Issue 02/2025

MAGAZINE

CASA HUNTER

DAY HUNTER 2025

EDITORIAL

The Day Hunter magazine is the result of a collective effort to register the work developed with rare patients and their caregivers in five Brazilian capitals: Sao Paulo, Rio de Janeiro, Salvador, Goiânia and Belo Horizonte (still being implemented). We had a significant number of services in 2024, which registered a growth of 22% when compared to the previous year

The data also speak of Brazilians seeking a diagnosis (46%), while dealing with a challenging economic reality: income below R$2,500.00 in 66% of cases. They are men, women and children who were received in an outpatient clinic and seen to simultaneously by a group of specialists, which includes doctors, physiotherapists, psychologists and neuropsychologists, speech therapists, nurses, nutritionists and social workers.

There, it is worth highlighting, lies the program's biggest difference! A multidisciplinary service, made up of specialists qualified to deal with complex cases, which involves the presence of doctors and professionals from other fields of health. In its 10 years of existence, the program has challenged models, shortened processes and proven the need to create protocols suited to the reality of rare diseases.

We invite you to dive into this document and discover the potential of this challenging program! Here, you are more than welcome!

Antoine Daher

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Casa Hunter: transparency, governance and social commitment

Casa Hunter has stood out as a reference in the care of patients with rare diseases in Brazil, going beyond medical care and offering a multidisciplinary approach, centered on solid principles of governance and social commitment.

For pharmaceutical industry executives and public health managers, Casa Hunter's trajectory offers an inspiring model of how health initiatives can combine innovation and responsibility in such a sensitive sector.

Structured governance and transparency

Casa Hunter's governance is composed of three essential boards: the Scientific Committee, which brings together doctors and specialists in rare diseases to guide the institution in best practices; the Audit Committee, responsible for the efficient and transparent administration of resources; and the Board of Directors, which defines the strategic guidelines. These committees not only ensure solid management, but also promote an environment of integrity and active participation by society. The creation of an efficient ombudsman's office reinforces

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transparency and ensures that all demands from patients and families are heard and responded to quickly, promoting continuous improvement of services.

ESG practices in the health sector

Casa Hunter also adopts practices in the field of ESG (Environmental, Social, and Governance). The institution seeks to reduce its environmental impact through initiatives such as the Zero Carbon project and the digitalization of administrative processes, aiming to reduce the use of paper. In the social field, Casa Hunter stands out for its commitment to inclusion, ensuring that patients with rare diseases and their families receive comprehensive support, with access to assistive therapies and legal, psychological and social support.

Comprehensive assistance and sustainability

The assistance provided by Casa Hunter goes beyond clinical support. The Day Hunter program, for example, offers a complete multidisciplinary assessment, providing humanized care to patients. In addition, the institution distributes basic food baskets and essential equipment, such as wheelchairs and respiratory devices, to vulnerable families, demonstrating its commitment to equity in access to treatments.

This model of governance and assistance is especially relevant at a time when the healthcare industry faces growing regulatory and social challenges. To build a new health ecosystem, Casa Hunter has sought global action, with a presence in global organizations such as Rare Diseases International (RDI), responsible for leading discussions on the topic with member countries of the United Nations.

The objective is one: to exchange experiences, reduce distances and accelerate the adoption of good practices throughout Brazil.

Consultations

THE JOURNEY OF A PATIENT WITH A RARE DISEASE: DAY HUNTER AS A GATEWAY TO STARTING TREATMENT IN THE SUS

Patients with rare diseases and their families often visit various services, public or private, seeking medical and multidisciplinary care. In many cases, these people do not have all of their needs met and, as a result, they experience many doubts and suffering.

Currently, there are few specialized services for treating patients with rare genetic diseases in Brazil, with 342 geneticists throughout the country, and most of these professionals work in the main urban centers, causing helplessness for a large part of this population and slowness in scheduling tests and consultations in the public health system.

Aiming to help minimize these difficulties, the Day Hunter program proposes a more agile, comprehensive care system that requires fewer family trips, shortening the waiting time for the diagnostic investigation process and assessment of the patient's general condition.

The program has multidisciplinary teams that work in the same place and on the same day, serving patients who do not yet have a diagnosis, as well as those who already have their diagnosis clarified, but still have doubts about the disease and the necessary treatment or have not yet undergone a multidisciplinary evaluation after receiving the diagnosis.

Multidisciplinary teams working in the same place on the same day

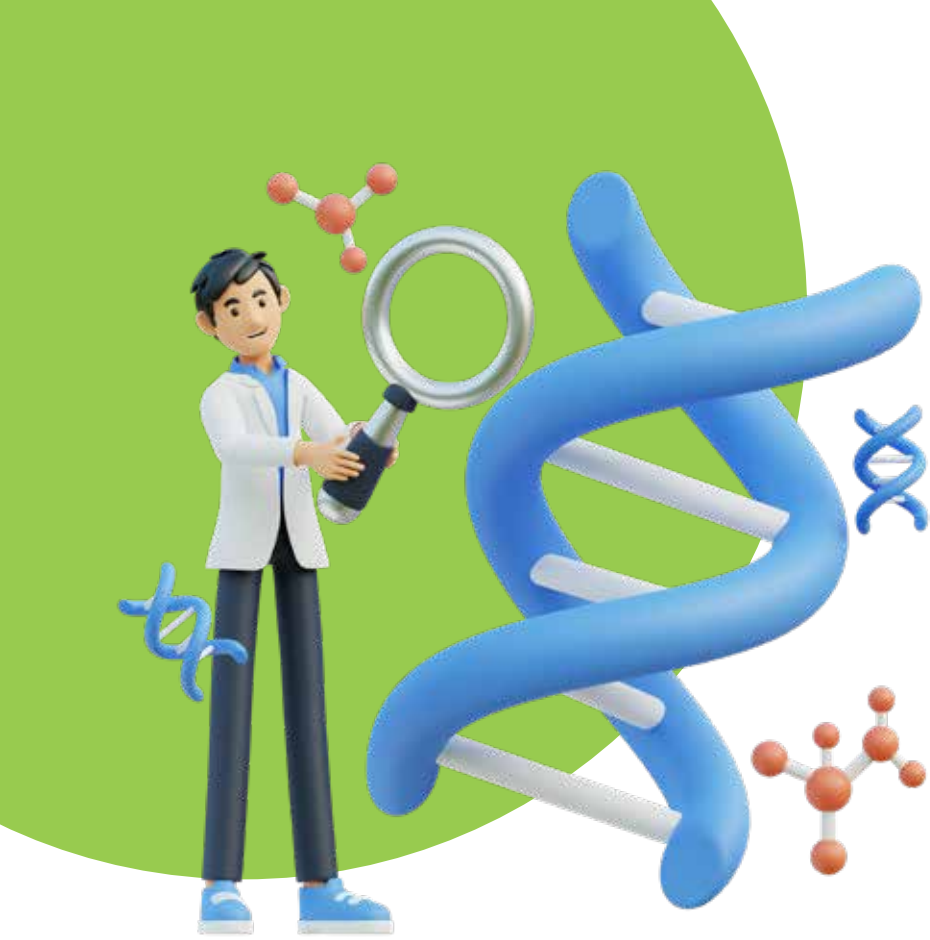
Follow-up

The program offers diagnostic research in an interdisciplinary manner, complemented by genetic counseling and psychological support for patients and their families. The patient and their caregivers are informed about the disease, treatments and prognosis and receive guidance on necessary care in different areas for a better quality of life. They also receive assistance in coordinating the public care network, both in the health and welfare areas, as many are unaware of their rights and the services they can access.

