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A collaborative clinical analysis service based on theory of evidence, fuzzy linguistic sets and prospect theory and its application to craniofacial disorders in infants

Arcangelo Castiglione *, Raffaele Pizzolante, Christian Esposito, Alfredo De Santis, Francesco Palmieri, Aniello Castiglione

Department of Computer Science, University of Salerno, Via Giovanni Paolo II, 132, I-84084, Fisciano (SA), Italy

HIGHLIGHTS

- A collaborative clinical analysis framework that can be implemented as a Grid or Cloud Service.
- Performs detection and characterization of pathologies by combining the knowledge of a group of experts.
- Combines prospect theory, theory of evidence and fuzzy linguistic sets for developing a computer aided diagnosis tool.
- We considered craniofacial pathologies in infant population as a practical example for better explaining the proposed solution.

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ABSTRACT

Nowadays, it is more and more important to diagnose several kinds of pathologies at their early stage, in order to take the necessary countermeasures before having permanent consequences. Unfortunately, though many pathologies are widespread, there does not exist a unique standardized reference or *Gold Standard* according to which it is possible to evaluate the patients, mainly when the pathology is in the early stages or is not very noticeable, and the doctor is not sufficiently expert in the problem domain. In this work, we deal with this problem, by envisioning new healthcare services supporting a collaborative clinical analysis of symptoms collected from the patients and forwarded to a group of experts, which are geographically distributed. The experts return back their assessment and diagnosis and the system combines these by means of the Theory of the Evidence, in order to provide a single response. The above services can be easily implemented on top of state-of-the-art distributed computing facilities such as Grids or Clouds, providing a connected environment for medical data distributed over different sites and allowing medical experts to collaborate without being co-located, thereby providing transparent access to data and computing resources. Additionally, such services can provide feedbacks to each expert, in order to improve its own knowledge and experience in the case of divergence between the expert response and the global combined diagnosis in recognizing and classifying the received symptomatic indexes from the patient. We have considered the craniofacial pathologies in infant population as a practical example for better explaining the proposed solution.

1. Introduction

Currently, the research community in health informatics is focusing its attention on approaches to support doctors and general practitioners in their job of recognizing pathologies from a series of evidences collected from the patients. The intent is to reduce the probability of misdiagnosis, which is currently quite high *i.e.*, it has been estimated that between 10% and 15% of medical diagnoses are incorrect, and implies high costs for the healthcare systems, *i.e.*, a recent analysis published in *BMJ Quality and Safety* estimated that malpractice claims in the last 25 years had an average price of $386,849 per claim and caused 40,000 to 80,000 hospital deaths yearly in the United States [1]. The problem of misdiagnosis is more serious if we consider that some pathologies can affect infants and leave them permanent deformations and/or

* Corresponding author.
E-mail addresses: arcastiglione@unisa.it (A. Castiglione), rpizzolante@unisa.it (R. Pizzolante), christian.esposito@dia.unisa.it (C. Esposito), ads@unisa.it (A. De Santis), fpalmieri@unisa.it (F. Palmieri), castiglione@ieee.org (A. Castiglione).

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health problems, which can be avoided with the early diagnosis of the pathologies and by assuming proper corrective, even simple, actions at the early stage of the pathology. A practical example of this is represented by craniofacial pathologies, such as *plagiocephaly*, *brachycephaly* and *scaphocephaly*, which are very common and widespread among infants, due to either wrong sleeping positions or to congenital problems. The deformation caused by such pathologies, if recognized early, can be smoothed out by simply placing the baby prone while awake or alternating the point of contact between the occiput and bed during sleep [2].

To avoid potential patient harm, reduce the costs caused by misdiagnosis and improve the probability of early diagnosis of pathologies in infants, hospitals and medical practices are turning their attention to clinical *Decision Support Systems (DSS)*, in order to assist them in diagnosing pathologies based on the symptoms manifesting in the patients. Generally speaking, DSS operations are based on the theory of the pattern recognition: the practitioner provides a series of inputs (acquired from patients with different methods), which are compared in a proper manner with a set of labeled data stored in the system, in order to provide as an output the label of the patterns more similar to the inserted ones. Therefore, the effectiveness of the DSS, *that is to say*, the response is exactly the ones we were expecting based on the provided inputs, strongly depends on the quality of the labeled patterns internally stored in the DSS, which is commonly denoted as *Gold Standard* (GS). However, a perfect GS, *i.e.*, the one that allows the DSS to always provide the correct answer based on the provided input, is impossible to be built, since in the healthcare practice there are no strict rules for the disease diagnosis. For example, in craniofacial pathologies, a GS would be a shape which best characterizes the cranial morphology of clinically healthy patients. In addition, the adopted diagnosis methodology substantially relies on subjective assessments, performed according to the skills and experience of the single doctor who carries them out. Specifically, craniofacial pathologies are recognized based on some indexes, such as *Cephalic Index (CI)*, *Oblique Cranial Length Ratio (OCLR)*, *Standard Deviation (SD)*, etc. Such indexes are not very indicative, especially for some mild cases, thus they do not provide the doctor with an immediate and significant support to detect and quantify the pathology, especially in the case of brachycephaly and positional plagiocephaly [3].

