

# Medical teleconsultation in the care of children with suspected rare diseases: a pilot study in Brazil during the COVID-19 pandemic

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## Abstract

**OBJECTIVE:** To evaluate teleinterconsultation in medical genetics for cases of children with suspected rare diseases. **METHODS:** Prospective study of qualitative and quantitative evaluation of the use and satisfaction with asynchronous teleinterconsultation for physicians from public hospitals in a city in the northern macro-region of the state of Minas Gerais. **RESULTS:** A total of 21 teleinterconsultations were performed from September 29, 2020, to January 07, 2021. Approximately 52.3% of them were able to establish the probable diagnosis in the first evaluation, and the others were within the protocols of rare diseases investigation. Of the 12 requesting physicians, 91.6% were female, 58.2% were pediatric residents, and 41.8% were pediatricians. About 58% of the requesting physicians had never used telemedicine before. There was 100% satisfaction with the teleinterconsultation with the specialist geneticist. **CONCLUSION:** Teleinterconsultation in medical genetics for children with rare diseases met expectations, modified the conduct of non-specialist physicians in a city lacking a geneticist, and guided diagnosis and conduct in all cases.

**Keywords:** Telemedicine; Interconsultation; Genetics; Pediatrics; User Satisfaction

## Resumen

**Teleconsulta médica en el cuidado de niños con sospecha de enfermedades raras: relato de experiencia de un estudio piloto en Brasil durante la pandemia de COVID-19.**

**OBJETIVO:** Evaluar la teleinterconsulta en genética médica para casos de niños con sospecha de enfermedades raras. **MÉTODOS:** Estudio prospectivo de evaluación cualitativa y cuantitativa del uso y satisfacción de la teleinterconsulta asincrónica para médicos de hospitales públicos de una ciudad de la macrorregión norte del estado de Minas Gerais. **RESULTADOS:** Se realizaron un total de 21 teleinterconsultas desde el 29 de septiembre de 2020 hasta el 07 de enero de 2021. Aproximadamente el 52,3% de ellas lograron establecer el diagnóstico probable en la primera evaluación, y las demás se encontraban dentro de los protocolos de investigación de enfermedades raras. De los 12 médicos solicitantes, el 91,6% eran mujeres, el 58,2% eran residentes de pediatría y el 41,8% eran pediatras. Alrededor del 58% de los médicos solicitantes nunca antes habían usado la telemedicina. Hubo 100% de satisfacción con la teleinterconsulta con el genetista especialista. **CONCLUSIÓN:** La teleinterconsulta en genética médica para niños con enfermedades raras cumplió con las expectativas, modificó la conducta de los médicos no especialistas en una ciudad sin genetista y orientó el diagnóstico y la conducta en todos los casos.

**Palabras clave:** Telemedicina; Interconsulta; Genética; Pediatría; Satisfacción del Usuario

## Resumo

**Teleinterconsulta médica no cuidado de crianças com suspeita de doenças raras: relato de experiência de um estudo piloto no Brasil durante a pandemia de COVID-19.**

**OBJETIVO:** Avaliar a teleinterconsulta em genética médica para casos de crianças com suspeita de doenças raras. **MÉTODOS:** Estudo prospectivo de avaliação qualitativa e quantitativa do uso e satisfação da teleinterconsulta assíncrona por médicos de hospitais públicos de um município da macrorregião norte de Minas Gerais. **RESULTADOS:** Foram realizadas 21 teleinterconsultas no período de 29 de setembro de 2020 a 07 de janeiro de 2021. Aproximadamente 52,3% delas conseguiram estabelecer o diagnóstico provável na primeira avaliação e as demais estavam dentro dos protocolos de investigação de doenças raras. Dos 12 médicos solicitantes, 91,6% eram do sexo feminino, 58,2% eram residentes de pediatría e 41,8% eram pediatras. Cerca de 58% dos médicos solicitantes nunca haviam utilizado a telemedicina. Houve 100% de satisfação com a teleinterconsulta com o geneticista especialista. **CONCLUSÃO:** A teleinterconsulta em genética médica para crianças com doenças raras atendeu às expectativas, modificou a conduta de médicos não especialistas em uma cidade sem geneticista e orientou diagnóstico

