

Mental Health Resources for JM Families

Chronic and rare disease is a challenging experience for anyone, and especially difficult on children and adolescents.

According to the 2013 Rare Disease Impact Report, 69% of patients with a rare disease experience depression and 82% of patients experience anxiety and stress.

In order to understand how our families cope with mental health, we collaborated with Andrea Knight MD MSCE, an expert in the autoimmune and mental health fields. We surveyed Cure JM patients and parents on the “Mental Health Needs of JM Patients and Potential Interventions” and found that 28% of JM adolescent and young adult patients reported depression and 33% reported anxiety. These percentages are two to three times higher than the 2016 national average of depression in adolescents (12.8%) and young adults (10.9%), according to the National Institute of Mental Health.

Researchers do not know whether mental health problems in JM are caused by the underlying illness (from inflammation and vasculitis), the treatments, genetics, or the challenge of living with a chronic and rare disease. But, we do know that patients, physicians, and researchers rank mental health as an important research priority for Cure JM.

Cure JM Foundation recommends that mental health assessment and treatment be an integral component of comprehensive care for children, adolescents, and adults with JM, and for patients in remission from JM. To this end, Cure JM Foundation is developing resources to educate physicians and patients about mental health issues in Juvenile Myositis. In addition, Cure JM is developing a plan to help pediatric rheumatologists screen for mental health issues and recommend further evaluation or treatment if warranted.”

ONLINE RESOURCES

For more information about:

- JM-Related Factors Leading to Depression and Anxiety
- The Signs of Depression
- The Signs of Anxiety
- How to Get Help
- Hotlines
- Insurance

visit www.curejm.org/fsn/mental-health-resources-article.php

Understanding the Underlying Genetics of Juvenile Myositis

Understanding the genetics of juvenile myositis is a research priority for Cure JM. In 2017, Cure JM partnered with the Center for Applied Genomics (CAG) at Children's Hospital of Philadelphia (CHoP) on a study to delve more deeply into the genetics of JM. CAG maintains a pediatric biobank, which contains biospecimen samples, like blood and tissue, along with medical information for those who have agreed to provide samples. To date, over 100,000 families have voluntarily donated samples into the biobank and shared medical and survey information. This massive store of information makes CAG's pediatric biobank the world's largest of its kind.

The Cure JM-invested research is led by Hakon Hakonarson, PhD, MD and Charly Kao, PhD. The team is using CAG's biobank, sequencing and genotyping technologies, and analytical tools to discover which genes associate with JM, and develop an understanding of why mutations in those genes can contribute to the development of JM. The researchers hope the results will lead to new drugs and new treatments that will improve the lives of those living with JM.

In order to conduct a thorough study, the team needed samples from both patients and their families. More samples mean more information, and more information improves the chances of understanding what causes JM. As of January, 2019, almost 100 Cure JM families have contributed to the study. During the last two National Family Conferences and several Walk Strong® events around the country, families filled out medical and family history paperwork and then gave blood or saliva. Samples were collected from JM patients, their siblings, parents, grandparents, and extended family.

"When conducting rare disease research, being able to collect samples from so many families is a unique opportunity," says Dr. Kao. "We were pleased that after just two visits to Cure JM conferences that we already collected samples from 300+ different individuals representing over 70 families. This speaks to the commitment, support, and enthusiasm of the Cure JM organization and their families."

Dr. Kao reports that they've made made progress in the Cure JM funded research, and plan to report early results in the coming months.

"We currently have encouraging preliminary results, and we are working on sequencing new families to replicate and search for other contributing genetic factors. Families where multiple members have a history of autoimmune disease or where more than one sibling or family member has/had JDM/JM are of particular interest and priority, since the genetic signals/contributions tend to be strongest in these settings. We anticipate at last some of these results will be available in June at the Annual Medical Conference and Family Education Event in Chicago, and we project to submit for publication later this year."



Hakon Hakonarson, PhD, MD



Charly Kao, PhD.

Autoantibodies in Juvenile Dermatomyositis

Antibodies are a normal part of the immune system which help fight infection by recognizing a unique protein from the harmful agent, called an antigen. Autoantibodies are antibodies that are directed against an individual's own proteins. Patients with inflammatory myositis often have autoantibodies which are specific to their disease, called myositis specific autoantibodies. They can also have autoantibodies that occur in myositis in addition to other connective tissue diseases, called myositis associated autoantibodies. The role autoantibodies play in inflammatory myositis is largely unknown. However, patients with the same myositis specific autoantibodies often have shared features of their disease. Knowing which patients have what myositis autoantibodies can be useful to doctors, because identifying who is at risk for certain disease features can help doctors better manage and treat their patients.

