalmos.

Remaining objectivity characterized by ligament laxity, muscle hypotonia, RT within the limits, bilateral flat foot (introduced corrective insole).

Notes and recommendations: a regular follow-up with complete abdomen ultrasound (especially for renal studies) and blood and urine tests is considered indicated.

It is advisable to continue the ophthalmological follow-up and rehabilitation care at the Chiossone Institute, to which reference is made, remaining available for any clarifications. NPI and development follow-up continues at our center every 8-10 months

Signature 



Patient: L.T.

Cod.

Gender: M

Date of birth: 20/04/2018

Age: 0

**Reason for access and therapy in progress:**

age: 7 months; birth: eutocic delivery

nystagmus (sent by Dott. XXXXXX)

since two months reported ocular fixation deficit for which L. performed a first ophthalmological evaluation in private regimen (Dr. YYYY) with finding of hyperopia, nystagmus and unstable fixation. Subsequent ophthalmological evaluation performed (November 2018) at our institute with finding of high hyperopia (+8.00) and prescription for optical correction.

Performed at the Ophthalmology Operating Unit (20/11/2018):  
- fundus examination with finding of probable hypermetropic pseudopapillitis

- PEV flash: tracing with irregular morphology in which the main components are barely recognizable, appearing with considerably reduced amplitude and increased latency time

- Scotopic ERG: layout with irregular morphology in which the main components are barely recognizable

- photopic ERG layout with irregular morphology in which the main components are barely recognizable

Recently performed Echo brain (9/11/2018) with normal results (small cystic formation in the left choroid plexus). Assessment of psychomotor development performed at the San Martino Institute in Genoa.

The mother reports that L does not turn away when called by name or in the presence of noise, but still seems to calm down and sometimes smiles in the presence of music or sounds. Reported presence of social smile

Feeding: breastfeeding in the morning and sometimes in the evening; weaning started with good acceptance of the proposed foods, no dysphagia reported.

Planned ENT evaluation with otolaryngologist in this Institute

Date: 26/11/2018

Signature ¥



Patient: L.T.

Cod.

Gender: M

Date of birth: 20/04/2018

Age: 0

**Reason for access and therapy in progress:**

High hyperopia and suspected severe visual impairment

**Medical objective examination:**

Scotopic ERG plot with irregular morphology in which the main components are barely recognizable

Photopic ERG plot with irregular morphology in which the main components are barely recognizable

Flash PEV plot with irregular morphology in which the main components are barely recognizable that appear to have significantly reduced amplitude and increased latency time

(see following graphs)

objective examination

OO anterior segment normal, isocoria, deep AC

pupillary reflex present, torpid uncertain with esotropia correction and slow oscillatory movements

performed PEV and ERG

OO fundus mydriasis: OO transparent dioptric means normal chorioretina, papilla with blurred margins without haemorrhages or exudates nor signs of congestion