*e conduta em todos os casos.*

*Palavras-chave: Telemedicina; Interconsulta; Genética; Pediatria; Satisfação do Usuário*

## Introduction

Rare diseases are those that affect up to 65 people per 100,000 individuals or 1.3 per 2,000 people. Although individually rare, as a group, they affect a significant percentage of the population<sup>1</sup>. In Brazil, it is estimated that about 13 million people have a disease that is considered rare<sup>2</sup>. In this group of diseases, 80% are genetic in origin. In 2014, the Ministry of Health published the ordinance that established the Policy of Integral Care for People with Rare Diseases within the Unified Health System (SUS). Rare diseases were classified according to their nature: genetic or non-genetic in origin. Those of genetic origin were subdivided into congenital anomalies vs. late manifestation and intellectual disability vs. innate errors of metabolism (IEM)<sup>1</sup>.

The early suspicion and, when possible, diagnosis of rare genetic diseases is fundamental and complex. The geneticist has a fundamental role in the early diagnosis and management of such diseases. Paradoxically, there is a shortage of geneticist physicians in Brazil, a country of continental dimensions with still evident social and geographic barriers. Data from the Medical Demography of Brazil estimates that there is a total of 300 active geneticist physicians throughout the country, the medical specialty with the second lowest number of professionals<sup>3</sup>.

The evaluation of persons with suspected genetic diseases requires careful attention to detail and anamnesis during physical examination and, when necessary, to specific molecular propaedeutics<sup>4</sup>.

The use of health information systems is useful in the epidemiological investigation of congenital malformations, one of the categories of rare diseases of genetic origin<sup>5</sup>. The Live Birth Information System (SINASC, in the Portuguese acronym), implemented in Brazil in 1990, aims to gather information on births occurring throughout the national territory. SINASC data come from the Certificate of Live Birth (DNV, in the Portuguese acronym). The DNV has a field called Field Six, with three options for completion regarding the presence of congenital anomalies: "yes" (when there are congenital anomalies), "no" (when congenital anomalies are absent) or "ignored" (when the information is not available to the professional who filled out the document)<sup>6,7</sup>. For SINASC to be effective

in monitoring and planning of public health actions, the information collected needs to be reliable. This is only possible with the correct filling of patient data and information in the collection instruments. When non-specialists in medical genetics evaluate and submit this data, there may be inconsistencies in the quality of information interfering with the overall evaluation of rare diseases<sup>7</sup>.

In this context, medical telegenetics, through teleinterconsultation between a geneticist physician and a non-specialist physician, is a helpful tool to address the demand for and concurrent lack of specialized professionals in the country. The COVID-19 pandemic further has highlighted its necessity, since lockdown has imposed isolation and subsequent social distancing. Thus, this study aimed to evaluate the use of teleinterconsultation in medical genetics in cases of children with suspected rare diseases, linked to public hospitals, in the main city of the northern macro-region of Minas Gerais.

## Method

This research was approved by the Ethics and Research Committee of the Rio de Janeiro State University under number CAEE 37441020.4.0000.5282.

This is an observational, descriptive, and cross-sectional study, with qualitative and quantitative analysis.

The following steps were followed to obtain the results: the selection and inclusion criteria of patients in the health macro-region in Minas Gerais with its respective main city were defined; invitations were sent to public hospitals with obstetrics services linked to the chosen main city to participate in the study; a specific form for teleinterconsultation in medical genetics was prepared; and the communication platform between the geneticist physician and the non-specialist physician was selected.

