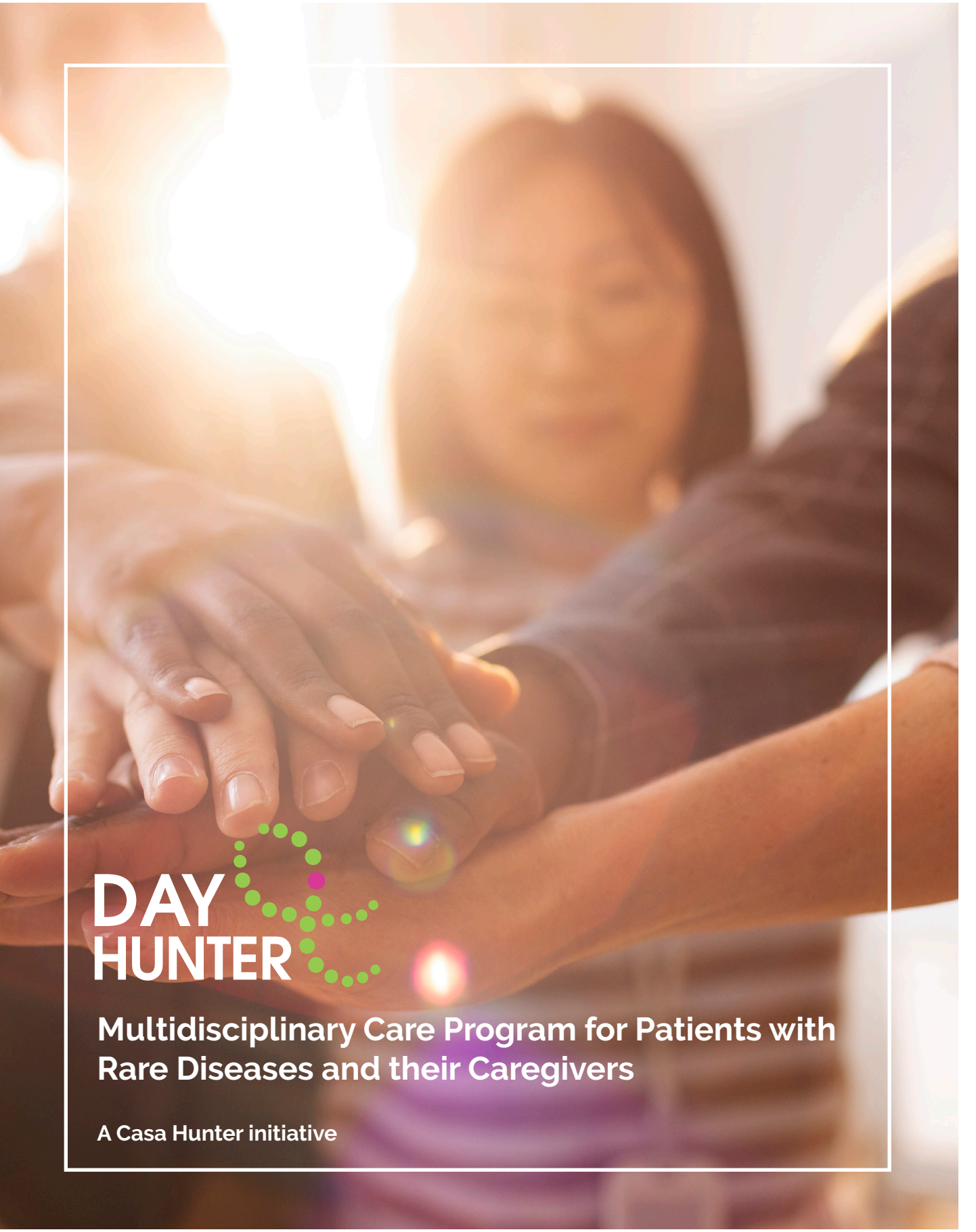


DAY HUNTER

Multidisciplinary Care Program for Patients with Rare Diseases and their Caregivers

A Casa Hunter initiative

EDITORIAL

It is with great joy we witness the formal presentation of the Day Hunter Program in this magazine. This is an innovative and successful initiative to help with the offer of quality care for people with rare diseases and their caregivers.

Here we talk about the care routine of not only 13 million Brazilians, but more than 50 million people who find themselves immersed in trying to find diagnosis, treatment and professionals prepared to care for their family and friends.

This model is bold, but time has proven that the idea is not only feasible, but also capable of producing quality service and savings for public coffers. An idea of exchange and partnership with centers of quality and innovation.

The certainty that we have found the way lies in the completeness and quality of the partner institutions involved. And also in the return of patients and caregivers, our most valuable companions in this journey. To everyone, thank you for your trust and investment in the program.

On behalf of myself and Fernanda, my wife and co-creator of this program, I state the commitment to continue with this project and move forward with innovative proposals focused on the patient, such as the House of the Rare - a dream that was born and managed with great care in the Day Hunter program.