After going through this initial process of care and assessment, the patient can continue their care in the SUS equipped with guidance and clarifications about their illness and proposals for interventions necessary for their well-being.

Day Hunter team professionals are available to talk, discuss cases and guide the professionals and teams that will carry out the patient's medical and therapeutic follow-up.

This is how the Day Hunter program can contribute to the dynamics of the SUS, assisting in the diagnostic process and providing the necessary information so that the referral and treatment process within the health system is streamlined and assertive.



SCIENTIFIC PRODUCTION AT DAY HUNTER: KNOWLEDGE THAT TRANSFORMS CARE

In addition to the humanized and multidisciplinary care offered to patients with rare diseases, Day Hunter also stands out as a fertile space for the production of scientific knowledge. Many of the professionals involved in the program, driven by the constant search for answers in an area still marked by so many gaps, have dedicated themselves to the systematic collection of data and the development of relevant studies.

This vocation for research arises from practice: from clinical observations, patient demands and the team's commitment to improving care through science. Day Hunter's collaborative environment offers professionals the opportunity to study and publish relevant information, contributing to the construction of more effective management guidelines and improving the quality of life of people with rare diseases.

Below, we present three scientific papers prepared by Day Hunter professionals that illustrate how clinical practice can boost the production of evidence and strengthen networking in the area of rare diseases.

Nely Barbosa de Matos, nutritionist

Nely Barbosa de Matos, team nutritionist **Day Hunter of Salvador**, presented at the III ATTRv-PN CONNECTION Event, held on June 30, 2023 in Fortaleza, the promotion of nutritional care within the multidisciplinary team for people with ATTRv-PR Amyloidosis.

The work entitled **Day Hunter Characterization: Multidisciplinary Care Model for Rare Diseases/SP** was prepared by the Day Hunter team and presented in the Original Scientific Work category in poster format by the professional André Luiz de Sousa (neuropsychologist) and Dr. Bianca Domit Erner Linnennenkamp at the III Brazilian Congress of Neurogenetics, organized by the Brazilian Academy of Neurology (ABN) held on March 9, 10 and 11, 2023, in the city of São Paulo/SP – Frei Caneca Convention Center.

André Luiz de Sousa, neuropsychologist, and Dr. Bianca Domit Erner Linnennenkamp

Correlation Between the Eating Assessment Tool (Eat-10) and Swallowing Quality of Life in Patients with Rare Neuromuscular Disease.

This is a work presented in 2021 at the Brazilian Congress of Neurology by speech therapist Deborah Sales. It was carried out with data from all patients with neuromuscular diseases treated at Day Hunter RJ, including Spinal Muscular Atrophy, Amyotrophic Lateral Sclerosis, Pompe Disease, Limb-Girdle Dystrophy and Myotonic Dystrophy. A total of 73 patients were evaluated, and the study showed that dysphagia is a prevalent symptom in these diseases and that it significantly impacts the quality of life of patients – it is worth noting that this symptom should be addressed during their follow-up.

THE IMPORTANCE OF SPECIALITIES IN THE TREATMENT OF RARE DISEASES

We understand that not all patients have diseases that can be treated with medication, but they all need and deserve to be cared for in a welcoming and individualized manner.

Promoting care for people with rare diseases is essential due to the multisystemic involvement of most of them. Furthermore, the needs of these patients go beyond health issues. Not only the person with a rare disease, but also their family group, may have needs in the psychological and social fields.

In view of this, the multidisciplinary teams at Day Hunter work to meet the needs of patients and their families, seeking to alleviate the setbacks and details involved in the experience of being rare and caring for or providing assistance to a rare patient.

They are teams made up of qualified professionals, who perform their roles with seriousness and care and who have an eye not only on the specificities of their areas, but also on the alignment of their observations with the views of all the other professionals on the team, highlighting the interdisciplinarity of Day Hunter.

Below is the list of specialties that make up the Day Hunter teams:

Psychologist

Psychological care at Day Hunter is essential to understand and welcome the emotional impacts experienced by patients with rare diseases and their families. The work is carried out with both the patient and their support network, promoting qualified listening, support and guidance. The service includes psychological support at the time of the consultation, as well as brief psychotherapy of four sessions and referral for ongoing support, when necessary. During this process, patients and family members can express emotions, expectations and coping strategies in a safe, judgment-free space. Psychology also works with neuropsychological assessment and psychoeducation, which contribute to understanding the cognitive and behavioral effects associated with certain syndromes, offering important support for more effective interventions. The results of this work are reflected in emotional relief, strengthening of self-esteem, greater adherence to treatments and improved quality of life. Psychological work at Day Hunter expands the care network and offers essential support to mental health of rare patients and their families.

The results include: relief, acceptance, hope, resilience, reduced levels of anxiety and feelings of helplessness, clarity about the impact of the health condition on their lives, greater possibilities for facing challenges, increased self-confidence and self-esteem and better adherence to treatments.

Social service

The purpose of social services is to discuss with families possibilities to guarantee rights and access to services and programs that promote the quality of life of patients with rare diseases and their families.

At Casa Hunter, social services are present at the beginning of the care provided to the family and the patient: introducing the institution, explaining the flow of our care and listening to initial needs, complaints and reasons for seeking care at Casa Hunter. It also collects data for the social classification of each family, in addition to working on opening new partnerships with places that can provide medical and therapeutic assistance to patients who were evaluated by Day Hunter.

During the patient assessment process, social services participate in case discussions with the multidisciplinary team and attempt to align with partner services and professionals to care for patients and families.

At the end of the evaluation process, this professional listens to the families and/or patients about any doubts and pending issues that may still remain after having been evaluated and guided by each professional on the Day Hunter team. Finally, we provide guidance on access to public policies in your municipality or state.

After our interventions, we observed the positive impact that social services have on the lives of patients and families when they feel heard, welcomed, guided and informed about services and rights that can improve their quality of life.

Nursing

Nursing work in rare diseases is essential for the management and monitoring of specific care due to the complexities of these conditions. At Day Hunter, nursing activities are divided into:

Service management – Production, preparation and presentation of scientific papers; analysis of indicators; preparation of specialized booklets; creation of routines and service protocols; teleconsultations; participation in clinical sessions; discussion of cases; organization of health education activities with patients; and continuing education with professionals.

