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Theoretical basis for a new approach of studying Emery-Dreifuss muscular dystrophy by means of thermography

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ABSTRACT

Introduction: Emery-Dreifuss muscular dystrophy (EDMD) is a clinical condition characterized by neuro-skeletal and cardiac impairments. By means of thermography, an image acquisition technique that allows the recording of the heat emitted by objects or bodies, news insight can be obtained insights about the evaluation and follow-up of this disease. Actually, musculoskeletal disorders are a major cause of counseling and access to rehabilitation services and are some of the most important problems that affect the quality of life of many people. There are urgent both clinical and research needs for the assessment and follow-up of patients with Emery-Dreifuss muscular dystrophy and the thermography is a rapid, non-invasive, easy to use and objective technique that analyzes the temperature of the examined tissue.

Hypothesis: The main aim is to offer a new possible hypothesis of validating the thermography techniques that support the evaluation and clinical follow-up of the Emery-Dreifuss dystrophy. To carry out this work we rely on the evidence of the existing bibliography. To perform this work and to evaluate the current situation on this topic, a systematic review was carried and after the application of an automatic and manual filter, inclusion and exclusion criteria, a total of 0 articles was obtained. Unfortunately, there is a lack of articles that relate the use of thermography in the Emery-Dreifuss muscular dystrophy. Due to the absence of information, we have expanded the search to articles concerning the use of thermography in relation to alterations of the musculoskeletal system compatible with those of Emery-Dreifuss, genetic diseases related to the X chromosome and more generally muscular atrophy. Based on other studies and results carried out in diseases that show signs and symptoms similar to Emery-Dreifuss Muscular Dystrophy, we believe that a new line of translational research could be opened with novel findings and we think that thermography could be an optimal tool for the clinical monitoring of this pathology. We believe that it would be of a great importance to carry out an observational study, to lay the foundations for future works, that relate thermography to the Emery-Dreifuss muscular dystrophies.

Introduction

Emery-Dreifuss muscular dystrophy (EDMD; MIM 310300) is a chronicle and degenerative clinical condition characterized by:

- Contractures and muscular stiffness of the tibialis, elbows and rachis, which begin in childhood.
- Initial loss in a correct Humero-peroneal articular distribution.
- Cardiac involvement as palpitations, poor exercise tolerance, heart failure, pre-syncope and syncope. [1,2].

With the term “thermography”, we define an image capture technique that allows us, by means of recorded heat, to interpret and understand different physiological and/or pathological states [3,4] (Fig. 1).

Considering that many of the muscular or articular alterations are related to changes in density, volume and temperature in the affected anatomical areas, and consequently mainly in the tegument of those areas, it is easy to deduce the reason for investigating the integumentary system throughout the use of thermography [5,6].

Despite the musculoskeletal system is fundamental for the

Abbreviations: BAT, brown adipose tissue; BVS, biblioteca virtual en salud; EDMD, emery-dreifuss muscular dystrophy; MIM, mendelian inheritance in man; MR, magnetic resonance; PET-CT, positron emission tomography/computed tomography; PRISMA, preferred reporting items for systematic reviews and meta-analyses; SSR, sympathetic skin response; XHED, hypohidrotic dysplasia

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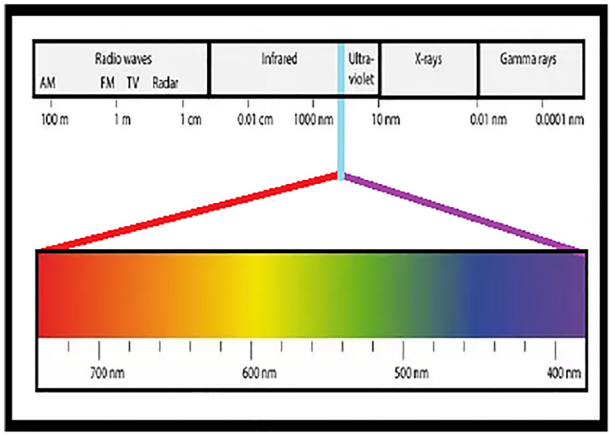

Fig. 1. The radiation recorded by the thermographic camera consists of the capture of long-wave radiation. This waves are represented in this electromagnetic spectrum image. *Figure from Tipler & Mosca, "Physics for Scientists and Engineers", 6th edition, p. 1041.*

functionality of everyday life activities, it has not been the subject of thermography in-depth studies and, indeed, the few publications available on the muscular system all relate to sport and physical activities [7–9].

Treatments received for musculoskeletal disorders during hospitalization services and subsequently in rehabilitation centers, normally vary from an urgent check by a physician in order to exclude possible injuries throughout physical and instrumental examinations, up to a scheduled or continuous follow-up to evaluate the general conditions of the culprit lesion [9,10].

