

esquerda e do 4° e 5° pododáctilos do pé esquerdo. A ecocardiografia constatou tetralogia de Fallot, com aorta cavalgando o septo em 50%, e comunicação interatrial. O resultado do cariótipo foi masculino, apresentando trissomia livre do cromossomo 13 (47,XY,+13) (sem evidência de mosaïcismo), compatível com diagnóstico de SP. O paciente foi a óbito aos 9 meses de vida devido a complicações de uma broncopneumonia. Conclusões: A SP caracteriza-se por um quadro clínico amplo. Contudo, existem achados que se destacam e que muito frequentemente levam ao seu diagnóstico. Estes incluem a micro/anofthalmia, a fenda labial bilateral/palatina e a polidactilia das mãos e/ou pés. Chama atenção em nosso paciente que ele não apresentava nenhum deles, o que dificultou a sua identificação. Este relato salienta que pacientes com a SP podem ter apresentações atípicas, diferentes da tradicional.

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### **BRAZIL BY THE NUMBERS: AN EVALUATION OF CRISPR/CAS HEALTH RELATED PUBLICATIONS**

CATEGORIA DO TRABALHO: PESQUISA

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CRISPR/Cas is being increasingly used for various applications, drug screening to functional genomics, in addition to therapeutic option. However, new technologies are introduced at different pace and purpose in different countries. In this study we perform a literature review on the progress of research using the CRISPR/Cas system in Brazil, focusing on health related applications. The PubMed database (<https://pubmed.ncbi.nlm.nih.gov>) was used to identify relevant articles about gene editing with CRISPR/Cas and patents were searched in the INPI (National Institute of Industrial Property) database, January 2010 to January 2021. These publications were pre-evaluated by reading their abstracts. Articles that did not use CRISPR/Cas technique were not considered, as well as articles that only mentioned the technique. Articles were classified in four groups, according to the subject of study. The "Health" group included articles on treatment and prevention of human diseases, which were read in full and further characterized. A total of 327 non duplicated articles (extracted PubMed) and 166 patents (not all health-related) were found. 220 articles were ed after excluding articles that only mentioned the technique or not used CRISPR/Cas technique. These 220 articles were divided according to subject into four groups: Plants (n=16), Animals (n=16), Microorganisms (n=98) and Health (n=90). The remaining 110 patents were classified into groups also, according to subject: Plants (n=20), Animals (n=5), Microorganisms (n=9), Disease (n=35) and Unspecified (n=41). Most of the 90 articles have authors based in São Paulo, Rio de Janeiro and Rio Grande do Sul. In 50 out of 90 articles, more than 50% of the authors were affiliated to Brazilian institutions, however only three patents are Brazilian owners. Different diseases are the target of experimental CRISPR/Cas studies in particular cancer, inborn errors of metabolism and neurological disorders. Most patents refer to generic applications but those with clear disease indications are for hematological and immunological disorders. Brazil, a developing country, is increasing its progress in CRISPR/Cas research for treatment of human diseases. Although many groups have international collaborations, much work is produced locally by groups formed by Brazilian researchers only. However, this growth is not accompanied by the protection of intellectual property products.

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### **ACHADOS DA AVALIAÇÃO AUDITIVA DE UMA CRIANÇA COM A SÍNDROME DE SHAH-WAARDENBURGH**

CATEGORIA DO TRABALHO: RELATO DE CASO ÚNICO

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SANTA CASA DE MISERICÓRDIA DE PORTO ALEGRE

Introdução: a síndrome de Shah-Waardemburgh é causada por uma anormalidade da migração ou diferenciação das células da crista neural durante o desenvolvimento embrionário. Nosso