We keep moving forward!

Antoine Daher

INDEX

ORIGIN OF DAY HUNTER

Hunter Day is an innovative program that aims to provide multidisciplinary guidance and support to patients with rare diseases. Its main objective is to create a **replicable model** of guidance and referrals that covers the entire journey from diagnosis to treatment.

Hunter Day offers essential information for the care journey, including referrals to specialists, ongoing monitoring, education and clarification of doubts for both family members and patients themselves.

In the Brazilian scenario, few rare disease centers offer multidisciplinary care, in which the patient is evaluated by professionals from different specialties at the same time. In many cases, the patient is referred to specialists, facing long waiting lists and misinformation about rare conditions. Hunter Day has stood out not only for filling this gap, but also for playing an important educational role in training healthcare professionals, especially in institutions with staff linked to universities.

The story behind the creation of Day Hunter

In 2012, Antoine Daher was faced with a new reality when he discovered that his son, Anthony, suffered from a rare disease known as Hunter Syndrome (Type II Mucopolysaccharidosis). Faced with this adversity, Daher eventually overcame it and also became a reference for others, founding **Casa Hunter** and dedicating his life to helping people affected by rare diseases in Brazil.

The Day Hunter project emerged in 2014, two years after Anthony's condition was discovered. This initiative was born from a long and arduous journey undertaken by Antoine and his wife, Fernanda Daher, from diagnosis to the search for adequate treatment for their son.

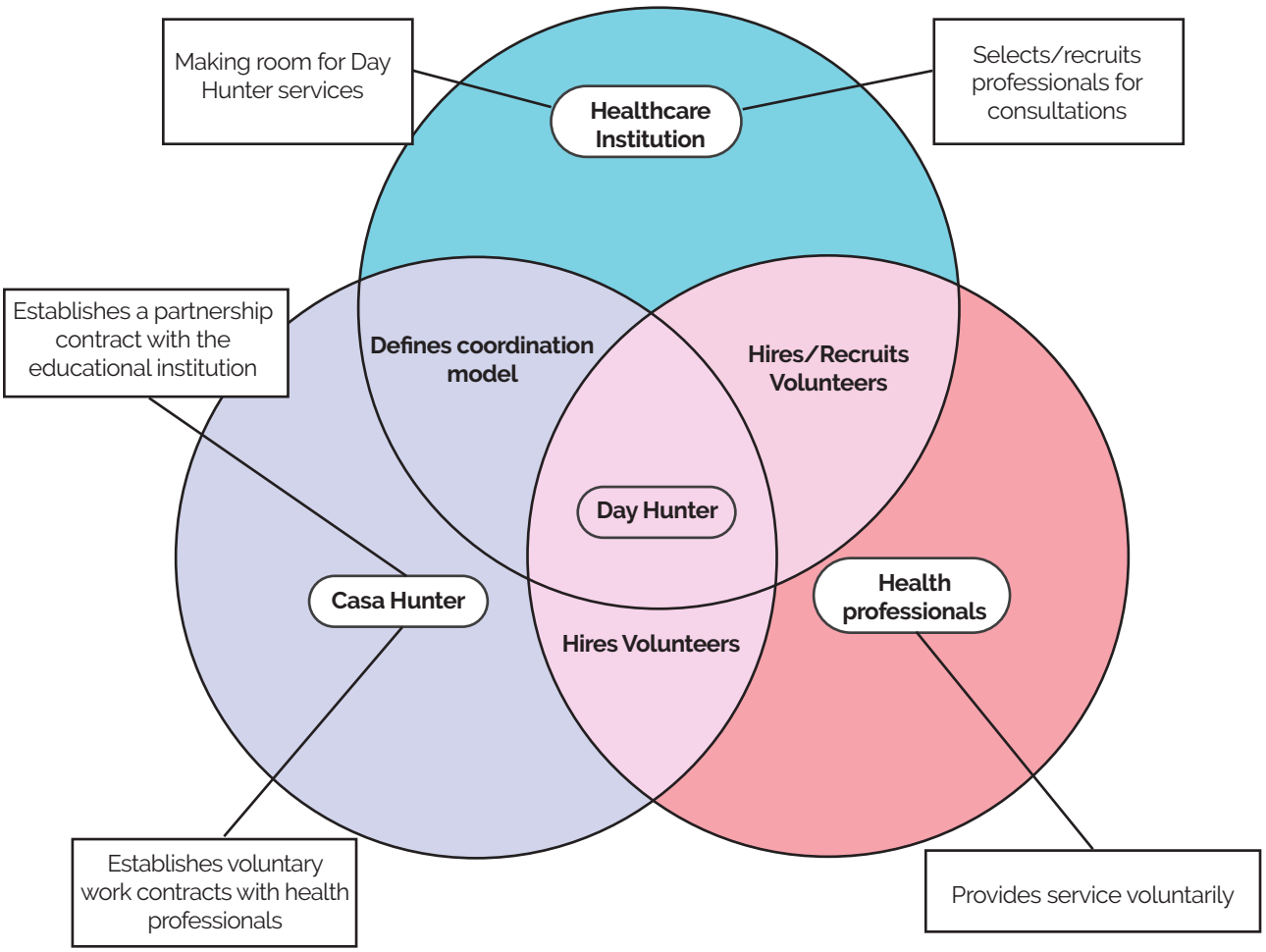
Rare diseases are often characterized by being multisystemic and presenting non-specific symptoms. This diagnostic complexity requires the intervention of health professionals from different specialties. Furthermore, the scarcity of trained health professionals and the lack of health centers specialized in these diseases make for a challenging scenario, which can prolong the process of obtaining a conclusive diagnosis for a period that commonly varies between 4 and 20 years.