Care management – Individual- and family-centered care provided through welcoming patients and their families; nursing consultations; active search for patients with difficult access; collection of laboratory and genetic tests; participation in the development of the therapeutic plan with the team; and establishment of effective communication between the patient and the other professionals on the team.

Nursing professionals perform their role with care, demonstrated by actions such as calling during the week to ask about the patient, offering a nutritious snack during the appointment, and organizing workshops, recreational activities and online meetings with patients, promoting interaction and exchange of experiences.

It is extremely gratifying and motivating to see that guidance and care have beneficial effects on users. And it is more rewarding when the patient returns to appointments grateful and showing improvements.

Speech therapy

The speech therapist is a professional trained to assess, diagnose and treat communication disorders (speech, language, voice and hearing) and changes in the stomatognathic system, orofacial motricity and dysphagia (swallowing disorder), without neglecting prevention and health promotion actions.

Our team is ready to serve adult and pediatric patients, using protocols according to age group and individual needs.

Most patients with rare diseases have dysphagia, which is a significant swallowing disorder, with a high risk of bronchoaspiration. The speech therapist performs immediate interventions, such as dietary adaptations, or even recommends an alternative feeding route (nasoenteral tube or gastrostomy), according to medical evaluation.

Improvements in patients are noted as a result of speech therapy interventions, from small guidelines for language stimulation and oral hygiene, to more delicate ones, such as in situations involving lung health, general well-being and patient development. The improvement is significant compared to the difficulties and/or risks observed in the assessment.

The speech therapist plays a fundamental role, both in improving communication with the social environment and in the pleasure, well-being and safety during a meal.

Physiotherapy

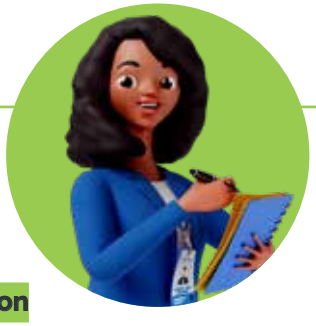
In rare diseases, the physiotherapist's approach must be comprehensive, as it aims to maintain as much functionality as possible, avoiding loss of strength, muscle mass and coordination.

Motor physiotherapy assesses strength, balance, tone, sensitivity, postural changes, gait, presence of deformities and delay in neuropsychomotor development. It uses validated tests and scales to assess any patient profile, diagnosed or seeking a diagnosis.

In **respiratory physiotherapy** we aim to prevent and treat diseases that affect the respiratory system. We provide techniques that help improve breathing, mobilize the respiratory muscles, facilitate the arrival of oxygen to the tissues and clear the airways. In the case of patients who use invasive or non-invasive mechanical ventilation, we evaluate and provide guidance on parameters and accessories, such as the necessary monitoring of saturation, vital signs or the presence of respiratory discomfort.

After the guidance, an improvement in the patient's quality of life is observed, as their guardians are guided on day-to-day care, whether with exercises, techniques and therapies learned in the care to adapt the ergonomics of the home. Prescriptions are made for orthoses, prostheses and walking devices that best suit the patient's needs.

The physiotherapist is an essential professional in monitoring people with rare diseases. It is highly capable of preventing possible injuries and rehabilitating patients, preventing damage related to disease progression and providing quality of life.



Nutrition

In nutritional consultations, health is addressed as a whole, seeking to understand how nutrition can improve patients' quality of life. It is necessary to take into account the culture, preferences, lifestyle and impact of the disease on the person's daily life. Among the main complaints of patients and their guardians are constipation/diarrhea, difficulty chewing and/or swallowing, loss of weight and muscle mass, food selectivity, overweight/obesity, diet via gastrostomy and the appearance of chronic diseases.

A dietary record is made, in which the patient's eating habits and routine are analyzed. An anthropometric assessment is also carried out (weight, height, body mass index and other important measurements).

During consultations, nutritional conduct is defined and a personalized eating plan is drawn up. Prescribing and implementing a meal plan can be challenging, as it involves food groups, proportions, diet consistency and hydration, in addition to the process that goes from food preparation to the meal itself.

Each time the patient returns, a new anthropometric assessment is carried out – and the eating plan is reviewed and readjusted if necessary.

In this way, it is possible to carry out nutritional therapy with a focus on understanding how food can help improve quality of life and symptoms, respecting the individuality of each person

Doctor

In the Day Hunter teams we have doctors from the fields of genetics, neurology and pulmonology, who are essential in the care of these patients, although we know that all medical specialties play an important role in this work, depending on the disease.

These are the professionals who receive patients who arrive at Day Hunter and, in a careful and caring manner, give them the news about the diagnosis, explain the disease, treatments, needs and clarify doubts.

They carry out a thorough, specialized and complete assessment and refer the patient for tests, aiming to obtain an adequate diagnosis and assertive treatment planning, outlining a line of care that includes medication (when possible), referrals and general guidance, essential for diagnosing and treating diseases and symptoms, mitigating the impact on the individual and their social context.

They provide support to the patient's support network, including family members, schools and professionals who accompany the patient in other services, with essential information about the disease and appropriate care.

They consider the support of a qualified multidisciplinary team to be essential for improving the quality of life of patients and their families. Collaboration between different specialties is vital to ensure that all nuances of the patient's condition are considered and an effective plan of action is developed.

The geneticist performs genetic counseling, which is a fundamental part of this work. Aimed at a couple, a patient or a family, it is a moment of guidance and support, in which not only the medical aspects are addressed, but also the emotional and social issues that may arise as a result of the diagnosis.

We have realized how grateful patients and family members are and how much more secure they feel when they receive a diagnosis and a path to follow. This reduces anxiety for families and improves the quality of life for patients and caregivers.

Social Worker:
Juliana Barica Righini

Nurses:
Diego Marques de Santana
Geovanna Liscio Pereira

Speech therapists:
Cíntia M. Gonçalves
Deborah Sales
Gabriela Prearo
Juliana Soave
Larissa Menezes
Regina Khoury

Physiotherapists:
Adriana Virginia Barris Faiçal
Mariana de Castro
Paula Tinel
Raquel Gonçalves de Paula
Vivian Pinto

Doctors:
Bruno Marcarini
Daise Larissa França
Eder A. Moura
Juliana Leão
Karina L. Pires
Lusmaia D. C. Costa
Marcela C. M. Costa

Nutritionists:
Mariana Sisdelli
Nelly Barbosa de Matos
Rayanne Souza

Psychologists:
André Luiz de Sousa
Carolina Carrijo
Cátia Ap. Cardoso de Macedo
Clarissa de Araújo Davico
Lucas de Medeiros Silva
Marylia Glenda Lopes Dep. Sousa

DAY HUNTER AND ITS TEAMS

The Day Hunter program proposes a care model that can be implemented in health services, enabling the expansion of the network of locations for treating patients with rare diseases. This expansion of the program already has 4 units.

