ECRD: THE EUROPEAN CONFERENCE ON RARE DISEASES & ORPHAN PRODUCTS - 10/12 MAY 2018 - VIENNA

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Ask and you will receive (Matthew 7:7) The experience of A.B.C. (Associazione Bambini Cri du chat) Italy.

It was April 2016 when some articles about costs and quality of life were published on Eur J Health Econ, a sort of "provisional report" of BURQUOL-RD. The main aim of BURQOL-RD was to develop a disease based model (instrument) to assess the impact of health policies, interventions and treatments in the field of rare diseases (RDs) quantifying the Economic Burden and Health-Related Quality of Life (HRQOL) for patients and their caregivers, from a macro societal perspective. Ten RDs were targeted in the pilot study: Cystic Fibrosis, Prader-Willi Syndrome, Haemophilia, Duchenne Muscular Dystrophy, Epidermolysis Bullosa, Fragile X Syndrome, Scleroderma, Mucopolysaccharidosis, Juvenile Idiopathic Arthritis and Histiocytosis. For the first-time scientists tried to face the problem of costs of a rare disease. Not only direct costs, nor only health related costs, but also social, indirect and intangible costs.

For the first-time patients and caregivers were asked to answer questions about their health problems and to weigh the impact of the disease on their quality of life.

The cost-of-illness (COI) approach was used. Information about the patients' and caregivers' HRQOL was estimated using the generic EQ-5D questionnaire; besides, the patients' disability was assessed through the Barthel Index and Whodas 2.0.

The diseases studied were different one form the other, and their prevalence was quite high, but we thought to ask the ISS (Istituto Superiore di Sanità=National Centre for Rare Diseases, Istituto Superiore di Sanità (ISS) Rome Italy) if it could be possible to figure something similar tailored on our problem: The Cri du chat syndrome.

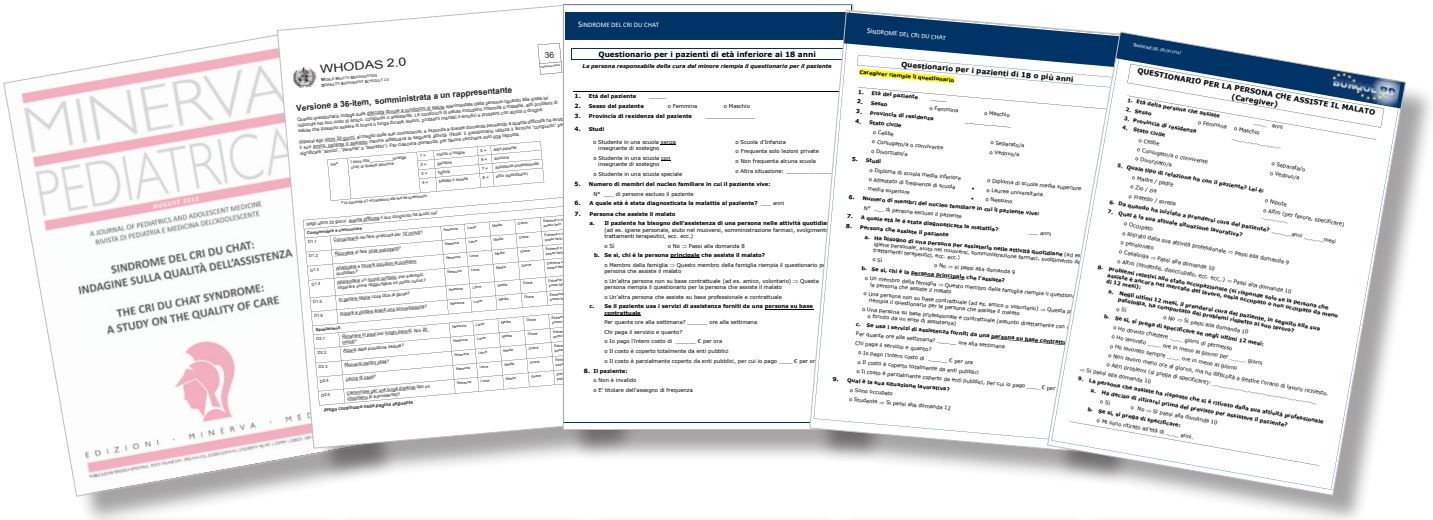
It took time to adapt questionnaires without altering them (so that the comparison with "the original" could be possible) and to convince families to fill them (even if boring and time wasting), but in the end we obtained a pretty high participation and a huge amount of data which are now being processed by economists.

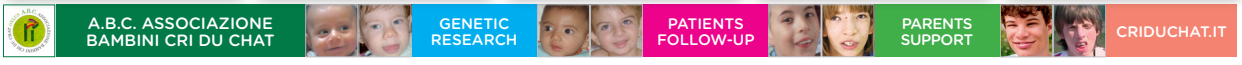
Already in this ECRD we have the possibility to read preliminary results, which will be precious to allocate resources and help policy makers and other stakeholders to plan wisely.

Our Association had already published in 2012 an article on Minerva Pediatrica on "*how much having a child with CdC Syndrome had modified parents life*" (*), but this new approach takes into account all different aspects of Quality of Life, and the answer comes from caregivers (very seldom Cri du chat patients are able to fill questionnaires themselves), not from "professionals". We want to point out that even "small" RD can obtain tailored investigations if the proper path is found.

RD require the combined efforts of health and social care professionals, politicians, managers and researchers to increase the availability of effective disease management tools to improve care and to extend both life expectancy and Health Related Quality of Life (HRQOL).

We thank ISS for their sensibility and hope to have opened a new form of collaboration between associations and institutions, patients and doctors and policy makers. **Just asking....**





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