Case study documents DC12y

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VERSIONING DETAILS

<table>
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<th>Version of the document</th>
<th>Version 1</th>
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<tbody>
<tr>
<td>Date of delivery</td>
<td>Apr 20\textsuperscript{th} 2023</td>
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Table of Contents
MEDICAL HISTORY ...........................................................................................................3
FUNCTIONAL CLINICAL REPORT FORM .......................................................................4
PROFESSIONAL OBSERVATIONS AND REPORTS .......................................................11
References ......................................................................................................................17

Instructions
The attached documents are designed to support the analysis of cases during the Multi-Disciplinary Team discussion simulation (see Lesson Plan LO3-D-E-1 "Promoting neuro-psychomotor development within the MDT)

The layout of the Functional clinical report was modified to facilitate the distribution of documents among the participating students, along with the selected professional reports. Additionally, video resources are included and listed on the final page of this document.

Simulation placed at November 2022
MEDICAL HISTORY

D.C.  gender M
Date of birth:  29/04/2010

ICD 10 Diagnosis Code
H47 Other disorders of optic [2nd] nerve and visual pathways

ICD 9 Diagnosis Code
37710 Optic atrophy, unspecified

Notes
suspected Leber optic atrophy
Medical history and aspects

Low vision (RE 1/50, LE 1/10) in a child with neuroradiological picture with underlying septo-optic dysplasia.

D.C was admitted to our institute in 2010, where he is currently receiving visual stimulation treatment, information technology, psycho-pedagogical support, learning support and mobility and orientation assistance.

In September of last year, D.C began attending the first class of lower secondary school, where he received individualized teaching and support from an educator. Holder of legal benefits for certification L.104 (paragraph 3).

During the June 2021 check-up, it was observed that the picture remained largely unchanged, with bilateral tibial external rotation and internal rotation of both feet. However, these rotations were significantly reduced when the patient wore shoes, and there was no indication for the use of insoles. Another check-up is anticipated in one year.

FUNCTIONAL PROFILE

D.C. presents as a shy and introverted child who takes a significant amount of time to establish a harmonious and cooperative relationship with the primary caregiver. This relationship is further hindered by reduced sustained attention span. In September, after switching therapists, D.C. has exhibited communicative and verbal resistance towards the new therapist. During sessions, D.C. demonstrates passive opposition through silence and initial refusal towards proposed activities. However, with great effort and limited motivation, D.C. eventually engages in the activities, whether they involve paper-based tasks or computer-based activities.

Despite several requests, D.C. did not agree to bring his PC, which could have facilitated relationship-building and dialogue. The use of a computer tool often encourages D.C. to express himself through writing, sharing his experiences and desires. Both verbal and non-verbal communication reflect D.C.’s introverted and shy nature, which significantly impacts his relationships with peers. D.C. tends to distance himself from the group, isolating himself and finding it challenging to interact with peers. While D.C. possesses the necessary pragmatic and intersubjective skills for social interactions, there is still a need for further development in psycho-affective aspects.

2. COGNITIVE/NEUROPSYCHOLOGICAL ASPECTS

D.C.’s overall cognitive function is normal; however, it is negatively impacted by tests that rely solely on visual characteristics and require eye-hand coordination. However, when D.C is presented with visual stimuli that have the appropriate characteristics and where he does not have a time limit to process what is presented, D.C demonstrates the ability to interpret and organize visual material that enables him to formulate hypotheses that are useful for solving new tasks.

D.C.’s capacity for focused and sustained attention is near the lower end of the normal range. D.C. experiences some difficulty in maintaining attention to auditory stimuli and suppressing distractions. Additionally, storing stimuli in short-term memory and
applying strategies to modify them based on specific requests can be challenging for the child. It is difficult for D.C. to regain concentration after interruptions, such as interventions from educators, parents, or teachers. D.C. also faces difficulties in retaining the correct sequence of an oral presentation or a theoretical lesson in memory.