In this section, we are pleased to introduce Day Hunter teams in different regions of the country. We are also providing updated addresses of where each of our units operate. For more information, you can visit the Casa Hunter website.

Day Hunter Goiás

Address: Hospital de Clínicas da Universidade Federal de Goiás – 1ª Avenida, s/n, quadra 68, Área 1 – Setor Leste Universitário – Goiânia-GO

Email: dayhunter.go@casahunter.org.br

Day Hunter Bahia

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Email: dayhunter.ba@casahunter.org.br

Day Hunter Rio de Janeiro

Address: Hospital Gafrée e Guinle – Rua Mariz e Barros, 775 – Maracanã – Rio de Janeiro-RJ

Email: dayhunter.rj@casahunter.org.br

Day Hunter São Paulo

Address: Instituto Superior de Medicina (ISMD) – Rua Conselheiro Rodrigues Alves, 213 – Vila Mariana – São Paulo-SP

E-mail: dayhunter.sp@casahunter.org.br

THE IMPORTANCE OF MULTIDISCIPLINARY CARE

The following case was handled by the Day Hunter team in São Paulo. However, it symbolizes the interdisciplinary work of all Day Hunter teams operating in the national territory.

We had the privilege of meeting an amazing girl, very smart and communicative! Her story is inspiring, and sharing it can provide comfort and hope to so many other families who need to feel embraced and encouraged to move forward, as well as publicize the importance of interdisciplinary work for people with rare diseases and demonstrate the valuable contribution of Casa Hunter as a companion to many patients and families on their journeys.

The family of Alice, who was 12 years old at the time, sought out Casa Hunter in São Paulo in search of more guidance and resources for the patient's clinical, therapeutic and social follow-up. Her interdisciplinary assessment involved professionals from the areas of genetic medicine, neuropsychology, physiotherapy, speech therapy and social work.

Alice presents hypothesis of **Cerebrocostomandibular Syndrome**, awaiting genetic tests. During pregnancy, the morphological ultrasound had already identified multiple alterations and fetal malformations. She has musculoskeletal alterations, such as severe mandibular micrognathia and glossoptosis, rib cage dysmorphisms, polyarticular juvenile arthritis, knee hyperextension, thoracolumbar scoliosis, gibbosity and thorax carinatum.

During the first months of her life, while still hospitalized after birth, she underwent several surgical procedures. She was discharged at 7 months of age and continued with homecare. She then developed multiple episodes of pneumonia, global neurodevelopmental delay and underwent several thoracic distraction procedures and rod placement for scoliosis. At the age of 4, she began to speak words and began to eat orally.

She had severe bilateral hearing loss, corrected with a hearing aid only on the left ear because, due to the malformation of the right ear, it is difficult to adapt the device to that side.

During the multidisciplinary assessment, she was collaborative, communicative and spoke very enthusiastically about her daily activities and her performance on social media. Despite the difficulties observed in expressive communication, Alice demonstrated a sense of humor, interacted, made spontaneous comments and questions, and answered everything that was asked. She also reported that, when she grows up, she wants to become a veterinarian.

She is literate. She was placed in a classroom for special needs children, supposedly due to physical malformations.

However, the results of the neuropsychological assessment indicated overall intellectual perfor-

Alice and her parents

mance, cognitive functions and adaptive skills within what was expected for her age.

And measuring these results was extremely important, as it made it possible to conclude that Alice had the potential and desire to advance in her academic knowledge and the conditions to attend a regular class.

The speech therapy assessment found well-developed language, despite the hearing impairment, and some difficulties in producing speech, voice and eating, caused by the anatomical changes in the mouth and teeth.

Physiotherapy observed that, despite the physical changes, Alice has autonomy and independence in most activities of daily living, with some reservations, such as bending down and tying her shoelaces.

Completion of the assessment and guidance to the family

During Alice's treatment, we were able to learn about aspects beyond her clinical condition. We got to know her dreams, desires, autonomy and a teenager who overcomes day after day the difficulties imposed on her by the genetic syndrome.

We also got to know her health history, professionals, procedures and interventions that helped her get to this point and certainly helped to achieve all of her goals.

Based on the outcome of the interdisciplinary assessment, Alice was enrolled in a regular public school and is following the curriculum and teaching methods presented very well. She is also developing well in social interactions, without the need for additional pedagogical support.

Additional genetic tests were requested to continue the genetic investigation and clarify the diagnosis.

All professionals agreed on the need to continue regular follow-ups with a medical team: orthopedics, otorhinolaryngology, pediatrics and genetics. Interventions with motor physiotherapy and speech therapy were maintained and Alice began psychological monitoring, with an emphasis on guidance for parents.

The family received guidance from social services about their legal benefits and ways to seek medical and therapeutic support.

After returning to the orthopedist, she is preparing for definitive spinal surgery due to the worsening of the scoliosis.

At Day Hunter, once assessed, the patient is monitored from time to time. For Alice, we suggested returning within a year for reassessment.

As presented above, Alice's interdisciplinary evaluation process took into account her multisystemic needs, making it possible to get to know her and delve deeper into aspects beyond her clinical condition. It was an assessment that demonstrated much of her potential.

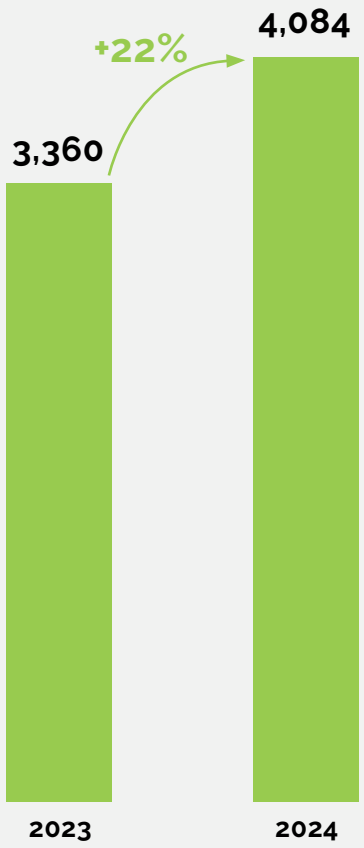
DAY HUNTER: IMPACT AND TRANSFORMATION IN THE RARE DISEASE JOURNEY

Transforming lives with multidisciplinary care

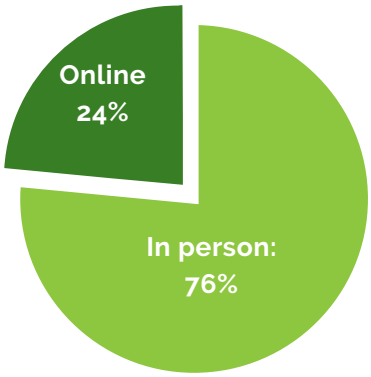
The Day Hunter Program, since its creation in 2014, has continuously evolved, expanding its reach and impact. Between 2019 and 2023, the number of consultations increased significantly, reflecting Casa Hunter's commitment to multidisciplinary care for patients with rare diseases. In 2019, 730 consultations were carried out; in 2024, this number jumped to an impressive 4,084 consultations, showing how much the program has consolidated itself as an important support in the patients' journey.