From a research by N Carboni et al. [11] having as its object a family with 5 patients affected by the recessive form of the muscular dystrophy linked to the X Emery-Dreifuss (mutation of the *STA* gene), it has been shown through magnetic resonance (MR) the muscular involvement of the soleus, the medial portion of the gastrocnemius, and the muscles of the thigh in the onset and progression of the EDMD, which until then had never been registered [11].

Since thermography, appears a very useful method in the early stages of medical diagnosis, it allows a simple, rapid and non-invasive evaluation [12] and EDMD is a pathology directly associated to changes in the muscular and articular system [1,2], we believe that the realization of this research is justified in order to better clarify other aspects of this pathology.

The objective of this work is to offer a new idea that supports the hypothesis of validation of thermography techniques in the evaluation and clinical follow-up of Emery-Dreifuss dystrophy.

To carry out this work, first, a systematic bibliographic review was conducted between September 2017 and February 2018, in which the PRISMA recommendations were followed [13] (Table 1).

In order to guarantee a depth investigation and the highest number of the gathered records, several searches were made in all databases combining different logical descriptors.

After the first search, a total of 213,968 articles were obtained. Filters, inclusion and exclusion criteria have been applied to all research examples.

The inclusion criteria (automatic) were: presence in the title/abstract of the terms thermography and/or Emery-Dreifuss and no restrictions of gender and ethnic group.

The exclusion criteria (manual) established were: articles at the preliminary stage, without conclusion and/or interpretation of the results and Pedro scale lower than 6 and Jadad less than 3 [14].

Automatic filters were: randomized clinical trials, systematic reviews, meta-analysis, human, language, last 10 years.

After the application of automatic and manual filter, inclusion and exclusion criteria, a total of 0 articles were obtained.

Due to the lack of information, we have expanded the search to articles concerning the use of thermography with alterations of the musculoskeletal system compatible with those of Emery-Dreifuss, genetic diseases related to the X chromosome and muscle atrophy more generally.

Proposal and justification of the hypothesis

In regards to the musculoskeletal system, we will define it as previously proposed as the whole of the osteoarticular and muscular system. As regard to the osteoarticular system, we refer to bones, joints and ligaments, while pertaining the muscular system, we refer to the muscles and tendons. [15–17].

According to a meta-analysis published in 2013 on the use of thermography for the diagnosis of injuries of the osseous, muscular and joint system, the use of this technique has not proved to be useful for the diagnosis of stress fractures [18].

Nevertheless, it should be emphasized that the sample of the analyzed articles (3 articles) was very small and so far stress fractures do not present themselves as a typical clinical condition of the diseases related to the emerins (protein complex involved in Emery-Dreifuss diseases).

In fact, according to the current bibliography, the issues of the osteoarticular system in patients with EMDM, do not depend on the bone structure itself, but on the specific deformations of the articular segments typical of this pathology [1,2,11].

The most evident cases of articular deformations recorded, all involve vertebral column, with scoliosis and/or rectifications that affect the whole vertebral spine. G Kwok et al. (2017) used thermography in patients with idiopathic scoliosis and showed differences in temperature between the concave and convex sides of the regions of interest. These differences according to the authors could be due to a greater emission of infrared radiations of the convex side of the observed area [19].

However, we must stress that: A) a relatively small sample of patients is used and; B) the spinal alterations of patients with EMDM are directly related to the genetic alterations present on the X chromosome and are therefore not of an idiopathic nature.

The use of thermography in X-linked genetic disorders such as hypodriotic dysplasia (MIM 305100; XHED causative gene) showed an abnormal distribution of cutaneous temperature derived from the altered peripheral vascular perfusion (RP Clark et al. in 1990) [20].

Although the genes involved in this pathology are different from those of the EMDM, both are linked to the X chromosome and present peculiar syndromic features.

In fact, on the one hand the hypohidrotic dysplasia (XHED) presents dysmorphic signatures at the level of teeth, forehead, lips, nose, skin and sweat glands and on the other hand the Emery-Dreifus (EDMD) shows them in the rachis, elbows and muscles of the distal part of the lower limbs the internal neuromuscular system being compromised [11,20].

In neuromuscular diseases, thermography is used as a diagnostic and monitoring tool for patients with nervous system disorders. Sudek syndrome and cerebrovascular accidents are the areas where thermography has more room in this medical specialty, however, EB Neves et al. (2015) argue that this tool has great potential for diagnosis in many other neurological diseases [21] (Fig. 2).

These facts are supported in another very recent study, by B Hegedus (2018) who described that the totality of patients with stroke had a microcirculation and function in the area affected, presenting differences in temperature between the healthy side and the pathological side [22].

This temperature difference has changed after a physiotherapy program highlighting a strong correlation between the improvement of the joint function and the change in temperature [22].

Although we are aware of the different nature between the