The form with clinical data for teleinterconsultation was modified and adapted from the form used in the Latin American Collaborative Study of Congenital Malformations (ECLAMC) to obtain the clinical data of patients with clinical suspicion of rare diseases in the teleinterconsultation protocol. ECLAMC is a clinical and epidemiological research program on developmental anomalies, working with hospital births

in Latin American countries. The case-control methodology is used to analyze potential risk factors in the causality of congenital malformations. In ECLAMC, charts are filled out for malformed patients and their controls. These forms contain information such as clinical examination, detailed description of the malformation, complementary exams, among others<sup>8</sup>. The form developed for information inclusion included specific fields for detailing information on family history, pregnancy, delivery, clinical findings, photographs, and imaging exams of the patient with suspected rare disease. The form was inserted in the remote assistance platform used.

For the selection of the macro-region to be studied in Minas Gerais, two criteria were selected: 1) the percentage and distribution of geneticist physicians in the 14 health macro-regions/main city of the state of Minas Gerais and 2) the percentage of filling in Field Six of the DNV as "ignored", through the evaluation of the data published in SINASC.

An uneven distribution of geneticist physicians was observed in the macro-regions of Minas Gerais, with most of them working in Belo Horizonte, the main city of the central macro-region (89.5%), while the other macro-regions had a shortage of specialists. In addition, when assessing the filling of DNVs through the analysis of SINASC data in 2017 (the most recent year in which data was available at the time the research began), the northern macro-region showed the highest rates of filling of Field Six as "ignored" (34%). Thus, this macro-region, with its respective main city (Montes Claros), was chosen as the object of this study.

The protocol for medical teleinterconsultation was as follows: the requesting physicians were registered; the Informed Consent Form (ICF), duly signed by the guardian of the target child, was inserted, and the detailed form for teleconsultation was filled out. The answer time from the specialist physician was up to 72 hours. In order to complete the information needed, audio contact was made for clarification and detailing. One geneticist physician sorted the first evaluation, and, in case of questions, two other geneticist medical specialists assisted.

To evaluate the level of satisfaction with the teleinterconsultation, the requesting physician filled in a supplementary form adapted from the Phrase Completion satisfaction level evaluation scale after receiving the expert geneticist's opinion. This form contained seven straight answer items and one open

item. In the first item of the questionnaire, the requesting physician was qualitatively questioned ("yes" or "no") about the previous use of a telemedicine platform. In the six subsequent items, the requester was asked to choose a score from 0 to 10 to the statements about how easy it was contacting the teleinterconsultation team in genetics, the ability to be guided by the teleconsultant, the satisfaction with the service received, the satisfaction with the information inclusion form, the intention of future use of teleinterconsultation in genetics, and the overall satisfaction with the teleconsultation. Satisfaction evaluated with a score of "0" means total absence of agreement and "10" means complete agreement. In the last item, the respondents were invited to criticize, suggest, or give a compliment.

## Results

In the period from September 29, 2020, to January 07, 2021, the two maternity hospitals participating in the study recorded a total of 4,345 births cumulative births. There were 21 requests for teleinterconsultation, demanded by 12 medical professionals (Table 1 and Table 2).

Approximately 11 patients (52.3%) were able to establish the probable diagnosis in the first evaluation, and the other 10 patients were within the protocols of rare diseases investigation based on the expertise of the medical geneticist. Of the patients evaluated, 13 (62%) were referred to genetic services for follow-up, four (19%) were awaiting reevaluation, two (9.5%) were discharged after evaluation, and there were two (9.5%) deaths (Table 1).

The main reason for teleinterconsultation request was the presence of dysmorphisms in the patient (10 patients; 47.6%). In addition, all the teleinterconsultation requests relating to congenital anomalies included multiple malformations; there was no request for isolated or minor congenital anomaly (such as: polydactyly, isolated myelomeningocele, pre-auricular alterations, among others).

In seven patients (33%), the diagnostic possibility of IEM was considered. The main clinical signs and symptoms suggesting the diagnosis of metabolic disease were positive family history of IEM, hypotonia, hepatosplenomegaly, anemia, congenital bilateral cataract, seizure, and coagulation alterations.