Innovating service with a new model

The conception of a care model focused on rare diseases emerged in 2014 at the **Child and Adolescent Institute of the General Hospital of the Faculty of Medicine of the University of São Paulo,** under the coordination of Dr. Chong Ae Kim. At the time, specialists in different areas developed a multidisciplinary and interdisciplinary care system that could be applied in other locations.

Professionals such as speech therapists, physiotherapists, psychologists, neuropsychologists and pediatric dentists collaborated on the first implementation of Day Hunter . They studied the most prevalent diseases and their clinical manifestations to guide patients and families on their journey searching diagnosis, treatment and rehabilitation programs. Experience has demonstrated the urgent need for multidisciplinary teams specialized in rare genetic diseases.

Day Hunter Structuring Model



A journey from project to program

In 2017, the Institute of Genetics and Inborn Errors of Metabolism (IGEIM) in São Paulo, led by Dr. Ana Maria Martins, began hosting Day Hunter services. The coordination of the program was handed over to Regina Bernardes Ferreira El Khoury, who also participated in the development of the service model. In the expansion of Day Hunter in São Paulo, the team now has a social worker to meet the growing demands for guidance in this area.

In 2018, the initiative expanded to Rio de Janeiro and began providing care at the neuromuscular diseases service at the Gaffrée and Guinle University Hospital, under the coordination of Dr. Karina Lebeis.

The program extended to Salvador, Bahia, in 2020, where a partnership was established with the Bahiana School of Medicine and Public Health, under the coordination of Dr. Marcela Câmara Machado Costa.

The program arrived in the Midwest of the country in 2021, with the signing of a partnership with the Department of Rare Diseases of the General Hospital of the Federal University of Goiás, coordinated by Dr. Lusmaia Damaceno Camargo Costa. The most recent expansion took place in Belo Horizonte, through the adherence to the program by the Higher Institute of Medicine (ISMD), founded by Dr. Raquel Vilela.

Partner institutions

IGEIM
INSTITUTO DE GENÉTICA E ERROS INATOS DO METABOLISMO

HU GAFFRÉE E GUINLE
UNIRIO

BAHIANA
ESCOLA DE MEDICINA E SAÚDE PÚBLICA

HC
Hospital das Clínicas
UFG

FACULDADE ISMD

Dr Chong Ae Kim

Dr Ana Maria Martins

The history of Day Hunter is directly linked to two medical references in rare diseases in the State of São Paulo: Chong Ae Kim and Ana Maria Martins. The two geneticists participated in the process of creating the model, now present in five Brazilian states.

For Dr. Chong, from the Children's Institute (ICr/HCFMUSP) of the General Hospital of São Paulo, the program materialized her dream of implementing a model of comprehensive and multidisciplinary care. "The care of our patients with genetic diseases by a multidisciplinary team was a dream. Although there is a multidisciplinary team in several other medical specialties, it did not exist for rare diseases", recalls the doctor.

For Dr. Ana Maria Martins, who opened the doors of the Institute of Genetics and Inborn Errors of Metabolism - (IGEIM) to the Day Hunter in São Paulo, the importance of multidisciplinary work is undeniable. "The doctor is essential for the diagnosis, but it is the guidance and care provided by professionals in the multidisciplinary team that help and change patients' daily lives, for a better quality of life", she says.

Service flow at Day Hunter

At Day Hunter the arrival of patients is the result of medical referrals, recommendations from institutions or the result of internet searches.

EVALUATION

Initially, families and patients are carefully welcomed, followed by a comprehensive assessment. This assessment, which can be completed in a single day or through scheduled sessions, is adaptable and in certain cases can be conducted online. The team meets to analyze each case individually, establishing personalized guidelines, appropriate therapeutic protocols and determining the necessary referrals.

GUIDANCE

After the evaluation, the patient and family members are advised about the disease, treatment and therapeutic approaches. Psychology professionals offer special sessions, helping them face the frequent challenges on this journey. In addition, information is provided on how to access SUS and other available services, as well as clarifications on the rights of patients with a rare disease. When necessary, there are referrals for tests, genetic counseling or other services.