D.C.’s ability to hold auditory/verbal information in memory while simultaneously performing operations on it, such as simple mental calculations, is only satisfactory.

Some difficulties encountered in daily life and at school can be attributed to the difficulties described above: estimating travel times during travel, for example, or remembering a sequence of activities to carry out. The mental effort required to deal with daily cognitive activities can cause fatigue and have repercussions on emotional experiences and the quality of interpersonal relationships. D.C experiences significant difficulties with scanning speed, visual stimulus recognition and in carrying out basic paper-pencil tasks compared to his peers.

In the tests conducted for this index, visual performance is assessed through timed tasks where the stimuli are reduced in size and presented in visually “confusing” contexts, such as numbers. Consequently, the results obtained by D.C. can largely be attributed to his visual impairment as well as the fact that he did not use compensatory strategies that would have been useful for his performance.
3. COMMUNICATION ASPECTS

Aspects of communication and conversation are intricately connected to the psychoaffective and relational aspects. D.C. faces considerable difficulties in spontaneous communication, exhibiting non-verbal cues characterized by withdrawal and closed-off behavior towards others. Aspects that need to be elicited, maintained and strengthened include visual engagement, spontaneous communication, attention and adherence to social rules that are inherent in conversations.

4. LINGUISTIC ASPECTS

D.C. predominantly utilises the verbal channel as his primary mode of communication. At the phonetic-phonological level, his ability to articulate sounds is proficient. With regards to lexical understanding, D.C demonstrates good comprehension, although encounters some challenges in recalling less frequently used words.

At the syntactic level, D.C. effectively employs both simple and complex sentence structures, displaying no difficulties in understanding syntax. When telling stories, he uses coordination and subordination to link sentences.

5. PRACTICAL MOTOR ASPECTS

D.C. experiences limitations in both gross and fine motor skills, which are partially attributed to the visual impairment.

6. ASPECTS OF THE GAME

Nothing to report. Loves board games.
7. ASPECTS of LEARNING
D.C reads at a slower pace compared to his reference age and makes numerous errors due to his visual and attention difficulties. His understanding of material is affected by his lack of attention and concentration when reading long material.

Writing: D.C writes in capital letters and struggles to grip the pen properly when writing, resulting in poorly legible and unclear handwriting. He makes many errors in dictation particularly with double letters, accents, and the use of the letter 'h.'

Mathematics: D.C. successfully applies calculation methods and independently solves the required operations by utilizing his usual compensatory strategies.

8. ASPECT OF AUTONOMY
D.C. is currently in the process of learning how to use the white long cane. He has gained the ability to navigate independently across multiple blocks and recognizes the appropriate time to make the first street crossings.

He has the potential for social autonomy.

9. SOCIO-RELATIONAL ASPECTS AND THE CONTEXT OF BELONGING
The family network is large and emotionally connected, showing traits of being overly protective.
10. COMPUTER REHABILITATION:  

11. NEUROVISUAL FUNCTIONAL ASPECT  
Visus evaluation: 1/50 was found for right eye and 1/10 in left eye, that cannot be improved with lenses.  
In terms of near vision, CP (closest point) 9 has been identified at approximately 10 cm, but only with single letters. When it comes to evaluating a CP of at least 20-24, D.C., due to their closed attitude with a bowed head, never engages in distant vision.  
Regarding D.C.’s ocular motility, there is a horizontal jerk nystagmus observed without any block position. The nystagmus jerks remain unaffected during convergence. There is a pronounced anomalous head position on the right shoulder. While the tracking movements and saccades can be examined to some extent, D.Cs lack of collaboration however makes the assessment limited.  
The chromatic sense, as measured by the Color vision test, falls within the normal range. However, there is a deficiency in contrast sensitivity. Further investigation of this aspect will be conducted as collaboration increases.  