Day Hunter Expansion and Growth

Until 2022, Day Hunter's significant growth was driven by expansion to other capitals: Rio de Janeiro (2018), Salvador (2020) and Goiânia (2022). However, even without new regional expansions, the program's progress remains robust, with an annual increase of over 20%.



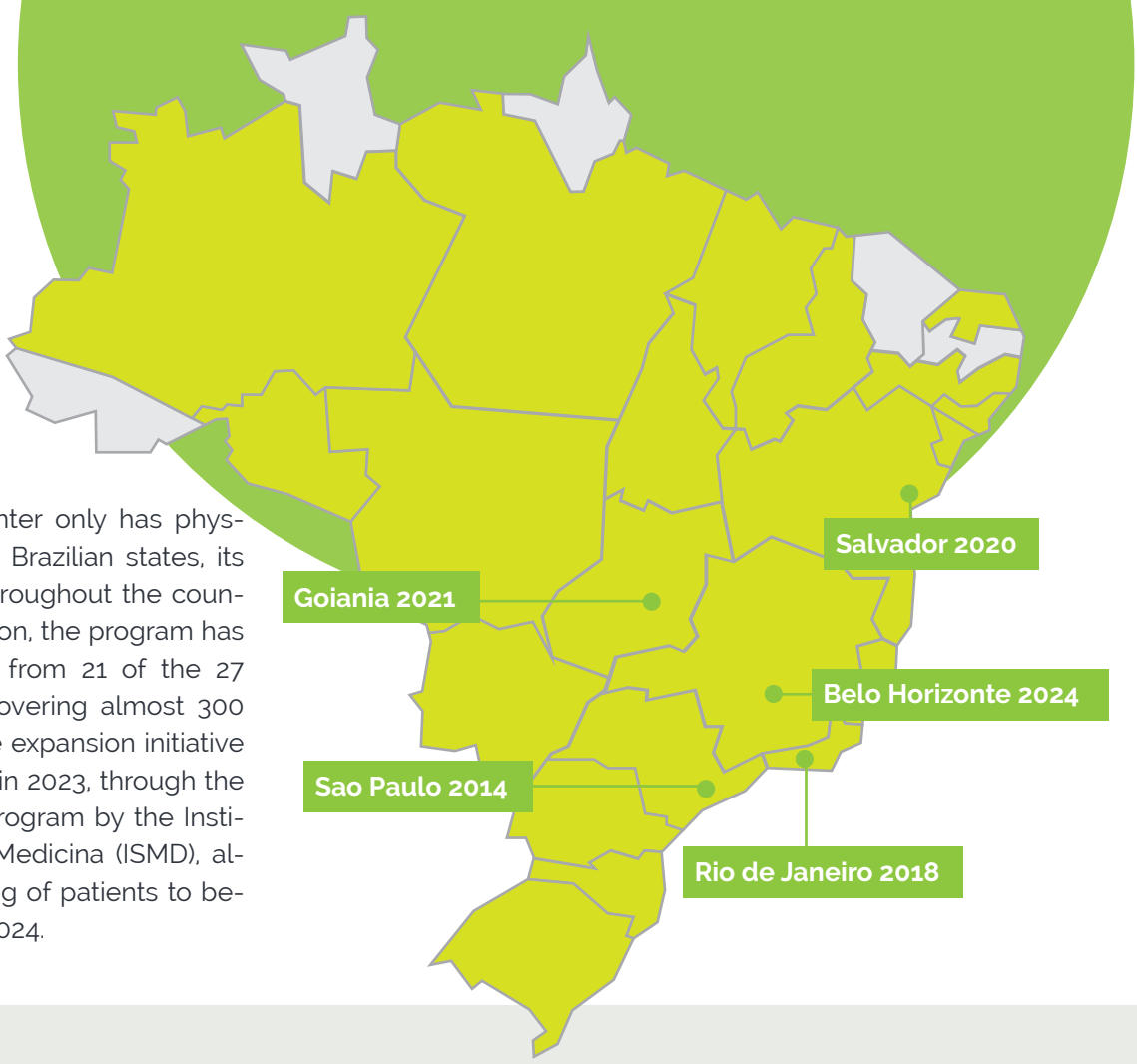
Online service: expanding access



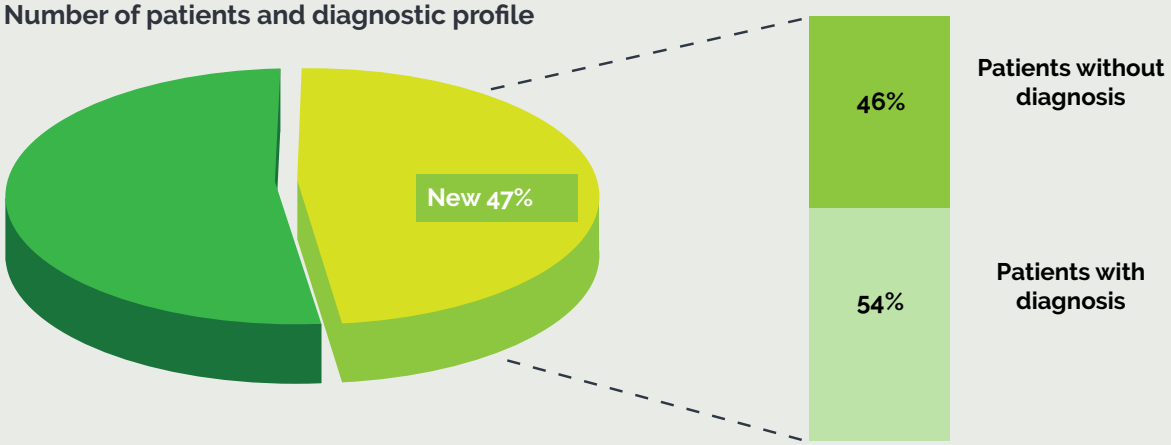
The online consultation modality, initiated during the Covid-19 pandemic, proved to be an essential tool for expanding access to the program. This strategy made it possible to reach patients who, due to geographical distance, would not be able to participate in the program in person, strengthening the impact of Day Hunter in remote regions.

National reach

Although Day Hunter only has physical units in some Brazilian states, its impact extends throughout the country. Since its creation, the program has received patients from 21 of the 27 Brazilian states, covering almost 300 municipalities. The expansion initiative to Belo Horizonte in 2023, through the adhesion to the program by the Instituto Superior de Medicina (ISMD), allowed the mapping of patients to begin at the end of 2024.



