


Chapter 2

AI-Powered Thermal Imaging for Early Detection of Down Syndrome in Children

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ABSTRACT

The early detection of Down syndrome in pregnant women is crucial for the appropriate care of the affected child, as it is a lifelong condition. Karyotyping and physical examinations are the foundation of traditional diagnostic techniques, which are intrusive, time-consuming, and inappropriate for mass screening. Furthermore, the existing automated systems' poor precision, specificity, and resilience result in incorrect positives and false negatives. The study describes a system that supports or enhances these techniques for monitoring individuals with Down syndrome by using deep learning (DL) algorithms to classify thermal images and recognize even the smallest temperature changes on the skin. For feature extraction and classification with high precision and recall, the model uses Convolutional Neural Networks (CNNs). According to a comparative performance review, the proposed system performs noticeably better than several other approaches, with an accuracy of 96.3%, precision of 95.8%, recall of 96.7%, and an AUC of 0.98.

DOI: 10.4018/979-8-3373-6796-5.ch002

INTRODUCTION

The quality of life for children with Down syndrome will be improved by early management and appropriate intervention; therefore, life-leading clues are crucial and achievable in the early stages of life. An extra chromosome, specifically chromosome 21, causes Down syndrome and leads to both physical and intellectual disabilities (Bose et al., 2024). Patients with the condition require early diagnosis and ongoing medical treatment. Testing for Down syndrome using traditional methods such as karyotyping, amniocentesis, and physical examinations is not the best option because it is invasive, labor-intensive, slow, and unsuitable for mass screenings. Few healthcare facilities provide these traditional, frequently risky diagnostic techniques. Consequently, early detection of Down syndrome is uncommon in many regions worldwide, especially in nations with limited resources. Improving outcomes for children with Down syndrome requires timely identification of existing diagnostic instruments and protocols. Therefore, it is imperative to develop scalable and non-invasive diagnostic methods to accurately and promptly discover Down syndrome. Thermal imaging is a camera type that can quantitatively measure the skin's surface temperature by detecting infrared radiation. Therefore, researchers have extensively studied the technology for medical diagnostic applications. Anthropomorphism and physiology naturally cause temperature changes in the human body, even in hereditary abnormalities like Down syndrome. Variations in blood circulation and other physiological factors may affect children's skin surface temperature. By detecting these temperature changes, thermographic images can provide local heat differentials that can be used to assess a child's developmental status (Sony et al., 2024).

The exponential growth of thermal technology by AI and DL has solved the issue of manually collecting and analyzing vast amounts of data and making precise diagnoses. CNNs, a DL model frequently used for image classification, were shown to be able to identify patterns in thermal images. A trained CNN was also able to differentiate between recordings that show early indicators of Down syndrome. The requirement for an early detection method for Down syndrome that is sensitive, specific, non-invasive, and economical and that can be used in clinical and large-scale population settings serves as an argument for the study. The cost-effectiveness, ease of use, and speed of current diagnostic techniques are insufficient for widespread adoption. Accessible instruments are also needed to bring these technologies to areas that aren't well developed or are far away, where targeted capabilities, benchtop diagnostic testing, and consumable diagnostic testing aren't possible (Anala et al., 2024).

AI-enabled thermal imaging could provide an expedient, affordable, and non-invasive substitute for traditional diagnostic techniques. Together with DL techniques, thermal imaging has the potential to significantly enhance early detection of Down

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