The genetic diseases suggested with diagnostic possibility in the period included: Jeune's asphyxiating

thoracic dystrophy, Noonan syndrome, Adenosine Deaminase Deficiency (ADA), Apert syndrome, Waardenburg syndrome, orofacioidigital syndrome, and trisomy 13. After the first evaluation, four patients (19%) required early stimulation and were referred to

physiotherapy, speech therapy, and occupational therapy care.

The youngest patient was seven days old and the oldest was 790 days old. The median age was 18.5 days. Ten patients (47.6%) were neonates.

Table 1- Description of the teleinterconsultation requests of the study (continues)

Date	Patient age, days	Pediatrician requesting?	Reason for the request	Diagnostic hypothesis	Conduct	Propaedeutics	Follow-up
10/01/20	15	Yes	Macrocrania, mesomelia, bell-shaped thorax	Jeune's asphyxiating thoracic dystrophy	Hearing evaluation, X-rays	Bone dysplasia panel: no changes. Karyotype: 46,XY.	Referred to Genetic Service - SUS.
10/05/20	8	Yes	Maternal half-brother with tyrosinemia type I	Healthy patient	Discharge from genetics for pediatric follow-up	-	Regular pediatric follow-up Discharge from genetics
10/10/20	28	Yes	Hypotonia, respiratory failure, hepatomegaly	IEM	IEM Screening	-	Follow-up with endocrinology and hematology; referred to Genetics Service - SUS
10/15/20	15	Yes	Suspected skeletal dysplasia	Apert Syndrome	Karyotype, x-rays and bone dysplasia panel	Karyotype: 46,XX	Referred to a specialist in medical genetics - private health care
11/15/20	90	Yes	Congenital clubfoot, type 1 laryngeal cleft, subglottic stenosis, and dysmorphisms	Chromosomopathy	Karyotype, hearing and ophthalmologic evaluation	Karyotype: 46,XY	Referred to Genetic Service - SUS
11/25/20	7	Yes	Family history of deceased siblings with Pompe disease	Family history of Pompe Disease (IEM).	Dosing of the acid alpha-glucosidase enzyme in leukocytes	Normal enzyme dosage.	Regular pediatric follow-up Discharge from genetics
12/10/20	790	Yes	Prolonged fever, hepatosplenomegaly, lymph node enlargement, anemia and malnutrition	Hemophagocytic lymphohistiocytosis IEM	IEM Screening Exome sequencing	Adenosine deaminase deficiency (ADA)	Death Genetic counseling for parents
12/21/20	9	Yes	Hypoplastic nasal bone, dysmorphisms (low ear implantation, short thorax and "tongue appendages" and postaxial polydactyly)	Chromosomopathy Orofaciodigital syndrome	Karyotype, whole body radiography, funduscopy and hearing evaluation	No response from the requesters of the propaedeutics performed	He was still in the hospital Awaiting reevaluation
12/29/20	30	Yes	Congenital knee dislocation, hypoplasia of buttocks, dysmorphisms (low ear implantation, micrognathia and hypertelorism)	Rasopathy Chromosomopathy Diabetic embryopathy	Karyotype, funduscopy and hearing evaluation	Karyotype: 46,XY	Referred to Genetic Service - SUS
01/06/21	-	No	12-week abortion	Chromosomopathy	Parental karyotype	No access to the result	Parents referred to the Genetics Service - SUS

Table 1- Description of the teleinterconsultation requests of the study (continued)

