



Social Economic Costs, Quality of Life and Disability in Patients with Cri du Chat in Italy

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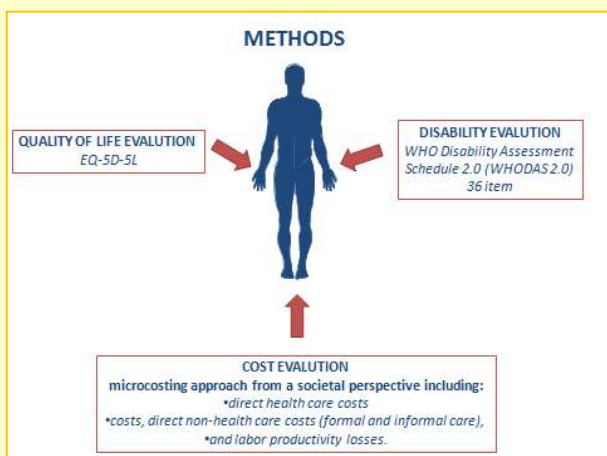
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Introduction

Cri du Chat syndrome (CdC) is a rare disease with chromosomal abnormality associated with intellectual disability and typical anatomical abnormalities. The incidence ranges from 1:15,000 to 1:50,000 live-born infants. No study at Italian and international level has assessed the costs, the impact of CdC on QoL and Disability through standardized quantitative tools.

Objectives

To estimate economic costs related to CdC from a societal perspective, to assess the QoL and Disability in patients with Cri du Chat and their caregivers in Italy.



Methodology

During 2017, we conduct a cross-sectional study of patients with Cri du Chat in Italy, recruited from the Italian Cri du Chat Association (ABC Associazione Bambini Cri du Chat Onlus). The microcosting approach was used to estimate costs from a societal perspective, including direct health care costs, direct non-health care costs (formal and informal care), and labor productivity losses. The QoL was measured with EQ-5D questionnaire and Disability with World Health Organization Disability Assessment Schedule 36item (WHODAS 2.0). The questionnaires were completed by the caregivers of patients with CdC.

Results

A total of 76 questionnaires were collected from caregivers (92% were mother/father) taking care of 42 adults patients and 36 minor patients. The mean age of patients is 19,6 years (SD=12,2) and the majority are male (68%); all patients need a carer to assist their daily activities. The principal caregiver is a family member (93%).

The EQ-5D VAS score for patients is 65.5 (SD=22.4) out of 100.

The most important compromised areas of QoL are 1) usual activities (100% reported problems in performing usual activities such as work, study, housework, family or leisure activities), 2) self care (98% reported problem in washing and dressing) and 3) mobility (92% reported problem in walking). Moderate problem is stated for anxiety and pain dimensions (figure 1). The overall WHODAS 2.0 score is 65% (where 0 = no disability; 100 = full disability), with higher score of disability for Life activities (84%), Understanding and communicating (72%) and Self-care (70%). Last, informal care is a relevant charge. First estimations show that 69% of adult patients' caregivers took them in charge from their birth and 86% of still working caregivers has difficulties to manage properly their working hours.

Concerning expenditure, first evidences show that Cri du Chat patients have a coverage similar to healthy citizens facing a high out-of-pocket quota of expenditure in rehabilitation (Table 1).

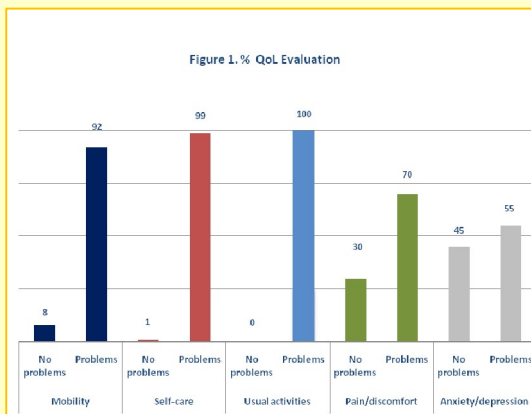


Table 1 Distribution of consumed healthcare services yearly by coverage

	Fully covered by NHS	Partially covered by NHS	Out of pocket
Laboratory exams	89%	9%	2%
Diagnostic exams	91%	7%	2%
Specialistic visits	44%	43%	13%
Rehabilitation	38%	3%	59%

Conclusion

Preliminary results highlights the burden of CdC in terms of its impact on QoL and Disability for patients and caregivers in Italy, with score much lower than that of general population. The disease is associated with considerable costs of informal care. Further patient and caregiver outcomes are under development.