LETTER TO THE EDITOR

About the article: Hurler syndrome. Report of a case

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Mr. Editor:

Hurler syndrome was first described by the German pediatric specialist Gertrud Hurler in 1919 and is one of 11 types of mucopolysaccharidosis (MPS) described.(1) Due to its low frequency, it appears with the code 93473 in the Registry of Rare Diseases and Orphan Drugs (ORPHA),(2) so it is considered very relevant the publication of a report of a case diagnosed in Villa Clara Province because, as the authors state, it is interesting and of interest for students and health professionals;(3) nevertheless, it is considered appropriate to share some considerations regarding the referred article: in the section on patient information, two photographs of the patient are shown in which the facial dysmorphic pattern characteristic of patients suffering from MPS type I is observed, but no reference is made to obtaining the informed consent of the parents or guardians, and the patient is 12 years old, with possible severe intellectual disability.(3)

Students and health professionals should be aware that a photograph in Genetics, because of the data it provides in the clinical evaluation of the patient and family, as well as the phenotype-genotype correlation, constitutes human genetic data of as much importance as a deoxyribonucleic acid (DNA) sample and as such should be subject to the same ethical standards for its collection, with written informed consent, as with other biological samples.(4)

The image of a person is part of the sensitive data that needs to be protected. To this end, it is necessary to obtain the informed consent of the patient or the person legally responsible, prior to its collection. Special care must be taken to ensure respect, not only at the moment of taking the image, but also in its use. Photography in medicine is only justified if it is taken for the direct benefit of the person from whom it was taken or for teaching or divulgation purposes, always maintaining anonymity.(5)

In medical practice, photography can be an invaluable tool for the clinical evolution of a case, especially in dermatological lesions and endocrine-metabolic diseases, but in genetic diseases, due to its low frequency, it has been an essential weapon in the delineation of clinical phenotypes and, in addition, its publication has contributed to the development of Clinical Genetics as a science. In any case, informed consent must be obtained even if the face is not shown and the eyes or particular physical signs, such as tattoos that allow identification, are covered.(4,5)

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The photograph is genetic data associated with an identifiable person, so it is confidential and its disclosure requires prior consent. The International Declaration on Human Genetic Data of the United Nations Scientific and Cultural Organization aims to ensure respect for human dignity and fundamental freedoms in the collection, use and storage of human genetic data, as well as to establish the principles that should guide states in developing their legislation and policies on these issues.[4,6]

Article 7 of this declaration relates to non-discrimination and non-stigmatization of individuals and Article 8 refers to consent to the collection of human genetic data or biological samples, while Article 14, which deals with privacy and confidentiality, states that States should endeavor to protect the privacy of individuals and the confidentiality of human genetic data associated with an individual, a family or, where appropriate, an identifiable group, in accordance with domestic law consistent with international human rights law.[6]

In Cuba, Ministerial Resolution No. 219/2007 of the Ministry of Public Health establishes the ethical norms for the protection of the genetic information of Cuban citizens undergoing diagnostic assistance or participating in research in which data relating to the individual and his/her family members is accessed and regulates the aspects related to informed and express consent to collect genetic data.[4,7]

Regarding informed consent in obtaining photographs in the practice of Clinical Genetics, another detail to take into account is that the universe of patients generally includes the pediatric population and also people with intellectual disabilities. When a patient is declared incompetent or incapable of giving consent, the immediate problem that arises for the specialist is who should make the decision and this enters the field of representation or substitution decisions. In order to decide properly the legal representative must receive an adequate amount of information, the same that the patient would have received had he or she been capable.[4]

The child must have a legal representative who collaborates in the search for direct benefit, which may be questionable in the case of photography because it may be considered that the greatest benefit is for the physician who uses it in scientific publications, teaching activities or case presentations and only generates a direct benefit for the patient or family when the diagnosis depends on a medical interconsultation coordinated by the Clinical Genetics Specialist in which photography could play a leading role. More than a direct benefit in this case we should speak of non-maleficence.[4]

In the case of medical publications there are regulations proposed by the International Committee of editors in its section "Privacy and confidentiality of patients and study participants" in which the idea of informed consent is made explicit and the exclusion of information that may identify patients such as names, photographs and family trees is proposed (it will only be included if this information is essential for scientific purposes and the patient or legal representative has given informed consent for publication; the article must be shown to the patients before being published). If the patient's identity is recognizable, the signature on the informed consent form should accompany the article submitted.[4,8]

On the other hand, in the Discussion section it is stated that "both parents need to transmit the defective gene for their child to develop this syndrome"

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but, in the opinion of the authors, it is more appropriate to use the term mutation to refer to the change in the genetic material, or even the term sequence variant, according to the most recent recommendations of the joint consensus of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology,[9] and it can be read that "the chromosomal alteration has been identified and is located on the short arm of chromosome 4", but the authors consider it appropriate to recall that this syndrome has a monogenic and not a chromosomal cause, caused by homozygous or heterozygous mutations composed of the more than 201 mutations described in the Iduroninase Alpha L gene (IDUA), as the article refers, so it is this gene that has been identified and mapped, which is located or has its gene locus on the short arm of chromosome 4 (4p16. 3).[1]

Finally, the authors claim that "genetic testing and counseling, as well as having a complete family history profile, are important for prospective parents with a family history of the syndrome", so it only remains to recall that in diseases that are transmitted with an autosomal recessive pattern of inheritance, as the vast majority of inborn errors of metabolism, and this is no exception, there is no family history of the disease in the members of the couple, since they are healthy carriers, so that in this type of disease it is more pertinent to inquire about possible parental consanguinity links or the history of the syndrome in other siblings of the *propositus* or index case.

With these modest observations, this article fully complies with the objective of increasing the level of knowledge of students and health professionals with respect to this genetic syndrome of low prevalence, as well as with respect to the ethical norms to take into account for the report of cases that include photographs of patients with genetic diseases.

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CONFLICT OF INTEREST

The authors declare that they have no conflict of interest.

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