Date	Patient age, days	Pediatrician requesting?	Reason for the request	Diagnostic hypothesis	Conduct	Propaedeutic s	Follow-up
01/13/21	30	Yes	Mild facial dysmorphism, camptodactyly of hands, overlapping and shortening of 4 and 5 toes of left foot, cutaneous syndactyly, seizures, sacral fossa (occult dysraphism?)	Dimorphic syndrome Chromosomopathy Diabetic embryopathy	Radiography of hands and feet and karyotype	No access to the result	He was still in the hospital Awaiting reevaluation
01/26/21	19	Yes	Dysmorphisms and bilateral congenital cataract	Fraser syndrome Numerical abnormality IEM	Karyotype, IEM screening	Karyotype: 46,XX	Referred to genetic service - SUS and referred to stimuli: speech therapy, physical therapy and occupational therapy
02/03/21	18	Yes	Dysmorphisms, ventriculomegaly, seizures, congenital clubfoot, iris and retinal coloboma	Dysmorphisms	Karyotype	No access to the result	Referred to Genetic Service - SUS
02/12/21	12	Yes	White hair in frontal region, rhizomelia	Waardenburg syndrome	Hearing and ophthalmological evaluation	No access to results	Referred to Genetic Service - SUS
02/12/21	9	Yes	Microphthalmia, polydactyly, dysmorphisms, aplasia cutis	Trisomy 13	Karyotype, Echocardiogram, Transfrontanelle ultrasound	Karyotype: 47, XX, +13	Referred to genetic service - SUS and stimuli: speech therapy, physical therapy and occupational therapy
04/01/21	120	Yes	Anemia, convulsion, diarrhea, NPMD, ascites, alteration in coagulation	CDG, tyrosinemia (IEM)	IEM screening Gene panel of neurodevelopment	No access to results	Death Parents referred to genetic service - SUS
04/05/21	240	Yes	Pulmonary valve stenosis, dysmorphisms, and mother with similar phenotype, karyotype 46,XY	Noonan Syndrome	Coagulation evaluation, ophthalmological and hearing evaluation	Propaedeutics without alterations	Referred to genetic service - SUS and stimuli: speech therapy, physical therapy and occupational therapy
05/26/21	32	Yes	Seizures of difficult control and hypospadias	Multifactorial disease IEM	Gene panel of neurodevelopment	Karyotype: 46,XY Neurodevelopment panel without alterations	Referred to a specialist in medical genetics - private health care
06/14/21	60	Yes	Tyrosine crystals in urine test Consanguineous parents (first cousins)	Tyrosinemia (IEM)	Dosage of plasma tyrosine and methionine. Urinary or plasma succinyl acetone dosage	No access to results	Awaiting reevaluation

Table 1- Description of the teleinterconsultation requests of the study (conclusion)

Date	Patient age, days	Pediatrician requesting?	Reason for the request	Diagnostic hypothesis	Conduct	Propaedeutic s	Follow-up
06/21/21	41	Yes	Hypotonia, arthrogryposis, dysmorphism	Structural chromosomopathy	Requested CGH-array	Karyotype: 46,X,Yqh+ Awaiting result of CGH-array	He was still in the hospital Awaiting reevaluation
07/01/21	180	Yes	NPMD, dysmorphism (short stature, low ear implantation, winged neck), pulmonary valve stenosis	Noonan Syndrome	Abdominal ultrasound, ophthalmological and hearing evaluation, laboratory review	Karyotype: 46,XX	Referred to genetic service - SUS and stimuli: speech therapy, physical therapy and occupational therapy

NPMD: Neuro-psychomotor Developmental Delay; ADA: Adenosine Deaminase Deficiency; CGH-array : Comparative Genomic Hybridization; CDG: Congenital Disorders of Glycosylation; IEM: Inborn Errors of Metabolism.

Of the 12 requests, most were physicians (91.6%), residents in pediatrics (58.2%), and had never used medical teleinterconsultation services (58.3%). The other 41.8% of the requests were pediatricians. (100%) and reported that, in case of new doubts regarding the genetic diagnosis of patients with suspected rare diseases, they would request the teleinterconsultation again (Table 2).