In order to address the above issues, we define a methodological framework for the collaborative clinical analysis of a patient, which involves the acquisition of data, along with the detection and characterization of pathologies by a group of experts, geographically scattered over the world. Such a framework, due to its multi-disciplinary and inherently distributed nature, can be easily implemented on top of state-of-the-art distributed computing facilities such as Grids or Clouds, providing a connected environment for medical data distributed over different sites and allowing medical experts to collaborate without being co-located, thus ensuring transparent access to patient records and biomedical knowledge and to computing resources needed for analyzing them. The resulting architecture, which natively provides services dealing with virtualization of distributed data regardless of their location, is able to support geographical transparency by allowing seamless access to the highly heterogeneous resources available in the biomedical domain, as well as to allow the distributed execution of complex analysis tasks, for the development of collaborative diagnoses and for sharing the knowledge between multiple medical centers.

Specifically, we have applied such a collaborative analysis methodology to the problem of early recognition of craniofacial pathologies in infants, but the methodology is generic enough for being applied to several types of disease. In detail, the first stage is the acquisition of data upon with the collaborative decision making works, in order to reach a diagnosis. A set of significant cranial features can be obtained by using thermoplastic material strips to acquire, in a very accurate manner, the contour of the skull. It is important to point out that such acquisition method turns out to be low cost, portable, deployable and non invasive. Subsequently, the patient's clinical data need to be digitalized and characterized by proper sets of points. Such digital inputs can be finally distributed within the identified group of experts by using proper interfaces and communication facilities.

The focus of this work is not on the means for distributing or accessing such data, which can be accomplished by using many mature interfaces, protocols and solutions available on modern grid or cloud platforms, but on the collaborative analysis and decision making methodologies used to implement an effective diagnostic service. More precisely, we combined Prospect Theory, Theory of Evidence and Fuzzy Linguistic Sets for developing a computer-aided diagnosis tool available over a Grid or Cloud through specific service interfaces (e.g., those provided within the Service Oriented Architecture framework). It is important to point out that the Theory of Evidence can be employed even in conflict resolution and context reasoning scenarios [4,5]. In detail, when an expert, which can be a doctor or a healthcare provider, receives the digital inputs of the cranial feature measurements, a proper recognition process is carried out according to the GS locally created by the expert from a training set of clinically healthy patients. We remark that the shape of such a local GS is conform with the one reported in the medical literature [6], as well as with doctor visual assessment and CI checking [7]. Any significant deviation with respect to the GS most likely denotes an anomaly in a given anatomical region of interest. In this work, we propose the method running at each expert side, which, besides being able to detect and quantify local deformations of the cranial shape (e.g., kinking, flattening and bumps), also detects other types of malformations, e.g., bilateral asymmetry, ear deviation and cranial elongation. It is important to emphasize that the method we propose also detects malformations that are highly symmetrical and in a mild form, such as the brachycephaly. Furthermore, we remark that our method may extend and improve the conventional analysis of cranial malformations based on the *Cephalic Index* (*CI*), which can take place in a collaborative manner. We also propose a method for the clinical characterization of eventually detected anomalies. In detail, our approach is based on morphological deformations which characterize a given pathology. We use this approach since it may happen that some pathologies do not occur in the canonical form defined in the medical literature, and a given patient may be simultaneously affected by several different pathologies, eventually mixed among them. Therefore, limiting the classification to the pathologies commonly defined in the literature is not very useful for analyzing malformations which do not fall into the common case histories. After running our detection approach, each expert has performs a multi-criteria assessment of the received input, where a digit and a severity degree have been associated to each assessment criterion for the considered pathology. Then, the multi-criteria assessment is returned back at the patient side and properly combined by using the Theory of Evidence, in order to obtain a single diagnosis concerning the kind of pathology affecting the patient. Such an outcome may be send back to the expert, in case of its wrong assessment, in order to provide a feedback for improving the expert GS (*i.e.*, the cranial features wrongly recognized as abnormal are stored in the GS so as to never repeat the misdiagnosis again in the future). We have used prospect theory in order to assess the consensus degree of a decision maker with the majority and to adjust its weight when participating in future collaborative decision making. Finally, we have used Linguistic Fuzzy Set to model the qualitative assessment of human experts and to combine them with the

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