Verification of the objectives at the end of the enabling project:  
Objectives achieved:  
- Approach with the techniques of the white long cane  
Ability to move in several blocks by making the first crossings with the recording of the exact moment to make them Fair spatial awareness.  

Objectives partially achieved:  
• Reduction of errors in reading and writing  
• Improve understanding of passages in long texts  

Monitoring of the study method and the compensatory tools used by D.C:  
• Autonomous use of the tool  
• Use of speech synthesis  
• Typing  

In orientation and mobility:  
• Problem solving along the journeys made with and without the help of passersby (for now the child interacts with the O.M. rehabilitator to improve the sphere of social skills).  
• Familiarise yourself with the concepts of the external environment (pavement, crossroads, roadway, etc.).  
• Acoustic decoding and use of the vicarious senses.
Proposed objectives

It is suggested to proceed with the ongoing project, maintaining the current progress and focusing on the reduced objectives. Additionally, the following objectives will be incorporated:

> Increase communication and social skills
> Support the development of greater self-esteem and self-awareness
> Strengthen executive functions.
> Improve relationship, collaboration and verbal communication with therapists
> Often and make eye contact
> Reduce oppositional attitudes
> Improve saccadic movements

The team of the Outpatient Rehabilitation Center
16/02/2022
________________________β β β β ___________
PROFESSIONAL OBSERVATIONS AND REPORTS

Simulation defined at November 2022

Patient: D.C.  Cod.  Gender: M
Date of birth: 29/04/2010  Age: 2

Reason for access and therapy in progress:
visual impairment in nystagmus

Medical objective examination:
Visual evoked potentials (VEP)
Electroretinography (ERG, FLASH PATTERN)

Pain evaluation
No pain

Diagnostic and therapeutic conclusions:
ERG both eyes: traced within the normal limits for morphology, amplitude and latency time, both in the photopic and scotopic components

Right Eye VEP within normal limits for morphology and latency time, slight reduction in amplitude

Left Eye PEV: traced within the normal limits for morphology, amplitude and latency time

Date: 05/12/2012
Signature __________ SSSSS__ -
ISTITUTO GIANNINA GASLINI GENOVA QUARTO U.O. OCULISTICA
SERVIZIO ELETTROFISIOLOGIA DIR. DOTT. CARLO SURLIATI

Patient: C
Birthday: 29/04/2010
Code: 1989/25
Exam dr.: 05/05/2012 10:52

SCOTOPIC 3.0 ERG PALPEBRA

Amplitude deviation (SD)
-8.9

RIGHT OD
A1 4.20 29.42 29.72 18.74 12.15 12.07 12.46 79.09
Amplitude (µV):
21.00 43.48 122.07 12.46 12.65 12.46 12.65 12.46
Peak time (ms):
280 ms

LEFT OS
A1 4.20 29.42 29.72 18.74 12.15 12.07 12.46 79.09
Amplitude (µV):
21.00 43.48 122.07 12.46 12.65 12.46 12.65 12.46
Peak time (ms):
280 ms

Ass. time (min): 250.0
Flash strength (cd/s/m2): 3.000
Spatial freq. (min): —
Diagnosis:
IPVGIONE

Events:
5
Frequency (Hz):
1.000
Spatial freq. (min):
—
Temp. freq. (Hz):
—
Contrast (%): —

Notes:
ERG OD SCOTOPICO TRACCIATO NEI LIMITI DELLA NORMA PER M.
ORIFOGIA E TEMPO DI LATENZA. LIEVE RIDUZIONE DI AMPIEZZA.

Notes:
ERG OD SCOTOPICO TRACCIATO NEI LIMITI DELLA NORMA PER M.
ORIFOGIA E TEMPO DI LATENZA. LIEVE RIDUZIONE DI AMPIEZZA.
Prova di ERG fotorreceptoriale (ERG) con speciale attenzione alla risposta palpebrale (PHOTOPIC 3.0 ERG PALPEBRA).