autorefractive mydriasis +8.00 approx

**Pain assessment:** no pain referred**Diagnostic and therapeutic conclusions**

tracings compatible with severe low vision and diagnosis to be defined

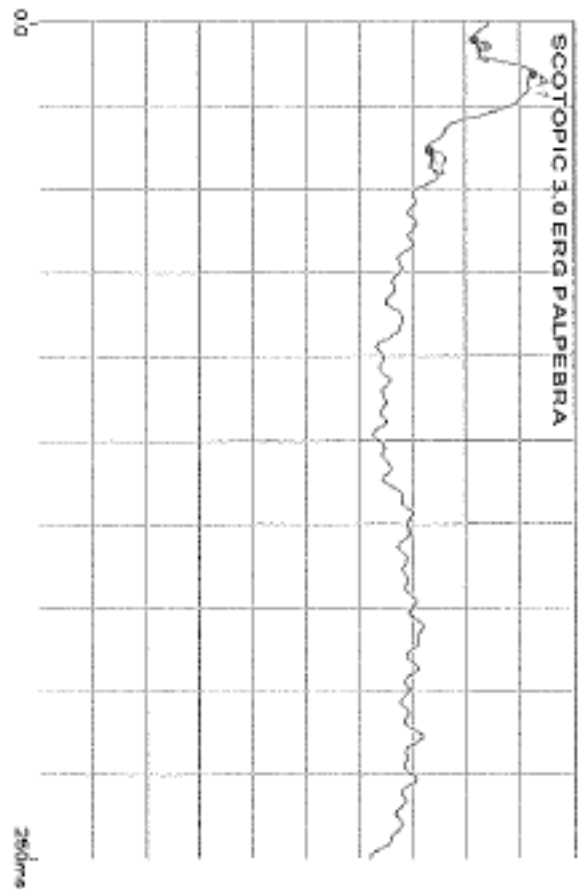
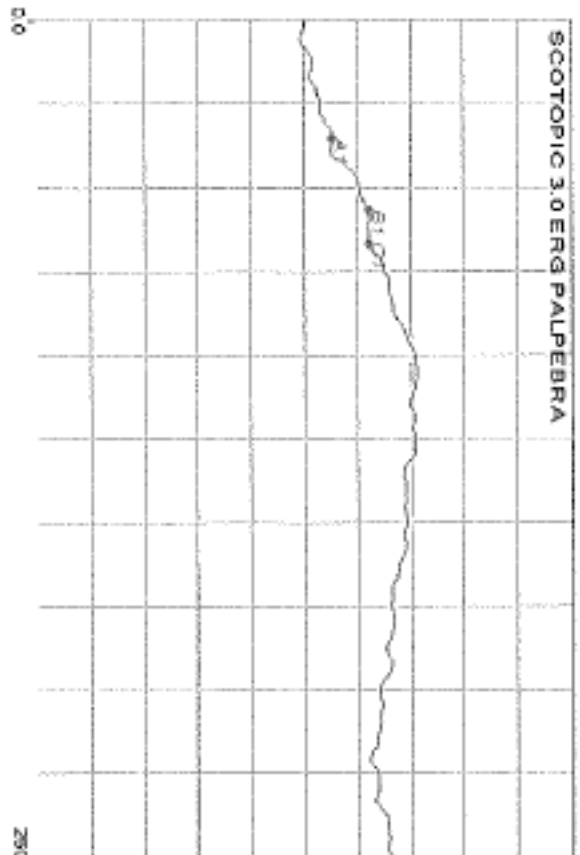
10/04/2019

Signature ¥



ISTITUTO GIANNINA GASLINI GENOVA QUARTO U.O. OCULISTICA  
 SERVIZIO ELETTROFISIOLOGIA DIR. DOTT. CARLO SBURLATI

Patient: ~~XXXXXXXXXXXXXXXXXXXX~~ Birthdate: 20/04/2018 Code: 179  
 Exam date: 10/04/2019 09:35



RIGHT - OD

	A1	B1	C1	A1-B1	B1-C1
Amplitude (uV)	2.83	20.27	20.45	11.44	0.18
Peak time (ms)	35.16	56.64	66.41	21.48	9.77

LEFT - OS

	A1	B1	C1	A1-B1	B1-C1
Amplitude (uV)	70.80	62.66	38.49	18.12	14.19
Peak time (ms)	15.14	4.88	38.09	10.25	33.20

Acq. time (ms): 250.0      Events: 5      Gain: 10000.0      High Pass (Hz): 1.0      Low Pass (Hz): 100.0  
 Flash strength (cd.s/m<sup>2</sup>): 3.000      Frequency (Hz): 1.000      Background (cd/m<sup>2</sup>): 0.0  
 Spatial form: ---      Spatial freq. (min): ---      Temp. form: ---      Temp. freq. (Hz): ---      Contrast (%): ---  
 Diagnosis:  
 NISTAGMO IPERMETROPIA 8 D