All participants were satisfied with the service provided

Table 2: Evaluation of satisfaction with teleconsulting in medical genetics

Have you used telemedicine platform for consulting purposes before?	Yes No	5 (41,7%) 7 (58,3%)
How easy was contacting the medical genetics telecare team. Consider 0: extremely difficult and 10: extremely easy	Grade	Number of participants
	7	2 (10%)
	8	4 (30%)
	9	2 (20%)
	10	4 (40%)
Consultant's ability to provide guidance on the case presented. Consider 0: extremely unable and 10: extremely able.	Grade	Number of participants
	10	12 (100%)
Satisfaction with the service received. Consider 0: extremely dissatisfied and 10: extremely satisfied.	Grade	Number of participants
	10	12 (100%)
Satisfaction with the presentation of the information inclusion form. Consider 0: extremely dissatisfied and 10: extremely satisfied.	Grade	Number of participants
	10	2 (16,66%)
	9	2 (16,66%)
	8	4 (33,3%)
	7	2 (16,66%)
	6	2 (16,66%)
In other cases where you are in doubt about a patient's genetic diagnosis, you would request the teleinterconsultation again. Consider 0: strongly disagree and 10: strongly agree.	Grade	Number of participants
	10	12 (100%)
Overall satisfaction with the teleinterconsultation in medical genetics received. Consider 0: extremely unsatisfied and 10: extremely satisfied.	Grade	Number of participants
	10	12 (100%)

The satisfaction questionnaire also had an open field, where the physician was asked to give his comments, doubts, or criticisms. Approximately 17% of the applicants filled this field, and the answers were positive, as exemplified below:

Answer 1: "Very satisfied and grateful, since where I work the evaluation of genetic syndromes is very limited".

Answer 2: "The geneticist was very helpful at all times, responded quickly to demands, and sent a report that made it easy to understand the case, enabling a good

follow-up of the patient".

## Discussion

This is a pioneer study on the use of medical telegenetics for children with suspected rare diseases in Brazil. This study indicates that the strategy may have been resolute in 61.8% of the cases in the first teleinterconsultation, since 52.3% of the cases had the probable diagnosis and 9.5% were discharged from the hospital. Our results also show that assisting

physicians of children with suspected rare diseases, especially pediatric residents, seek a second opinion from a geneticist, although most of them do not use telemedicine as a tool in their clinical practice.

There were 21 requests for teleinterconsultation, requested by 12 different professionals during the study execution period, in agreement with the literature in similar studies with a mean of less than or equal to 50 participants<sup>9,10,11</sup>.

The use of medical telegenetics is still poorly evaluated<sup>9,12-16</sup> and similar studies address the role of telegenetics in genetic oncology counseling<sup>12-14</sup>, but few report the role of telegenetics in the clinical evaluation of patients with congenital anomalies and IEM<sup>15,16</sup>.

Similar to our study, Gold et al. evaluated the use of telegenetics from the perspective of user satisfaction. In that study, health professionals' satisfaction with telegenetics was assessed when applied in the care of children with altered neonatal screening for IEM during the initial months of the COVID-19 pandemic. An online survey was sent to healthcare professionals registered on the Metab-L server, an international email list for discussion of clinical care in IEM. Participants completed the survey using an online questionnaire containing 69 objective questions and four open questions. Participants' answers were registered, and satisfaction was assessed using the Likert scale. The questionnaire was answered by 44 health professionals from eight different countries and several professional categories involved in the care of children with abnormal newborn screening (physicians, genetic counselors, nutritionists, and social workers). There was no grouping of the assessment by professional category of the participants. More than 93% of participants practiced telegenetics for care of patients with abnormal newborn screening in the first months of the COVID-19 pandemic. About 50% of users agreed that telehealth is effective for evaluation of abnormal newborn screening<sup>17</sup>.