**Amplitude deviation (AV)**

- **RIGHT - OD**
  - A1: 0.95
  - B1: 32.89
  - C1: 4.67
  - S1-C1: 27.92

- **LEFT - OS**
  - A1: -2.80
  - B1: 27.72
  - C1: 17.64
  - A1-B1: 30.51
  - S1-C1: 10.05

**Peak time (ms)**

- **RIGHT - OD**
  - 12.21
  - 34.67
  - 99.12
  - 22.45

- **LEFT - OS**
  - 64.48
  - 17.58
  - 34.18
  - 42.97
  - 16.60
  - 8.79

**Notes:**

ERG OD FOTOPICO NEI LIMITI DELLA NORMA

**Diagnosi:**

IPERVISIONE

**Eventi:** 5

**Freq. (Hz):** 1.000

**Gain:** 10000.0

**High Pass (Hz):** 1.0

**Low Pass (Hz):** 100.0

**Riflessi:**

**Temp. ris.:** ---

**Temp. freq. (Hz):** ---

**Contrast (%):** ---
ISTITUTO GIANNINA GASLINI GENOVA QUARTO U.O. OCULISTICA
SERVIZIO ELETTOFILOGIA DIR. DOTT. CARLO SBURLATI

Patient: C
Birthday: 29/04/2010
Code: 1999/20

Exam date: 06/12/2012 11:00

VEP FLASH OD 1C50EV

RIGHT - OD
Amplitude (v): 0.33
Peak time (ms): 43.95
Acq. time (ms): 300.0
Flash strength (cd/m²): 3000
Spatial form: —
Diagnosis: —

N1  P1  N2  N1-P1  P1-N2
0.33  2.42 -0.79  2.09  3.21
43.95 59.77 04.92 18.82 35.16

LEFT - OS
Amplitude (v): -3.81
Peak time (ms): 44.53
Gain: 30000.0
High Pass (Hz): 1.0
Low Pass (Hz): 30.0

N1  P1  N2  N1-P1  P1-N2
-3.81 -0.37 -3.01 3.14 2.84
44.53 66.67 85.45 19.34 24.61

Event: 100
Frequency (Hz): 1.000
Spatial freq. (min): —
Temp. form: —
Contrast (%): —

Notes:
DEV. OD TRACCIATO NEI LIMITI DELLA NORMA PER MORFOLOGIA E TEMPO DI LATENZA, LIEVE RIDUZIONE DI AMPIEZZA

Notes:
PEV OD TRACCIATO NEI LIMITI DELLA NORMA PER MORFOLOGIA AMPIEZZA E TEMPO DI LATENZA
Cognitive evaluation
01/02/2021

Surname, Name: C..........D.......... 
Date of birth: 29/04/2010

FUNCTIONAL CLINICAL PROFILE
D.C. is subjected in October 2020 to an assessment of cognitive functioning through the administration of the WISC IV scale (Wechsler Intelligence Scale for Children 4th edition).
It was decided to implement an update of the previous cognitive assessments carried out in 2016 and even earlier in 2014, in order to understand the strengths on which to focus and the weaknesses to enhance and compensate. In the meetings dedicated to administering the tests, the child proved to be euthymic and in tune with the examiner, despite the difficulty in maintaining concentration on the task and his easy distractibility.

COGNITIVE SKILLS
The WISC IV test allows for the computation of four indices: Verbal Comprehension, Visual Perceptual Reasoning, Working Memory and Processing Speed. The quantitative analysis of the index profile has a dual objective: to evaluate the cognitive abilities implicit in the execution of the tests and to determine whether some specific modes of functioning are a facilitating or hindering factor in the subject's daily life and learning. All indices have a mean of 100 and a standard deviation of 15. Each unitary index can be interpreted if it exceeds the threshold value.