MONITORING

There is a commitment to maintaining regular contact to monitor patient progress and the implementation of guidance provided by the team. This stage may include necessary re-evaluations and adjustments. Each patient is monitored at least once a year, with the possibility of online sessions. During this period, relevant information is collected to help analyze the evolution of the disease over time.

Cases and Testimonials from Patients and Caregivers

Day Hunter Goiânia

Mikaely, 13 years old, resident of Balsas, Maranhão, had chronic respiratory problems since he was 7 months old. She was admitted to Day Hunter Goiânia after a series of complications and malnutrition. The team carried out evaluations with doctors, psychologists, nutritionists and a social worker, as well as various diagnostic tests. After a hospital stay, the group discussed the final diagnosis (Bronchiectasis Secondary to Post-infectious Bronchiolitis Obliterans) with the family and also the disease management plan. She began treatment with specific medications, physiotherapy, night ventilation, oxygen therapy at home and diet, leading to improvements in lung function and weight gain.

> *We came to the right place, one that was able to really tell us what she had. I am very grateful to the team that welcomed us and is treating her. It was a very good experience. We were well attended to from the moment we arrived by all the professionals we had contact with", says Mikaely's mother.*

Day Hunter São Paulo

aRafael's mother, from Espírito Santo, came to Casa Hunter looking for a diagnosis for her 9-year-old son. Since pregnancy, she suspected a syndrome due to changes in her ultrasound. The team carried out a thorough assessment, covering social, medical and neuropsychomotor aspects. Virtual consultations with a social worker and geneticist preceded the in-person consultation, where it was possible to identify facial dysmorphisms, specific physical characteristics and developmental delays. Previous genetic testing had been inconclusive. Therefore, the Day Hunter team, in partnership with a private laboratory, carried out Complete Exome Sequencing, which revealed two changes: deletion in chromosome 6 and duplication in chromosome 14. This allowed for a correct diagnosis and an effective treatment and follow-up plan. With this, the Day Hunter team offered detailed guidance, referrals to specialists, specialized educational support, adapted therapies, psychotherapeutic and speech therapy support. An interdisciplinary report also allowed the mother to access essential benefits and information.

> *It's inexplicable how much this has changed our lives. Gratitude to everyone who, directly or indirectly, is part of the project. What you are doing for the lives of our rare people and for our lives is priceless", thanks Rafael's mother.*

Day Hunter Rio de Janeiro

A 69-year-old patient from Rio de Janeiro, Jucéa had experienced numbness in her upper limbs and choking since 2011. In 2017, an electromyography confirmed the condition of Amyotrophic Lateral Sclerosis (ALS), whose symptoms worsened, leading to falls, loss of speech, gait, dyspnoea, among other symptoms. Despite using specific medication, there was no improvement until April 2021. At the end of the same year, the Day Hunter team took over the treatment. The patient had grade 2 muscle strength, in addition to mobility difficulties. Due to the loss of strength of the orofacial muscles and alveolar hypoventilation, nocturnal ventilation and lung exercises were recommended, but limited financial resources prevented adherence. After presenting bradycardia during physical examination, cardiological screening was performed. Thus, the Holter exam identified complete atrioventricular block (AVB), requiring the use of a pacemaker. Speech therapy and nutrition identified severe dysphagia, and a soft diet and alternative feeding route were suggested. A gastrostomy was performed in 2022, leading to weight gain and improved swallowing. Psychological support was also essential. Currently, the patient appears lucid and oriented, clinically stable, communicating by writing on a cell phone.

"The first consultation got me emotional. I have never been treated so well anywhere! Everyone is so affectionate! We all have our problems, but this team leaves them out there and only gives love, affection and care. This made me calmer in the face of the problems I have faced since 2011."

