



TRABALHOS CIENTÍFICOS: POSTER

P-045 - GENERATIVE ARTIFICIAL INTELLIGENCE AS A TOOL FOR LEARNING GENETIC SYNDROME FACIAL GESTALT FABIANO POSWAR (HOSPITAL DE CLÍNICAS DE PORTO ALEGRE)

Introdução: A clear pattern of facial features is well-established for many Genetic Syndromes (GS), allowing Medical Geneticists and other healthcare practitioners to quickly hypothesize a GS based on specific facial dysmorphisms, even with incomplete information. Generative artificial intelligence (AI) has emerged as a powerful technology capable of generating new content, such as text, images, and music. Unlike AI models focused on classification or prediction, generative AI models utilize existing data to create content. One such model is Stable Diffusion, an open-source generative AI model.

Objetivos: To assess the ability of Stable Diffusion to generate representative images of people with selected recognizable syndromes.

Metodologia: A total of 20 syndromes with a recognizable facial gestalt were selected for this study. All syndromes are classical conditions included in the textbook Smith's Recognizable Patterns of Human Malformation. Images were generated using the Stable Diffusion model using an internet resource, without providing an additional training set. Accuracy of the generated photographs was assessed using Face2Gene (FDNA INC), regarding both the position of the syndrome among the suggested syndromes (top 1, top 10 or top 30) and the Gestalt score (high, medium or low).

Resultados: Stable Diffusion model was able to generate high quality images for Down and Rubinstein-Taybi syndromes (both top 1, with a high and medium gestalt score, respectively). For other 6 generated images, the syndrome in the text was either in the top 10 (n=4) or top 30 (n=2), with either medium (n=1) or low (n=5) gestalt score. The syndrome suggested in the text was out of the top 30 suggested diagnoses by Face2Gene for the other 12 syndromes (60%).

Conclusão: The ability to recognize Genetic Syndrome gestalt is an expected competence for doctors specializing in Medical Genetics and Genomics. It saves time and reduces costs by optimizing clinical assessments and minimizing the need for expensive genetic testing. Traditionally, residents rely on illustrations, published photographs, and online databases to supplement their training. However, there is a scarcity of available material, particularly for ultra rare conditions. Generative AI has the potential to address this issue by generating new content. Similar approaches have been successful in the Radiology field, especially after domain-adaptation of the AI model. While this study found low accuracy in generating images for most syndromes, it already demonstrates the algorithm's ability to generate high-quality images for some selected syndromes. Providing additional training sets, and fine-tuning the model for this purpose is expected to yield improved results.