Number of patients and diagnostic profile



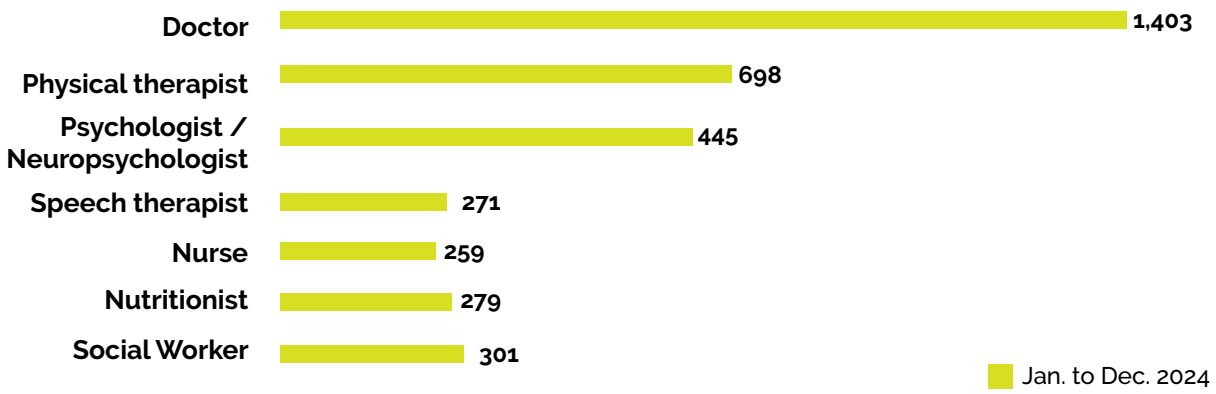
By 2024, more than **900 patients** were served by the program, with almost half accessing Day Hunter for the first time. Among these new patients, approximately **46%** sought a diagnosis or diagnostic confirmation, while the others already had an established diagnosis and were looking for guidance on managing the disease and ways to access the health system.

In addition, the program has already provided care to patients with **240 different rare diseases**, and, each year, around 20 new diseases are added to the list of cases treated. This growth reflects the complexity of the world of rare diseases and reinforces the importance of Day Hunter as a reference center in supporting these patients. The complete list of diseases covered can be found on the last pages of this magazine.

Medical specialties and healthcare demands

Medical assessment continues to be the most requested service within Day Hunter. Consultations in other specialties vary according to the specific needs of each pathology, reflecting the diversity of profiles served.

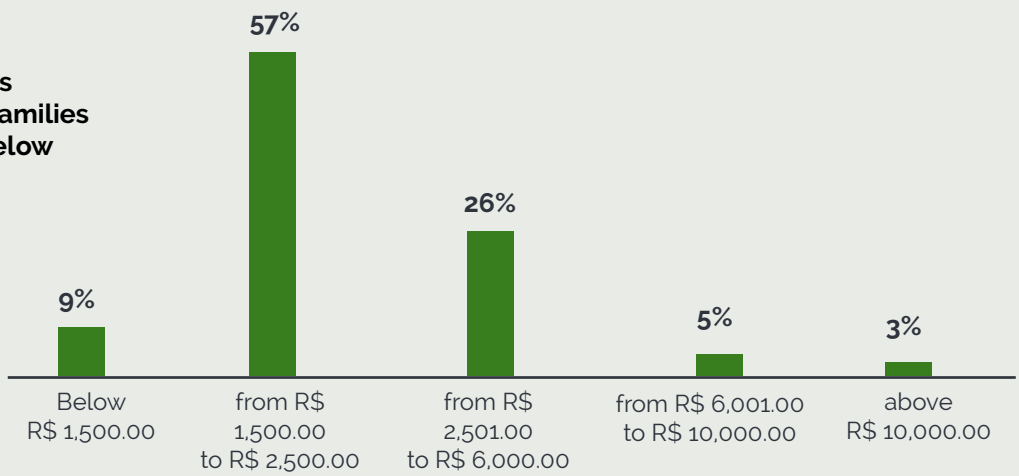
Consultations by specialty



Socioeconomic profile of families served

The socioeconomic survey of families assisted by Day Hunter reveals that 66% have an annual income of less than R$2,500, and the average family size is four people, including the patient. This means that, by serving these patients, the program directly impacts more than 3,600 people, expanding its social relevance.

2/3 of the patients treated are from families with an income below R$2,500.00



A program in constant evolution

Day Hunter continues to consolidate its role as a reference in the care of patients with rare diseases in Brazil. Its growth, even without new regional expansions, demonstrates the relevance and need for the program. Expanding access through online services and presence in different states demonstrate the positive impact of the initiative, which continues to transform lives and offer essential support to thousands of families.

Free care for dozens of patients every month

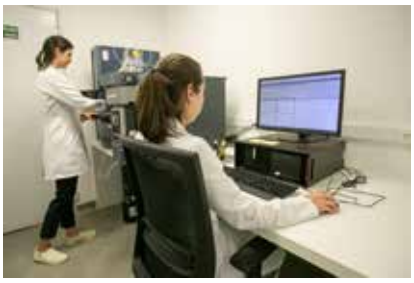
Casa dos Raros: transforming the lives of patients with rare diseases

An inspirational model for Casa dos Raros, the Day Hunter program is changing the reality of patients with rare diseases by providing fast and accurate diagnoses

In less than 100 days, patients who have been waiting for years for answers receive the long-awaited diagnosis, essential to begin treatment for rare diseases, which affect millions of Brazilians. Opened in February 2023 in Porto Alegre (RS), Casa dos Raros stands out as the first center in Latin America to offer comprehensive care, education and research on these diseases, completing 2 years of significant impact.

Since its opening, the facility has provided free care to dozens of patients, mostly children and adolescents, offering a multidisciplinary team, including geneticists, physiotherapists, psychologists, among others. Patients like Miguel Martiny Ferreira, who had been waiting for a genetic diagnosis for five years, finally received the answer through Casa dos Raros. "I thought we would never be able to get the test for Miguel," says his mother, Jessica Patricia Martiny.

Initially, the service was offered through the registration of patients or their family members who expressed interest in the service. In the next step, an



Casa dos Raros, for the rare

agreement was signed with the Government of the State of Rio Grande do Sul. For 42 months, 1,050 patients will be referred to Casa dos Raros, totaling an estimated 31,500 appointments. Since the beginning of this agreement, more than 900 patients have already passed through the center.

An innovative model

Casa dos Raros was created to propose a differentiated model for serving people with rare diseases. It is estimated that 30% of patients will not reach the age of 5, since many of these diseases are progressive and disabling. On the other hand, the journey to achieve a diagnosis and effective treatment can take up to 10 years, involving different professionals and even having to cross the country in search of specialists or procedures that are often expensive and sophisticated.

The institution has 51 employees and was the first in the state to offer the complete exome exam through the SUS, allowing the identification of genetic diseases with high precision. In addition, it promotes research and education projects, with 29 training events held to date and will soon begin testing new medicines.

