

IRSF presents...



HER Knight Information Sheet

Hello Dads, Grandfathers, Brothers, Uncles and Friends!

- Have you been interested in becoming involved and doing something to help raise funds for research?
- Have you just not known exactly what you can do?
- Would you like to take part in October Rett Syndrome Awareness Month?
- Would you like the chance to receive an iPad2 for the Rett angel in your life?

If you answered YES to any of the questions above...here is your chance!

Please take part in this EASY fundraising and awareness campaign that will take place now through October. Take a look at information contained on this page as well the PDF with HER Knight Challenge details—you will find everything you need to get started.

Simply gather a picture or two (you may want to share pictures of just your Rett Angel or of you and your Rett Angel together), write your letter and send off to all of your family and friends! Firstgiving, IRSF's online giving tool, is fun and easy! You can watch your donations grow daily!

Get started today...don't delay!

Ideas of what to include in your letter/email:

- Brief background on your Rett Angel (name, age, date of diagnosis)
- Story that tells what an inspiration your Rett Angel is and just how special she is
- Explanation of what Rett syndrome is (*see below*)
- Explanation of research (*see below*)
- Request to make a donation
- Firstgiving link (*see below*)
- Be sure to tell people their donation is tax deductible and will be acknowledged by IRSF

What is Rett Syndrome?

Rett syndrome is a unique developmental disorder that is first recognized in infancy and seen almost always in girls, but can be rarely seen in boys. Rett syndrome has been most often misdiagnosed as autism, cerebral palsy, or non-specific developmental delay.

Rett syndrome is caused by mutations on the X chromosome on a gene called MECP2. There are more than 200 different mutations found on the MECP2 gene. Most of these mutations are found in eight different “hot spots.”

Rett syndrome strikes all racial and ethnic groups, and occurs worldwide in 1 of every 10,000 to 23,000 female births.

Rett syndrome is a developmental disorder. It is not a degenerative disorder.

Rett syndrome causes problems in brain function that are responsible for cognitive, sensory, emotional, motor and autonomic function. These can include learning, speech, sensory sensations, mood, movement, breathing, cardiac function, and even chewing, swallowing, and digestion.

Rett syndrome symptoms appear after an early period of apparently normal or near normal development until six to eighteen months of life, when there is a slowing down or stagnation of skills. A period of regression then follows when she loses communication skills and purposeful use of her hands. Soon, stereotyped hand movements such as handwashing, gait disturbances, and slowing of the normal rate of head growth become apparent. Other problems may include seizures and disorganized breathing patterns while she is awake. In the early years, there may be a period of isolation or withdrawal when she is irritable and cries inconsolably. Over time, motor problems may increase, but in general, irritability lessens and eye contact and communication improve.

Rett syndrome is confirmed with a simple blood test to identify the MECP2 mutation. However, since the MECP2 mutation is also seen in other disorders, the presence of the MECP2 mutation in itself is not enough for the diagnosis of Rett syndrome. Diagnosis requires either the presence of the mutation (a molecular diagnosis) or fulfillment of the diagnostic criteria (a clinical diagnosis, based on signs and symptoms that you can observe) or both.

Rett syndrome can present with a wide range of disability ranging from mild to severe. The course and severity of Rett syndrome is determined by the location, type and severity of her mutation and X-inactivation. Therefore, two girls of the same age with the same mutation can appear quite different.

Rett syndrome presents many challenges, but with love, therapy and assistance, those with the syndrome can benefit from school and community activities well into middle age and beyond. They experience a full range of emotions and show their engaging personalities as they take part in social, educational, and recreational activities at home and in the community.

Or just share the link directing donors to the Rett syndrome section on the IRSF website! <http://www.rettsyndrome.org/content/blogcategory/17/1105/>

What's Happening with Research?

Since the identification of the MECP2 gene in 1999, research on Rett syndrome has made important advances. Through research we have gained a greater understanding of the molecular mechanisms underlying Rett syndrome which result from MeCP2 dysfunction.

Scientific research has yielded some exciting developments:

- Numerous target genes have been identified that are regulated by the MeCP2 protein
- Genetic studies have begun to identify additional causative genes such as CDKL5 and FOXP1
- Valuable animal models have been developed that effectively reproduce the disease and demonstrate the potential reversibility of the disease
- Neurobiological studies have enabled us to determine some of the neurological underpinnings of Rett Syndrome pathogenesis

The International Rett Syndrome Foundation has played a critical role in driving this progress forward through grant funding. Since 1998 IRSF has awarded over \$24M in direct support of research grants towards innovative basic and preclinical research. These grants are primarily focused on understanding the mechanisms that contribute to Rett syndrome and its associated complications.

Or just share the link directing donors to the research section on the IRSF website!
http://www.rettsyndrome.org/index.php?option=com_content&task=blogsection&id=6&Itemid=944

You could also share this exciting wording about research!

- Gene discovered (we know what causes it!) - 1999
- Animal models created (we can study it!) - early 2000's
- Reversed in a mouse model (we can fix it in the lab!) - 2007
- iPS cell lines created (Rett in a petri test from real skin cells of girls with the disease - we can test real drugs on real cells - mice are not people!) - 2010
- IGF-1 drug trial begins in Boston, shows early promising results (first trial to try a FIX for symptoms, not just treat them!) - 2011
- Pace is dizzying, the research dollars invested are really paying off today, please support IRSF!

Directions for Setting up your Firstgiving Page

- Go to www.firstgiving.com/rettsyndrome/herknight
- Click on Fundraise

- Enter your email address
- Sign in or create a new profile
- Create your fundraising page
- Create your title
 - *Ideas include but are not limited to “Megan’s Knight in Shining Amour” or “Stacey’s Warrior” or “Molly’s Hero”...ideas are endless; be creative!*
- Create your URL
- Personalize your page by clicking on “edit your page”
- Share your individual link with others (please note—IRSF suggests emailing your link vs. sharing it directly from Firstgiving as the email may be routed to the recipient’s spam folder)

GOOD LUCK!!!