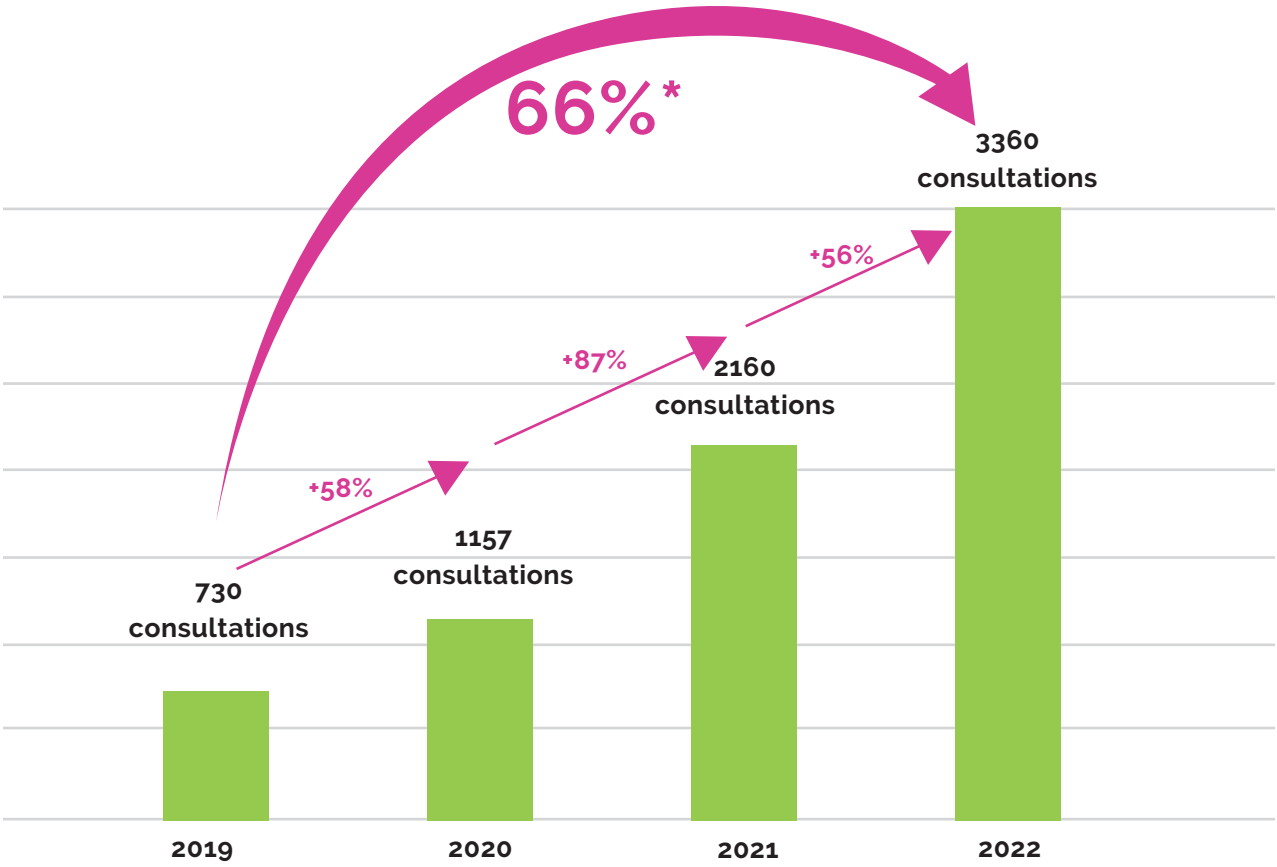
Day Hunter Salvador

The patient Letícia, resident of Camamu, Bahia, had motor delay since she was 7 months old, unable to stand or sit without support. At 14 months, she would only crawl. Spinal Muscular Atrophy (SMA) Type 2 was diagnosed at two years of age. She arrived at Day Hunter Salvador in 2020, after being referred by the neuropediatrician on suspicion of the rare disease, which was confirmed by genetic testing. Since then, she has undergone regular monitoring with the multidisciplinary team, with returns every four months. Her care plan includes disease-modifying therapy associated with multidisciplinary rehabilitation (Physiotherapy, Psychology, Speech Therapy, Nursing and Nutrition). Drug therapy began at age 3, with excellent adherence and regularity to treatment by the family, who created tools to help the child adapt to daily activities (such as a parapodium, equipment used to help maintain posture, made of wood and support bars with PVC pipes). On the 10th dose of medication, Letícia recorded gains in mobility and breathing.

Everywhere we went, no one understood what SMA was and here we truly felt welcomed. The professional guidance and care are incredible. I just have to say thank you", says the patient's mother.

The Day Hunter in numbers

Total of Consultations



(*) Compound growth per year

The expansion of the program has been notable, with a **growth of almost 5 times** in the number of consultations over the last 4 years. This increase is due not only to the opening of new units, but also to the increase in services in each of them.

Virtual services, which played a crucial role in expanding the reach of the Day Hhunter program, were introduced in 2020 in response to the Covid-19 pandemic. The objective was to guarantee the continuity of follow-up services, in addition to providing essential guidance on how to maintain therapies and rehabilitation amid the new conditions imposed by the pandemic.

Rare Diseases already covered by Day Hunter

Over the past four years, Day Hunter has registered consultations related to almost **200 different rare diseases**. Among the pathologies that required a greater volume of consultations are those that require more frequent interventions.

20 Pathologies with the highest number of consultations in 2022

Amyotrophic Lateral Sclerosis (ALS)	Amyloidosis
Spinal Muscular Atrophy (SMA)	Pompe disease
Duchenne Muscular Dystrophy (DMD)	Charcot-Marie-Tooth Disease (CMT)
Girdle Muscular Dystrophy	Sotos Syndrome
Cystic Fibrosis	Arthrogryposis Multiplex Congenita
Myotonic Dystrophy or Steinert's Disease	Williams Syndrome
Spinocerebellar Ataxia	Dravet syndrome
Mucopolysaccharidoses (MPS) Type I, Type II, Type III, Type IV and Type VI	Congenital Muscular Dystrophy
DiGeorge Syndrome	Huntington's disease
Myasthenia Gravis and Congenital	Tuberous Sclerosis (Bourneville Syndrome)

Number of patients treated

Since its inception in 2014, the **Day Hunter** program has carried out assessments on more than **five thousand patients**. In 2022, considering the gradual resumption of activities and services that were normalized due to the pandemic, the program provided assistance to approximately **900 individuals**.