HOW TO BE SERVED BY CASA DOS RAROS:

- Register your interest on the Casa dos Raros website (www.cdr.org.br) by filling out a form with your personal and case details.
- Patients can also be referred by the public health network.

HOW TO DONATE:

Casa dos Raros is maintained by donations and support, which can be made via https://doe.cdr.org.br/ in different payment methods, including recurring donations.

Virtual Library

In this edition of Day Hunter magazine we are pleased to start the Virtual Library section!

Here you will find tips on books, films, series, plays, documentaries, interviews, among other suggestions about the world of rare diseases.

VIVER É R

Our first recommendation is the series **"Living is Rare"** (1st and 2nd seasons). In this documentary we have the opportunity to learn a little about the lives of some of our patients, who give viewers the chance to follow their care routines, medical and specialist follow-ups, their dreams, goals, experiences and their families. The series moves us through its portrayal of the challenges, uncertainties and, above all, the victories of the protagonists!

Season 1

Ep. # 1

Laissa has SCA, **Spinal Cord Atrophy** , a degenerative disease that affects your body's movements. But this doesn't stop her from dancing ballet and being a Paralympic athlete.

Ep. # 2

Rafael Lellis is 31 years old, is a youtuber and has a rare disease called **Duchenne dystrophy**, which does not allow the muscles to develop correctly.

Ep. #3

The teenager **Theo Colker** shows strength to live with the fragility of one's own skin. EB, **Epidermolysis Bullosa** causes pain and wounds throughout the body.

Ep. # 4

Beatriz Valetim was – and will continue to be – an inspiration to friends, family and all those seeking to deal positively with life's challenges. Since the discovery of **Pulmonary Arterial Hypertension**, at age 20, until the last minute, she valued each victory, living one day at a time.

Ep. # 5

David Ishii is 3 years old and has a very rare condition: a type of dwarfism called **Achondroplasia**. His parents, Arielle and Daniel, have become true ambassadors for the cause.

Ep. # 6

At 2 years old, **Lara** was diagnosed with **ASMD**, a genetic syndrome that affects their growth and makes it difficult for them to eat. She shares her struggle with her mother and grandmother.

Ep. # 7

Miguel is a 17-year-old boy passionate about football who lives with a very rare disease: **Hemophilia** – a condition that affects blood clotting and causes numerous consequences.

RARO

Season 2

Ep. # 1

At 8 years old, the little one **Bryan** faces the limitations caused by **Mucopolysaccharidosis** (MPS), a disease that impairs his growth and breathing.

Ep. # 2

When **Mona** discovered that she was a carrier of **NMO**, a disease that took away the movement of her legs, her love for art was fundamental in finding ways to resist.

Ep. # 3

Dea is passionate about life and has made this passion her greatest fuel to deal with the diagnosis of a rare disease she received at the age of 45.

Ep. # 4

At 1 year old, **Leonardo** was diagnosed with **Cystic Fibrosis**, a rare disease that affects several organs and causes symptoms related to the respiratory system.

Ep. # 5

Regina Furuta faces an unpredictable disease, the **Paroxysmal Nocturnal Hemoglobinuria** (PNH), while dealing with the challenges of reentering the job market.

Ep. # 6

Vinicius, active and passionate about sports, was surprised at the age of 43 by **Pompe disease**, which affects the muscles and causes weakness, respiratory failure and other symptoms.

Ep. # 7

Marcela overcame a series of deprivations and a diagnosis of **Lupus** to build a career in law and now fights for rare people too.

Wonder is a film released in 2017 that tells the story of Auggie Pullman (played by Jacob Tremblay), who was born with a genetic syndrome called **Treacher Collins**. The film portrays the nuances of coexistence in the school environment, the challenges in forming bonds, as well as the family atmosphere. It's a moving story of love and overcoming.

Five Feet Apart tells the story of Stella Grant (Haley Lu Richardson), 16, who is different from other teenagers: due to **cystic fibrosis**, she spends a lot of time in the hospital, between treatments and medical monitoring. One day, she meets Will Newman (Cole Sprouse), a boy who has the same condition.

"Uma gota de esperança" tells the story of Larissa and her son, Théo, highlighting the journey to the diagnosis of **Glutaric Aciduria type** 1. The documentary addresses the struggle to understand this rare condition and highlights the importance of early detection to improve quality of life.

We'll be back soon with more suggestions. If you have any suggestions, please share them with us!

LIST OF RARE PATHOLOGIES REGISTERED IN HUNTER DAY CONSULTATIONS

A

Glutaric acidemia types 1 and 2
Methylmalonic Acidemia
L2 Hydroxyglutaric Aciduria
Adrenoleukodystrophy
Alkaptonuria
Medullary Aplasia
Acheiropodia
Takayasu arteritis
Arthrogryposis Multiplex Congenita
Severe Asthma
VACTERL Association
Ataxia
Friedreich's ataxia
Spinocerebellar Ataxia
Spinal Muscular Atrophy (SMA)

B

Beta-propeller protein-associated neurodegeneration (BPAIN)

C

Keratoconus
Craniosynostosis

D

Congenital disorders of glycosylation (CDG)
Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase (LCHAD) Deficiency
Biotinidase Deficiency
Cerebral creatine deficiency
Alpha 1 antitrypsin enzyme deficiency
Glucose-6-Phosphate Dehydrogenase (G6PD) Deficiency
GM3 Synthase Deficiency
Lysosomal acid lipase deficiency (LAL-D)
Dermatomyositis
Diabetes Insipidus
Nephrogenic Diabetes Insipidus
Hereditary Spastic Paraplegia
Primary Ciliary Dyskinesia (PCD) or Kartagener Syndrome
Dysferlinopathy / Dysferlin-type Muscular Dystrophy
Corpus callosum dysgenesis
Anhidrotic Ectodermal Dysplasia
Polyostotic fibrous dysplasia (McCune-Albright Syndrome)
Bone Dysplasia
Scapular Dystrophy
Myotonic Dystrophy or Steinert's Disease
Muscular dystrophy
Congenital Muscular Dystrophy
Ullrich's congenital muscular dystrophy or Ullrich's disease
Becker Muscular Dystrophy
Girdle Muscular Dystrophy
Duchenne Muscular Dystrophy (DMD)
Facioscapulohumeral Muscular Dystrophy (FSHD)
Oculopharyngeal dystrophy
SETBP1 haploinsufficiency disorder
Autoinflammatory disease – TRAPS
Addison's disease / adrenal insufficiency
Behçet's disease
Castleman's disease
Charcot-Marie-Tooth Disease (CMT) Type 2z or Neurodevelopmental Disorder Associated with the MORC2 Gene
Fabry disease
Huntington's Disease or Syndrome
Kennedy's disease
Landing's Disease or Norman-Landing Disease or GM1-Gangliosidosis
McArdle Disease or Glycogen Storage Disease Type V or Metabolic Glycogen Myopathy
Norrie's disease
Pompe disease or glycogen storage disease type II
Tarui disease or glycogen storage disease type VII
Tay-Sachs Disease B1 Variant
Motor Neurone Disease
Interstitial Disease or TTF1 Deficiency
Mixed connective tissue disease (MCTD)
Mitochondrial Disease
Neuromuscular Disease
Degenerative Neuromuscular Disease
Caroli's Disease or Syndrome
Calcium pyrophosphate deposition disease (CPPD)
Interstitial lung disease
Duplication of chromosome 22nq13