WISC IV test I.Q. total calculated: 85

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<th>Regulated score</th>
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<th>Regulated score</th>
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<tr>
<td><strong>Verbal Comprehension</strong></td>
<td>94</td>
<td><strong>Visual Perceptual Reasoning</strong></td>
<td>104</td>
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<tr>
<td>similarities</td>
<td>10</td>
<td>drawing with cubes</td>
<td>8</td>
</tr>
<tr>
<td>vocabulary</td>
<td>9</td>
<td>illustrated concepts</td>
<td>11</td>
</tr>
<tr>
<td>comprehension</td>
<td>8</td>
<td>reasoning with matrices</td>
<td>13</td>
</tr>
<tr>
<td>information</td>
<td>(9)</td>
<td>completion of figures</td>
<td>(4)</td>
</tr>
<tr>
<td>reasoning with words</td>
<td>/</td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Working Memory</strong></td>
<td>85</td>
<td><strong>Processing Speed</strong></td>
<td>65</td>
</tr>
<tr>
<td>digit memory</td>
<td>9</td>
<td>search for symbols</td>
<td>/</td>
</tr>
<tr>
<td>letter-number reordering</td>
<td>6</td>
<td>cipher</td>
<td>4</td>
</tr>
<tr>
<td>arithmetic reasoning</td>
<td>(5)</td>
<td>cancellation</td>
<td>4</td>
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An analysis of the profile is performed both for each index and for the total scores, to evaluate specific modes of functioning that can be a facilitator or an obstacle in daily life. The total score of 85 is interpretable (considering the placement of index scores) but cannot be considered an accurate descriptor of the general functioning of D.C. It is more useful to evaluate a composite score: the resulting General Ability index is 99.

The General Ability index differs from the I.Q. because it doesn't directly affect performance on Memory tasks. The mental effort to deal with daily cognitive activities, even for the sequence of activities to be performed, can cause fatigue and have emotional repercussions and on the quality of relationships.

The score obtained in Processing Speed (65) in fact reveals great difficulties in scanning speed, in the recognition of visual stimuli and in carrying out simple paper-pencil tasks (compared to the performance of peers). Furthermore, the trials that make up this index are characterized by a time limit and by visually confusing stimuli (cipher); the result obtained can therefore largely be attributed to the severe visual impairment, in addition to the fact that the child does not seem to have elaborated useful compensatory strategies.

RELATIONAL AFFECTIVE AREA

Self-esteem
D presents himself as a shy and introverted child, keeps his head down and hides from the eyes of the speaker, only after having acquired sufficient confidence does greater security emerge.

Motivation
Motivation for trials requiring maintenance of cognitive effort over time is flexible; he struggles to stay in the context of the performance for extended periods of time. Difficulty maintaining concentration and easily distracted.

Relations with the adult
Very positive

Relationships with peers
Good relationships with peers

SUMMARY CONSIDERATIONS

In summary, the operating framework of the tests appears harmonious and evolving with respect to the past.

The proposed General Abilities composite score is average for chronological age and indicates the ability to listen to the question, reason and answer, express one's thoughts.

Verbal Reasoning confirms ample vocabulary and sufficient ability to use deductive and inferential reasoning, with the ability to abstract. The Comprehension score (8) indicates that the most tiring element is language related to practical, everyday situations.

As regards the Visual-Perceptual Reasoning, once the stimulus has been presented with suitable characteristics (with reference to its visual impairment) and without a time limit, it demonstrates the ability to organize the material and formulate hypotheses useful for solving tasks.
The Working Memory Index (85) is in the lower average for the chronological age. To consider the ability to maintain focused attention to auditory stimuli, inhibiting any distractors, storing stimuli in short-term memory - a task in which D finds difficulty. He shows difficulty remembering verbal information and at the same time performing operations on them, difficulty concentrating and maintaining cognitive effort for prolonged periods of time.