Notes:  
 ERG OO TRACCIATO A MORFOLOGIA IRREGOLARE IN CUI NON SONO RICONOSCIBILI LE PRINCIPALI COMPONENTI

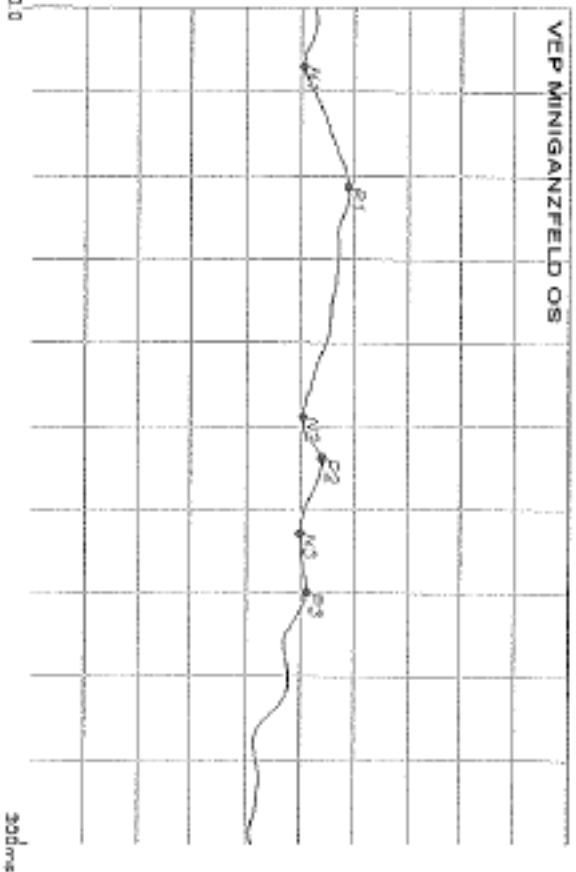
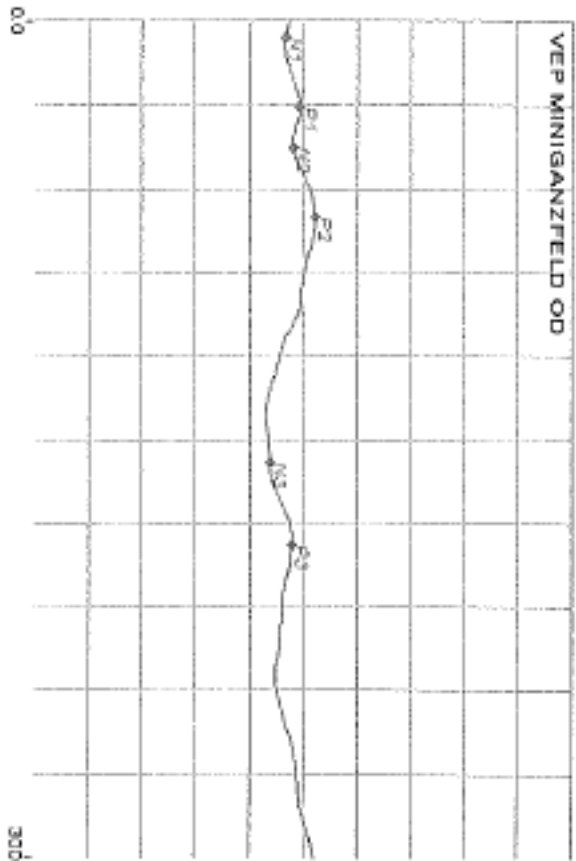
ISTITUTO GIANNINA GASLINI GENOVA QUARTO U.O. OCULISTICA  
SERVIZIO ELETTROFISIOLOGIA DIR. DOTT. CARLO SBURLATI

Patient: ██████████

Birthdate: 20/04/2018

Code: 179

Exam date: 10/04/2019 09:42



RIGHT - OD	NI	PI	N2	NI-P1	PI-N2	LEFT - OS	NI	PI	N2	NI-P1	PI-N2
Amplitude (uV)	-1.88	-0.38	-1.10	1.50	0.72	Amplitude (uV)	0.20	2.97	0.14	2.77	2.83
Peak time (ms)	5.27	30.47	45.12	25.20	14.65	Peak time (ms)	20.51	63.87	146.48	43.36	82.62

Aca. time (ms): 300.0 Events: 100 Gain: 30000.0  
 Flash strength (cd.s/m<sup>2</sup>): 3.000 Frequency (Hz): 1.000 Background (cd/m<sup>2</sup>): 0.0  
 Spatial form: --- Spatial freq. (min): --- Temp. form: --- Temp. freq. (Hz): ---  
 Diagnostic: NISTAGMO IPERMETROPIA S D Contrast (%): ---

Notes: PEV OD FLASH TRACCIATO A MORFOLOGIA LIEVEMENTE IRREGOLARE CON AMPIEZZA RIDOTTA E TEMPO DI LATENZA AUMENTATO  
 Notes: PEV OS FLASH TRACCIATO A MORFOLOGIA LIEVEMENTE IRREGOLARE CON AMPIEZZA RIDOTTA E TEMPO DI LATENZA AUMENTATO





Patient: L.T.

Cod.

Gender: M

Date of birth: 20/04/2018

Age: 0

**Reason for access and therapy in progress:**

High hyperopia and suspected severe visual impairment

**Medical objective examination:**

7/10/2019 control visit

MRI of 01/31/2019 does not show alterations of the supra and subtentorial nervous tissues, microphthalmic bulbs in OO, prescribed +8 sph that the child is not wearing

during observation in the dark, the child seems to slowly follow a light source below and in the primary position

OO slow shake nystagmus

performed cycloplegia OO +9.00 sph

**Objective examination**

OO normal anterior segment, isocoria, uncertain pupillar reflex, with esotropia correction and slow oscillatory movements

performed PEV and ERG

OO fundus (mydriasis) pallor middle retinal periphery

transparent dioptric means

autorefr +8.00, optical correction confirmed

**Pain assessment:** no pain referred**Diagnostic and therapeutic conclusions**

severe low vision with diagnosis to be defined, awaiting genetic investigations at Mondino. Control in 6-7 months

07/10/2019

Signature ¥

## References

Video 1 4y tactile

Video 2 4y walking

Video 3 4y taking space references