E

Mitochondrial Encephalopathy
Epilepsy
Myoclonic Epilepsy with Absences
Inborn Errors of Immunity
Amyotrophic Lateral Sclerosis (ALS)
Primary Lateral Sclerosis
Multiple sclerosis
Tuberous Sclerosis or Bourneville Syndrome

F

Ossifying fibroma
Cystic Fibrosis
Fructosemia

G

Eosinophilic granulomatosis with acute polyangiitis

H

Congenital Adrenal Hyperplasia (CAH)
Pulmonary hypertension
Hypophosphatasia (HPP)
Homocystinuria

I

Incontinentia Pigmenti or Bloch-Sulzberger Syndrome

L

Leukodystrophy
Leukoencephalopathy with Evanescent White Matter
Neuronal Ceroid Lipofuscinosis Type 2 (CLN2) or Batten Disease
Neuronal Ceroid Lipofuscinosis type 3 (CLN 3)
Neuronal Ceroid Lipofuscinosis type 6 (CLN6)
Neuronal Ceroid Lipofuscinosis type 7 (CLN7)
Lupus Erythematosus

M

Macrocephaly
Idiopathic Fibrosing Mediastinitis
Myasthenia Gravis and Congenital
Microcephaly
Chromosome 1 microdeletion
Chromosome 2 microdeletion
22q11 microduplication
Myopathy

Central Core Myopathy (CCM)
Congenital Myopathy
Congenital Centronuclear Myopathy (CNM)
Bethlem myopathy
Distal myopathy
Inflammatory myopathy
Myofibrillar Myopathy (MFM)
Native American Myopathy / STAC3-related Myopathy
Nemaline Myopathy
Mitochondrial Myopathies or Kearns-Sayre Syndrome
Myositis
Inclusion body myositis (IBM)
Congenital Myotonia
Mononeuropathy multiplex
Monosomy of Chromosome 7
Mucopolysaccharidosis (MPS)
Mucopolysaccharidosis (MPS) Type I / Hurler/ Hurler-Scheie/Scheie Syndrome
Mucopolysaccharidosis (MPS) Type II / Hunter Syndrome
Mucopolysaccharidosis (MPS) Type III / Sanfilippo Syndrome
Mucopolysaccharidosis (MPS) type III B
Mucopolysaccharidosis (MPS) type III C
Mucopolysaccharidosis (MPS) Type IV / Morquio Syndrome
Mucopolissacaridose (MPS) tipo VI / Síndrome Maroteaux-Lamy

N

Neurofibromatosis type 2
Neurofibromatosis
Neurofibromatosis type 1 (Von Recklinghausen's disease)
Neuromyelitis Optica
Peripheral neuropathy
Neuropathies
Neurosarcoidosis
Giant Congenital Melanocytic Nevus

O

Osteogenesis Imperfecta and Osteogenesis Imperfecta type 3

P

Autosomal Dominant Spastic Paraplegia
Polymyositis
Polyneuromyopathy
Polyneuropathy
Familial Amyloid Polyneuropathy (FAP) or Amyloidosis
Chronic Demyelinating Polyneuropathy (CIDP)
Peripheral Axonal Polyneuropathy
Sensory-Motor Polyneuropathy
Porphyria
Hepatoerythropoietic Porphyria
Pseudo Hypoparathyroidism

R

Retinitis Pigmentosa

S

Pierre Robin sequence
Cardio-cranial syndrome, Pfeiffer type / Pfeiffer Singer-Zschiesche Syndrome
Cutaneous Cardio-Facio Syndrome (CFC)
Cerebrocostomandibular Syndrome
Craniofaciocutaneous syndrome Chromosome Duplication Syndrome 7Q
Chromosome 4q deletion syndrome
Andersen syndrome
Angelman syndrome
Arnold Chiari Syndrome
Ataxia-Telangiectasia Syndrome
Bainbridge-Ropers Syndrome
Bartter syndrome
Beals Syndrome or Congenital Contractural Arachnodactyly
Beckwith-Wiedemann syndrome
Bloom Syndrome (Genetic mutation in the BLM gene)
Borjeson-Forssman-Lehmann Syndrome
Aminiotic Band Syndrome
CHILD Syndrome (Congenital Hemidysplasia with Ichthyosiform Erythroderma and Limb Defects)
Coffin-Siris Syndrome
Doose syndrome
Down Syndrome / 21 Trisomy
Dravet syndrome
Eaton Lambert syndrome
Edwards Syndrome / 18 Trisomy
Ehlers-Danlos syndrome
Emery Dreifuss syndrome
FIRES Syndrome (Febrile Infection-Related Epilepsy Syndrome)
Goldenhar Syndrome
Follicular ichthyosis, alopecia and photophobia syndrome (IFAP)
Jacobsen syndrome or 11q Deletion Syndrome or Monosomy 11q
Jeune Syndrome or Asphyxiating Thoracic Dystrophy
Klippel Trenaunay syndrome
Larsen syndrome
Leigh syndrome
Lesch-Nyhan syndrome
Lewis-Sumner syndrome
Marfan syndrome
22q11.2 Microdeletion Syndrome / DiGeorge Syndrome / 22q11.2 Deletion Syndrome / Velocardiofacial Syndrome
Distal 17p13.1 microdeletion syndrome
1q21 microduplication syndrome
22q11.2 microduplication syndrome
Moebius syndrome
Niemann-Pick Syndrome or Disease
Noonan syndrome
QRICH1-related intellectual disability-chondrodysplasia syndrome
Pfeiffer syndrome
Pitt-Hopkins syndrome
Prader-Willi syndrome
Rett Syndrome
Sotos Syndrome
Stickler syndrome
Sturge-Weber syndrome
Tatton-Brown-Rahman syndrome
Townes-Brocks syndrome
Troyer's syndrome
Turner syndrome
Walker-Wauburg Syndrome or Muscle-Eye-Brain Dystrophy
West Syndrome
Williams Syndrome
Wilson syndrome
Ring chromosome 18 syndrome
Hyper IgE Syndrome
Fragile X syndrome
Ellis Van Creveld Syndrome
Hunter-McAlpine Syndrome

T

Alpha Thalassemia
Tetrasomy 18p
Tetrasomy of 15q 21q 23
Partial tetrasomy 5q
Tyrosinemia
Opitz Trigonocephaly
Partial trisomy of the short arm of chromosome 8

V

Vasculitis C ANCA
Vasculitis of the PNS

X

Xanthomatosis
Cerebrotendinous Xanthomatosis

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