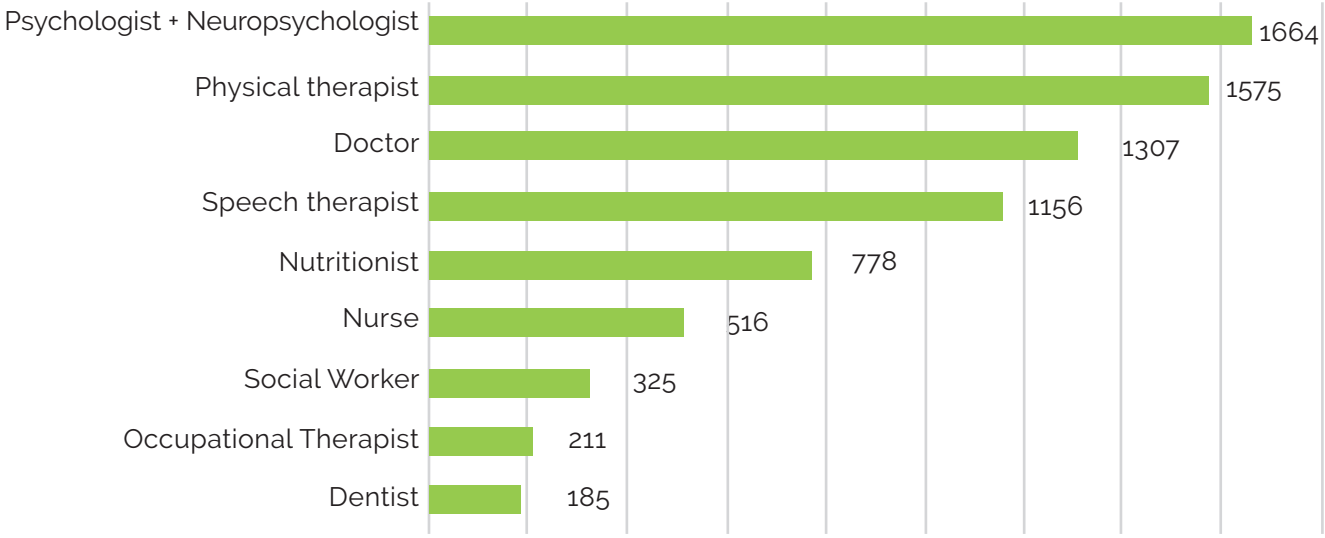
The work of the multidisciplinary team

Day Hunter teams are made up of several health professionals, including doctors and other specialists, favoring a **multidisciplinary approach** that is extremely necessary for diagnosis, guidance and referrals of patients with rare diseases. The composition of these teams may vary according to the clinical profile of the patients and the location of the consultation.

Multidisciplinary care is crucial both for rare diseases that require pharmacological treatment and for those for which there is still no specific treatment. In these latter cases, orientation on rehabilitation is crucial for improving the patient's quality of life.

Between 2019 and 2022, a significant number of consultations were registered, particularly in the areas of Psychology, Physiotherapy and Speech Therapy. These are specialties that often require more than one appointment to welcome and evaluate patients. In 2022, Day Hunter already had a team of more than 30 dedicated professionals, working in the five service units.

Consultations by Specialty (2019 to 2022)



Regardless of the pathology, the **welcoming care** provided to patients and their families by psychologists has, since the beginning, been a fundamental element of Day Hunter. Across all staff, there is an ongoing commitment to ensuring patients and caregivers feel truly welcomed, understood, respected and supported.

Interdisciplinary practice in the clinic is beneficial not only to patients, but also to working professionals. The exchange of ideas and information promotes the expansion of knowledge and the improvement of clinical reasoning among employees. This means training and qualification of more health professionals in rare diseases, as well as generating data that can contribute to initiatives aimed at the cause.

Who makes Day Hunter happen

Doctor: has the crucial role of analyzing the complexity of each patient to establish accurate diagnoses, especially in the case of rare diseases. They explain the details of the disease and its implications to patients and families, highlighting the importance of multidisciplinary monitoring and genetic counseling when appropriate.

Psychologist and Neuropsychologist: offer welcoming care, support and neuropsychological assessments to help with the diagnosis and clarification of common disorders and comorbidities in several rare pathologies.

Physiotherapist: provides diverse and specific treatment for each pathology, explains diseases and guides the use of equipment. Works as a team on complex cases and provides guidance on physical activities and care at home and school, as well as access to health services.