INTERVENTION PROPOSALS

It is advisable to reinforce, both in school and in the family environment, the positive behaviors that D adopts to learn, supporting and enhancing his autonomy. It is advisable to work on the metacognitive aspects, enhancing the ability to analyse tasks and the understanding of his own personal functioning, reflecting on the strategies to be used. It is advisable not to replace him in his homework but to help him compensate as much as possible for the visual impairment with appropriate tools recommended by therapists and by structuring attention spans. Enhancing autonomy must favor the process of growth and maturation.

The psychologist

______________________________
**hospital discharge letter**

Patient: C. D.
Gender: M
Date of birth: 29/04/2010
Admission: 23/11/2018
Discharged: 27/11/2018

**Reason for hospitalization:**
diagnostic tests in patient with optic atrophy

**Relevant Anamnestic recent notes**

In July 2018 referred nervous tic when under emotional stress, referred irritability. since then reported stability

selective feeding, eat little pasta and vegetables. regular bowel movement, polyuria and enuresis several times a week

Familiar context mother (28 y.o.) healthy, father (28 y.o.) healthy, sister (4 y.o.) healthy

eutocic delivery at term, vaccinations performed

**Medical Hystory**

at 4 months of age evidenced nystagmus and diagnosis of optic atrophy at 6 months, followed up in the ophthalmological clinic.
Recent visit with finding of bi-ocular vision 1/100 with normal ERG, unspecified VEP alterations.

The 5-year psychometric evaluation showed a slight delay.
Denies school problems unless related to the visual impairment.

In the last year, episodes of motor tics and state of anxiety. Brain MRI never done.
Regular sleep, never epileptic seizures.
July 2018 neurological examination shows a slight increase in patellar reflexes and
nystagmus, overall within the norm. Evaluation by brain MRI and panel for optic atrophy is required, a fundamental diagnosis for the certification of Rare Disease.

**Neurological admission exam:** patient attuned and cooperative, turns away if called, fair understanding of simple orders without gestural support. Known hypovisus. Apparently undamaged explorable cranial nerves. Normal tone, trophism and muscle shape. Coordination tests done correctly, not dysmetria. Absent involuntary movements. Romberg negative. Independent ambulation without pathological notes, possible ambulation on toes, heels and in tandem. Complete motor autonomy.

**Brain MRI:** performed with a 1.5T scanner and TSE, 3DT1, FLAIR, SWI and DRIVE techniques on the three axis.

Results: dysmorphism of the midline structures is documented, characterized by subtotal agenasia of the septum pellucidum (small remnant in the right posterior area). It is associated with marked hypoplasia of the optic nerves, chiasma and optic tracts bilaterally. The morphology and signal of the remaining intraorbital structures were within normal limits. It presents physiological neurohypophysial T1 hyperintensity, with pedicle in axis and of normal thickness.

The corpus callosum and anterior commissure appear normal. Adjust the morphology of the structures making up the inner ear, bilaterally, the olfactory bulbs are visible.

**Conclusions:** findings attributable to the spectrum of optic dysplasia.

Specialist endocrinological counseling and neurogenetic reevaluation are recommended.

Date: 27/11/2018

Signature _____WWWWWWW_________
Patient: D.C.
Cod. 
Gender: M
Date of birth: 29/04/2010 
Age 7

Reason for access and therapy in progress:
control visit
expected services: orthoptic evaluation and study of the visual field

Medical objective examination:

Orthoptic evaluation:

Autoref. Miotic:
RE -5,75 sph 0,25 cyl 100°
LE -2,75 sph 0,25 cyl 45°

VISUS
RE no signal
LE 1/10 (letters)

FUNDUS OO: chorioretina normal, optic nerve hypoplasia
ocular motility: fine horizontal jerk nystagmus, no blocked position

Medical objective examination:

Visual Field: manual kinetic perimetry in binocular vision (right eye without sight)
marked absolute isopteric contraction at 30 degrees temporal (left VF) and 60 degrees nasal (right VF)
contraction and left lateral hemianopsic attitude

23/07/2021

Signature AAAAAAABBBBBBBBBB
References

Video 1 12y protection indoor
Video 2 12y long cane indoor