



**Canadian
Ectodermal Dysplasia Syndromes
Association**



Introducing Michael Boutilier

**Michael Boutilier will be assisting
CEDSA's executive director,
Meghan Howard**

Much of our work at CEDSA is made possible through the generosity of our Director Emeritus and Megram Consulting Services president Robert C. Cross. Bob has kindly provided the office space and services of his Megram team to help develop and grow CEDSA.

Megram and CEDSA team members Bonnie James (accounting), Irene Sauve (graphic design) and Devon Lesk (website design), along with CEDSA Executive Director Meghan Howard would like to welcome Michael Boutilier, our newest team member, aboard. Michael joins CEDSA as a senior writer, using his communications background to provide research and articles for our website and newsletter. Michael will be working approximately one day per week with CEDSA. He currently resides in Renfrew, Ontario, with his wife and son.

**Visit us at
www.ectodermaldysplasia.ca**



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- email us today!***

Hypomyelination and ectodermal dysplasia?

The mysterious world of genetics – mutations, dominant, variations, mapping, recessive, x-linked – all words that float over your head while the geneticist scribbles on paper upside down in front of you trying to clarify how it all connects. After waiting months, even years, to finally get an answer it can be heartbreaking to finally understand what the geneticist is really telling you: “We can’t give a clear diagnosis.”

For the Sprung family of Ontario, it has been years of testing and doctors appointments for their young son. At first, doctors suggested autism, then possibly a connection to Cleft Lip/Palate-Ectodermal Dysplasia Syndrome (CPLD) without the clefting.

Now geneticists have found their son carries a triplication of the PVRL1 gene (on Chromosome 11). That gene, apparently when mutated a specific way, has been linked to autosomal recessive cleft lip/palate ectodermal dysplasia. When that gene is shut off, it causes major defects and developmental delays. But for this little man, the gene isn’t shut off; instead he has it in triplicate. MRI tests have also revealed some evidence of hypomyelination.

While the parents are now awaiting their own genetic tests to try and unlock another piece of the puzzle, they are asking if any member of CEDSA or a member of an international ectodermal dysplasia association have heard of hypomyelination as part of an ectodermal dysplasia syndrome. Brief online research has not revealed any studies on this, and the geneticist involved in this case says he is “unclear of the significance of his clinical presentation.”

This little boy also has a lot of trouble digesting certain foods; as many of us know, some ectodermal dysplasia syndromes can be characterized by food intolerances and/or allergies.

If you have information you would like to share with the Sprung family, either about the PVRL1 gene, hypomyelination in connection with an ectodermal dysplasia syndrome or just some general advice to pass along, please contact executive director and president Meghan Howard, CEDSA at meghan@megram.com and she will make the connection.



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Arctic Heat donates to help a Canadian child enjoy the summer outdoors

Two days after approaching Arctic Heat USA about its cooling vest projects, the Canadian Ectodermal Dysplasia Syndromes Association (CEDSA) received the following wonderful news.

"I would be more than happy to supply you with an XS Arctic Heat Cooling Vest for a child with ectodermal dysplasia to demo," wrote Dean Sainsbury, Arctic Heat USA. "I have been a junior sports coach for the past 20 years, so I am always keen to help kids stay cool and be active."



In addition, Mr. Sainsbury is providing CEDSA members with a 10 per cent off discount code for online purchases of adult sizes over \$150 available in blue or white. The company does carry some white children's cooling vests which are adjustable. In addition, they can manufacture the cooling vest in custom-made sizes based on chest measurements with a zip front – they are made in two inch decreases in size, for example: XXS (29 inch), XXXS (27 inch), XXXXS (25 inch) etc. Custom orders are \$220 U.S. each and take three to four weeks for delivery.

The vests are lightweight and work using a two-stage cooling process. Each vest contains a gel-like substance that can be frozen or chilled and will stay cold for up to two hours. They retail for just over \$200 U.S.

You can special order an Arctic Heat Body Cooling Vest in a child's size — in blue or white

The Canadian Ectodermal Dysplasia Syndromes Association would like to thank Arctic Heat for its support.

"The value of this donation goes beyond dollars – it means feeling normal, playing outside, being with friends and staying active – experiences all children deserve to have. Too often children, and adults, with an ectodermal dysplasia syndrome are unable to do the sports they love because of their reduced ability to perspire," says Meghan Howard, CEDSA executive director and president. "Arctic Heat just gave a child the gift of summer."

Watch for the upcoming article on the Arctic Heat Cooling Vest after being tested by a young man with XLHED. The vest will then be entered into a draw for CEDSA members to win!



*Check out the
CEDSA
Sharing Forum*

One of the best things about our organization is the networking opportunities it offers for a cross-Canada community of people whose lives are impacted by ectodermal dysplasia syndromes. Members include individuals who have an EDS, their friends and family members, and also medical health professionals who treat patients with one of the 150-plus disorders.

Our mission at CEDSA is to enhance the quality of life of those impacted by ectodermal dysplasia syndromes, and what better way to do so than by sharing the voices of our members with our entire community?

That's why we're introducing the CEDSA Sharing Forum, an online message board where you can share your advice, stories, questions and more with the CEDSA family. Your comments can be submitted via email to mike@ectodermaldysplasia.ca, and they will be posted in a timely fashion.

We look forward to hearing from you! Check out our Sharing section for more information.



