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Short Communication

Oral Manifestations in Rare Diseases

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The complexity of the pathologies that characterize Rare Diseases (RD) requires a multidisciplinary approach. A significant number of rare diseases, especially the syndromic forms, are associated with oral manifestations that have a very broad spectrum of severity [1-4]. Dentistry is almost systematically involved in both diagnostic procedures and specific treatments, which makes it essential to have the presence and specific expertise of dentists in multidisciplinary groups dealing with rare diseases.

Main issues in a hypothetical project should be:

1. Identification and classification of oral manifestations in the most common rare diseases in the Italian pediatric population.
2. Collection of data for epidemiological and statistical purposes (incidence, prevalence).
3. Drafting of protocols and guidelines for clinical use.
4. Establishment/consolidation of a multidisciplinary group within pediatric hospitals in which the dentist plays a transversal role, of diagnosis, treatment and timing, connection/suggestions for further investigations, continuous discussion of individual cases, publication of case histories on the events oral in rare syndromes required for observation.

The rare diseases are a complex of over 5000 pathologies that represent 10% of the entire human pathology. [1-20].

The European Commission in 2001 decided to consider rare those pathologies whose incidence does not exceed 5 cases per 10,000 inhabitants [1-4].

Overall, 80% of rare diseases (about 4000), are genetic in origin, while the remaining 20% are acquired.

The RD have some characteristics in common:

a) Chronic and disabling character, which constitutes an important social problem

b) Difficulty of diagnosis

c) Difficulty of treatment: Often there are no effective therapies, no targeted drugs are available (due to the limited market, there is little interest in developing the research and production of so-called orphan drugs).

d) Limited number of specialized centers for diagnosis and treatment

e) Difficulties of identification and access for patients

f) Fragmentation of patients between centers, which is an obstacle to controlled clinical trials and consequently to therapeutic innovations

Since a large part of rare diseases are of genetic origin, most of them occur in the pediatric age. The Pediatric Hospitals are, therefore, the natural locations for the realization of finalized projects.

In 1997 Orphanet was born in France, a European project that aims to help improve the management and control of rare diseases and those without precise diagnosis ("orphans"), offering updated information, written by professional experts, written in understandable form, able to adapt to the needs of patients and their families, doctors and researchers, associations and industry. The Orphanet project has effectively created a virtual and integrated network of information, services and data relevant to rare diseases. The network includes France, Italy, Austria, Belgium, Germany, England, Holland, Portugal, Ireland, Greece, Spain, Switzerland, Hungary, Cyprus, Finland, Romania [1-4].

Orphanet has a directory that contains a database on rare diseases and orphan drugs in 6 languages, available online.

Genetic-based RD are divided into syndromic and non-syndromic.

The oral manifestations in rare diseases, in turn, can be part of specific syndromes or represent complications of an underlying disease or undesirable effects of therapies [1-20].

The number of syndromes with oro-dento-facial manifestations is enormous. A comprehensive discussion of these syndromes is found in The Oxford Monography on Medical Genetics No. 19 [2]. The syndromes are divided into chromosomal, genetic, metabolic and classified according to symptom prevalence criteria.

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