A related study developed by Mena et al. also evaluated the role of teleinterconsultation in medical genetics. However, unlike our study, in which the teleinterconsultation request could be made by any medical specialty involved in the care of patients with rare diseases and in which the teleinterconsultation was performed asynchronously, the requesters included by Mena et al. were exclusively pediatricians and the evaluations were performed synchronously. Initially, patients were evaluated in person by a

pediatrician in Santo Domingo, Dominican Republic, a region with a shortage of geneticist physicians. After this, if there was an indication for genetic evaluation, the patient was evaluated remotely through video conference by a medical geneticist from the Cincinnati Children's Hospital Medical Center, USA. During the teleinterconsultation, the pediatrician performed the physical examination, which included neurological examination and dysmorphology evaluation guided by the geneticist. A total of 66 individuals with suspected genetic disorders were evaluated from 2015 to 2020. Similar to our study, after the evaluation, the geneticist issued an expert opinion to the pediatrician, guiding the clinical evaluation and conduct<sup>16</sup>.

Similar to our study, dysmorphisms were one of the main reasons for referring children to teleinterconsultation in the study of Mena et al. They also reported lower diagnostic acuity than that found in our study after the first clinical evaluation by teleinterconsultation (7.6% and 52.3%, respectively). The main diseases clinically diagnosed in that study were: neurofibromatosis type 1, Poland syndrome, and rasopathies. It is noteworthy that, unlike our study, diagnostic molecular tests were available and offered to the patients when necessary. After molecular propaedeutics, the diagnostic acuity increased to 59%. Unlike our study, no user satisfaction survey was performed. However, a high level of satisfaction with safety, efficiency, and diagnostic skills were reported through informal feedback from all patients' families<sup>16</sup>. It is worth remembering that dysmorphisms, the main reason for requesting teleinterconsultation in our study, are part of the vast majority of genetic syndromes<sup>18</sup>. A study by Wenger et al. evaluated the role of telegenetics for clinical evaluation of patients with dysmorphisms admitted to an intensive care unit. A geneticist physician performed an in-person physical examination that was synchronously transmitted to another geneticist physician who was geographically distant. Subsequently, the physical examination performed remotely was compared with the physical examination performed in person by the specialists. For image transmission, an AMD-2500 camera with a 50x zoom lens was used to transmit the recording of the physical examination to the geneticist physician who was remote through the InTouch Health RP-Lite telemedicine platform<sup>10</sup>. The authors concluded that the clinical evaluation by means of telegenetics was able to identify 93% of the dysmorphisms reported in the in-person examination.

Similar to our study, all patients evaluated by Weng et al. were polymalformed; however, they had a smaller age variation than the one found in our study (21 to 140 days versus 7 to 790 days, respectively)<sup>10</sup>. Moreover, in our study, the patients were not reevaluated in person by the medical geneticist, and there was no comparison between on-site and remote evaluation, as was done in that study. However, our study also used a validated platform for communication between the geneticist and the non-geneticist physicians. The use of a validated communication platform is essential for the security and confidentiality of sensitive data from telemedicine users. Both studies are pioneers in the evaluation of medical teleinterconsultation in genetics.

It is important to highlight that our study obtained a high level of satisfaction among physicians who used teleinterconsultation. Similar statements are reported in the literature, such as the study by Gorrie et al. These authors conducted a literature review in the Pubmed database with descriptors related to telegenetics in the period from 2001 to 2019. Twenty-one articles were included in the review. Most of the included studies presented methodology similar to our study; they were qualitative-quantitative analyses, they used teleinterconsultation among the modalities of telecare, and they developed their own online form for the evaluation of user satisfaction<sup>9</sup>.

In that study, the overall satisfaction of health professionals with the use of telegenetics was high in most of the studies included in the review. The authors highlight reports from physicians about the benefits of telegenetics regarding the speed between the request and the call, especially in places where distance was a limiting factor<sup>9</sup>. A similar observation was also made in our study, with reports from users that one of the benefits of teleinterconsultation was the speed of the expert opinion.

Therefore, the teleinterconsultation in medical genetics for children with suspected rare diseases met the expectation of non-specialist physicians in a city lacking a geneticist and guided diagnosis and management in all cases. Our data highlight the importance of appropriate clinical evaluation by a geneticist of children with suspected genetic disease.

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