Speech therapist: treats speech and language delays, speech and swallowing disorders. Promotes adaptations for safe eating in cases of dysphagia, adjusting strategies as the disease progresses. They also seek to interact with professionals who will continue the patient's rehabilitation.

Nurse: advises on risks of falling, skin injuries, choking and care with gastrostomy or tracheostomy. They also offer advice on the use of orthoses, home adaptations and medication administration, in addition to guiding families to look for local health centers.

Occupational Therapist: focuses on improving patients' independence and safety in daily tasks, creating personalized strategies and adaptations. Access to the rehabilitation network is a challenge, leading the therapist to advise on rehabilitation at home, with accessible resources.

Nutritionist: carries out anamnesis and nutritional assessment, taking into account lifestyle and eating habits, symptoms and difficulties of the disease. The guidelines are personalized, aiming for quality and balance in the diet. The professional monitors patients who are malnourished or in risk situations.

Social Worker: plays a transversal role, assessing the socioeconomic situation of families, providing guidance on social benefits. They provide information on inclusion laws for children with disabilities, and work to facilitate access to social and health services. They also look for institutions and assistance services, to provide guidance on places where the patient can seek care.

Dentist: performs oral and dental assessment, offers guidance on oral hygiene and advises according to the patient's needs and disabilities. Patients can be referred for specialized care when necessary.

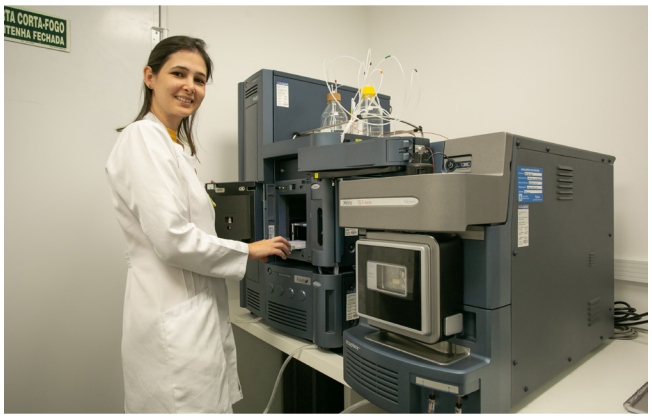
DAY HUNTER & HOUSE OF THE RARE

Over its 9 years of existence, Day Hunter has accumulated experience and refined its approach. Today it is clear that interdisciplinary care provided by a multidisciplinary team brings benefits to patients, their families and the doctors and specialists themselves.

What began as a project now has the solidity of a consolidated program, driving gains in knowledge about rare diseases, the development of strategies and the approach to treatment and rehabilitation, in addition to contributing to the training and improvement of new professionals.

Although Day Hunter does not intend to replace the public system, its mission is to contribute to the expansion of a service model that has been proven to be effective and sustainable. Healthcare professionals who have gone through Day Hunter become advocates and promoters of this model, which can be adopted in other health institutions.

Antoine Daher
and Fernanda Daher



Mariluce Riegel-Giugliani and
Prof. Roberto Giugliani

The effectiveness of Day Hunter and the ability to replicate its model were the inspiration for the creation of the House of the Rare , in Porto Alegre. An unprecedented initiative in Latin America, it was created with the purpose of developing an interconnected network dedicated to comprehensive care for people with rare diseases.

The main objective of **House of the Rare** is to offer comprehensive and multidisciplinary assistance, starting with rapid and accurate diagnosis, going through advanced treatments and clinical research focused on rare genetic diseases, and culminating with training and qualification of healthcare professionals to work in this specific field. All of this was possible thanks to the collaboration between **Casa Hunter**, with its founder Antoine Daher, and the **Instituto Genética para Todos** (Genetics For All Institute), with its creator Prof. Roberto Giugliani. Visit **www.cdr.org.br** and learn more about the House of the Rare.

LIST OF RARE PATHOLOGIES REGISTERED IN DAY HUNTER CONSULTATIONS

A

Glutaric acidemia types 1 and 2
Methylmalonic Acidemia
L2 Hydroxyglutaric Aciduria
Adrenoleukodystrophy
Amyloidosis
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 deficiency (LCHAD)
Biotinidase Deficiency
Cerebral creatine deficiency
Alpha 1 antitrypsin enzyme deficiency
Glucose-6-phosphate dehydrogenase (G6PD) deficiency
Lysosomal acid lipase deficiency (LAL-D)
Dermatomyositis
Diabetes Insipidus
Nephrogenic Diabetes Insipidus
Hereditary Spastic Paraplegia
Primary ciliary dyskinesia - PCD (Kartagener Syndrome)
Dysferlinopathy (Dysferlin-type Muscular Dystrophy)
Corpus callosum dysgenesis
Polyostotic fibrous dysplasia (McCune-Albright Syndrome)
Bone Dysplasia
Scapular Dystrophy
Myotonic Dystrophy (Steinert's Disease)
Muscular dystrophy
Congenital Muscular Dystrophy
Ullrich congenital muscular dystrophy (Ullrich 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 type 2z - CMT (Neurodevelopmental disorder associated with the MORC2 gene)
Fabry disease
Huntington's disease (Huntington Syndrome)
Kennedy's disease
Landing's Disease (Norman-Landing Disease/GM1-Gangliosidosis)
McArdle's Disease (Type V Glycogenosis/Gycogen Metabolic Myopathy)
Pompe Disease (Glycogenosis Type II)
Tarui Disease (Glycogenosis Type VII)
Tay-Sachs Disease B1 Variant
Motor Neurone Disease
Interstitial Disease (TTF1 Deficiency)
Mixed connective tissue disease (MCTD)
Mitochondrial Disease
Neuromuscular Disease
Degenerative Neuromuscular Disease
Calcium pyrophosphate deposition disease (CPPD)
Interstitial lung disease
Duplication of chromosome 22nq13