Dr. Sara Sabbagh's research in the Muscle Disease Unit in The National Institute of Arthritis, Musculoskeletal, and Skin Disease, National Institutes of Health, focuses on assessing the prevalence and associated disease features in children with myositis that have a myositis associated autoantibody called anti-Ro52. Anti-Ro52 autoantibodies are known to occur in adult patients with myositis. They can be seen alongside myositis specific autoantibodies, but also have distinct features in adult patients with myositis. Using 302 juvenile myositis patient samples and comparing to juvenile healthy controls, Dr. Sabbagh tested for these autoantibodies in patients with juvenile dermatomyositis by enzyme-linked immunosorbent assay (ELISA), which is a technique that detects and quantifies antibodies.

Her research revealed that anti-Ro52 autoantibodies were found in 14% of juvenile myositis patients overall and found to occur in higher rates in juvenile myositis patients with lung disease. In addition, patients with anti-Ro52 autoantibodies were found to have more severe disease overall than myositis patients who were negative for anti-Ro52 autoantibodies. Knowing which patients are at risk for certain disease features based on their autoantibody status can help doctors better manage their patients. Sometimes autoantibodies can determine what screening tests are appropriate for a patient. Other times, it can direct therapy choices. Overall, Dr. Sabbagh's data suggest that anti-Ro52 autoantibodies may impart clinical significance. This work was recently accepted for publication in *Annals of Rheumatic Disease*.

Meet Andrew Heaton, Cure JM CSO

The Cure JM family can anticipate some exciting and bold new directions, as Dr. Andrew Heaton moves into the second half of the first year of his appointment as Cure JM's first Chief Scientific Officer.



Andrew Heaton, PhD

Dr. Heaton brings a 30-year career in pharmaceutical discovery, development, and translational studies, and most recently served as the CEO and President of Heaton-Brown Life Sciences and Novogen North America. Dr. Heaton, ushered several oncology compounds from the laboratory through Phase I, II and III clinical trials.

According to Cure JM Executive Director James Minow, "Andrew has already had tremendous impact in further focusing our research priorities and strategic planning. He brings exceptional experience and a strong track record of innovation in the fields of biotech, drug development and business development. His dedication to finding treatments to rare and life-threatening diseases and compassion for patients and families make him the ideal choice to lead our research program to the next level of achievement."

The CSO serves as Cure JM's science and research leader, proactively driving the growth and effectiveness of Cure JM's grants and clinical research program. In addition, the CSO provides strategic oversight, guidance, and leadership to the Cure JM Research Priorities. Dr. Heaton will serve as key staff to Cure JM's Research Committee and Medical Advisory Board, and along with Research Committee co-chair Suzanne Edison and MAB member Dr. Lauren Pachman has developed the program for this year's Cure JM Medical Conference.

Dr. Heaton received his Bachelor of Science degree from the University of Tasmania, his doctorate from James Cook University of North Queensland, his executive MBA from Melbourne Business School, and completed a sabbatical at Imperial College, London. Dr. Heaton is the senior author on over 100, globally granted-patents, covering a wide range of molecular architectures and diseases and has published multiple papers in peer-reviewed scientific journals.

Cure JM Funding Juvenile Myositis Patient Registry, Biorepository through CARRA

Cure JM has provided financial support to several patient registries and biorepositories, and most recently has partnered with the Childhood Arthritis and Rheumatology Research Alliance (CARRA) to launch a multi-center JM patient registry. The CARRA JM patient registry is a database that collects information at the time of diagnosis and at regular intervals about how a patient is doing, medications used, and side effects. Pooling all this information lets CARRA researchers conduct larger studies that will ultimately help JM families make more informed decisions on treatment options.

According to Dr. Peter Nigrovic, chair of CARRA's Translational Research and Technology Committee, "There are many questions about JDM that require answers. For example, are there different types of JDM that need different approaches to treatment or are at risk for different complications? Can we predict which patient will respond to which therapy? How do some children enter long-term drug-free remission, when most autoimmune diseases last for a lifetime? Why does JDM occur at all?"

"The commitment from the Cure JM Foundation to fund a JDM Biobank in addition to the registry along with the willingness of patients and families to donate samples and clinical data, will allow investigators within CARRA and elsewhere to begin tackling these questions. We expect that this collection of data and biosamples will become a powerful resource for transformative research in JDM,"

This Cure JM supported CARRA JM patient registry went live in December 2017 at 18 CARRA sites, with expansion planned to a total of 75 sites in 2018.

In addition to the CARRA registry and biorepository, Cure JM continues to support the largest U.S.- based registry and biorepository at Dr. Lauren Pachman's lab at the Stanley Manne Research Institute/Lurie Children's Hospital in Chicago. This Cure JM supported CARRA JM patient registry went live in December 2017 at 18 CARRA sites, with expansion planned to a total of 75 sites in 2018.