E

Mitochondrial Encephalopathy
Epilepsy
Inborn Errors of Immunity
Amyotrophic Lateral Sclerosis (ALS)
Primary Lateral Sclerosis
Multiple sclerosis
Tuberous sclerosis (Bourneville's disease)

F

Cystic Fibrosis

G

Eosinophilic granulomatosis with acute polyangiitis

H

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

I

Incontinentia pigmenti (Bloch-

Sulzberger Syndrome)

L

Leukodystrophy
Leukoencephalopathy with vanishing white matter
Neuronal Ceroid Lipofuscinosis type 2 - CLN2 (Batten Disease)
Lupus Erythematosus

M

Myasthenia Gravis and Congenital Microcephaly
Chromosome 1 microdeletion
Chromosome 2 microdeletion
22q11 microduplication
Myopathy
Central Core Myopathy (CCM)
Congenital Myopathy
Congenital Centronuclear Myopathy (CNM)
Distal myopathy
Inflammatory myopathy
Myofibrillar Myopathy (MFM)
Native American Myopathy (STAC3-related Myopathy)
Nemaline Myopathy
Mitochondrial myopathies (Kearns-Sayre Syndrome)
Myositis
Inclusion body myositis (IBM)
Congenital Myotonia
Mononeuropathy multiplex
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 IV (Morquio Syndrome)
Mucopolysaccharidosis (MPS) Type VI (Maroteaux-Lamy Syndrome)

N

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

O

Osteogenesis Imperfecta and Osteogenesis Imperfecta type 3

P

Polymyositis
Polyneuromyopathy
Polyneuropathy
Familial Amyloidotic Polyneuropathy (FAP) Amyloidosis
Chronic Demyelinating Polyneuropathy (CIDP)
Peripheral Axonal Polyneuropathy
Sensorimotor polyneuropathy
Porphyria
Pseudo Hypoparathyroidism

R

Retinitis Pigmentosa

S

Pierre Robin sequence
Cardio-Facio-Cutaneous Syndrome (CFC)
7q11. 23 Chromosomal duplication syndrome
Chromosome 4q deletion syndrome
Angelman syndrome
Arnold Chiari Syndrome
Ataxia-telangiectasia syndrome
Bartter syndrome
Beals syndrome (congenital contractural arachnodactyly)
Beckwith-Wiedemann syndrome
Bloom Syndrome (Genetic mutation in the BLM gene)
Borjeson Forssman Lehmann Syndrome
Aminiotic Band Syndrome
Doose syndrome
Down Syndrome (21 Trisomy)
Dravet syndrome
Eaton Lambert syndrome
Edwards Syndrome (18 Trisomy)
Ehlers-Danlos syndrome
Emery Dreifuss syndrome
Goldenhar Syndrome (Oculo-auriculo-vertebral Spectrum - OAVS)
Follicular ichthyosis, alopecia and photophobia syndrome (IFAP)
Jacobsen Syndrome (11q Deletion Syndrome - 11q Monosomy)
Jeune Syndrome (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)
1q21 microduplication syndrome
22q11.2 microduplication syndrome
Moebius syndrome
Niemann-Pick Syndrome (Niemann-Pick Disease)
Noonan syndrome
QRICH1-related intellectual disability-chondrodysplasia syndrome
Pitt Hopkins Syndrome
Prader–Willi syndrome
Rett Syndrome
Sotos Syndrome
Sturge-Weber syndrome
Tatton-Brown-Rahman syndrome
Troyer's syndrome
Turner syndrome
Walker Wauburg Syndrome (Muscle-eye-brain dystrophy)
Williams Syndrome
Wilson syndrome
Hyper IgE Syndrome
Fragile X syndrome
Hunter-McAlpine Syndrome

T

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

V

Vasculitis C
PNS vasculitis

X

Xanthomatosis
Cerebrotendinous Xanthomatosis

CASA HUNTER
ASSOCIAÇÃO DE DOENÇAS RARAS

Ombudsman
ouvidoria@casahunter.org.br

Contact us
casahunter@casahunter.org.br
pacientes@casahunter.org.br