EDSA Canadian
CSDE Ectodermal Dysplasia Syndromes
Association

Introducing the Paisley Protectives Canadian-made cooling vest

Any form of cooling comfort is welcome in the summer — especially for people with an ectodermal dysplasia syndrome. The advent of cooling garments, such as vests, scarves, and hats, is a helpful reprieve for those who wish to join in fun summer activities.

As luck would have it, Paisley Protectives of Canada, an organization that markets surface protection solutions, has a cooling garment that is manufactured right here in Canada: an adjustable cooling vest with pockets to insert cooling packs. These cooling vests are made of specially-designed material with two chest pockets and two back pockets to hold cooling inserts that keep the body cool for up to five hours. This adjustable vest will accommodate a wide range of sizes.

The vests (\$142) and four-pack of cooling inserts (\$94) retail for \$236; however, Paisley has generously extended an offer to all CEDSA members to receive a 10 per cent discount on all cooling vest/cooling insert orders.

For more information, visit Paisley Protectives online (www.paisleypro.com) or contact Mary Lou Sayers at (marylou@paisley.ca) or telephone at 1-877-769-9845 ext. 223.



Techni-Ice inserts deliver up to five hours of cooling relief.

Adjustable four-pocket cooling vest is designed to keep your body cool during hot weather or physical activity

Branching out:

German student looks to Canada for experience in the timber industry

This spring, CEDSA received an email from the president of the German-Austrian-Swiss ED Support Group, Andrea Burk. Her son, Fabian, is looking to come to Canada and is reaching out to our members to see if anyone can assist him achieve his dream of working in B.C. to finalize his degree.

If anyone has any advice, contacts or is able to provide work experience in the timber industry for Fabian, contact CEDSA executive director and president Meghan Howard at meghan@megram.com who has Fabian's CV and email information.

My name is Fabian Burk and I'm studying Wood Technology in the 4th semester in Rosenheim, Germany. Next semester an internship is planned. From September 2011 on each student spends 18 weeks in a business of his choice. I'd like to spend 4.5 months in Vancouver, Canada.

Wood Technology is an engineering degree program with the same basic studies such as mechanical engineering, but it is aimed at the timber industry.

Primarily I'd be interested in a wood processing operation. However, I would also be suitable for an operation in the mechanical engineering sector.

After I completed my studies, I want to go in the direction of machine design and development. I'd be happy to learn more about other areas, such as quality assurance in a production, process optimization, or execution and supervision of a test setup.

Thank you!

Fabian Burk



Canadian Ectodermal Dysplasia Syndromes Association

Mom in Spain reaches out to Canadian families for information

Mónica and her son, two-year-old Miguel, live in Murcia, in the south-east of Spain. Recently, Miguel was diagnosed with Rothmund-Thomson Syndrome (RTS), an extremely rare ectodermal dysplasia syndrome. There are roughly 300 documented cases of RTS worldwide.

Due to the rarity of

RTS, it can be very challenging to find specific information about the basic care needs of children such as Miguel. Mónica is now reaching out to the international ectodermal dysplasia community for help. If you have RTS, are the parent of a child with RTS or you are a health care professional with knowledge that you can

share, we'd love to hear from you.

Mónica is fluently bilingual in English and Spanish, so if you are interested in connecting directly with her, please contact CEDSA by email to: meghan@megram.com or by telephone at 613-432-9491 and we will forward your name and email address to her. Alternately,

you can send us any information you have about RTS, such as your advice on living with the day-to-day challenges of this syndrome, and we will forward the information directly to Mónica and Miguel and post it on our website for others to reference.

Thank you very much for your help!

The Benefits of Membership

For only \$20 a year, you are helping build this dynamic young organization and are reaching out to other families across Canada. Benefits of membership include:

- Networking and outreach with other Canadians;
- Access to new medical and research information;
- Connections to the international ectodermal dysplasia community;
- Support and tips on living with the day-to-day challenges of having an EDS;
- Entry into draws for cooling vests;
- Information and discounts on products;
- Quarterly newsletters;
- Advocacy and funding support.

Join today through our easy on-line membership form or by sending your name, mailing address, and let us know if you are a friend or family member of someone with an EDS, have an ectodermal dysplasia syndrome or are a member of the medical community. Please send your \$20 payment to the Canadian Ectodermal Dysplasia Syndromes Association, 3-247 Barr Street, Renfrew, Ontario, K7V 1J6. A receipt and membership card will be mailed to you. Cheques and money orders can be made out to the Canadian Ectodermal Dysplasia Syndromes Association.

All current members and new members who join before August 30, 2011 will be entered into a draw to win a cooling vest, size XS.

Fundraisers!

The rain clouds hovered and the bugs turned out in droves – but neither deterred people from attending the first CEDSA fundraiser held on June 26, 2011 in Renfrew, Ontario. A huge garage sale was held at the home of board director Bob Cross with items donated by friends, family and co-workers from across the Ottawa Valley.

The kindness and generosity of those who donated items, and those who simply came to shop, was overwhelming. While many did not have any prior knowledge about ectodermal dysplasia syndromes, they asked questions and reached into their hearts to contribute. Over \$330 was raised with many items left over to be used for a second sale to be held later this summer.

The proceeds will go towards purchasing cooling vests for CEDSA members to help them cope with summer sun and enable them to participate in outdoor activities without fear of overheating.

We would like to recognize the following volunteers and donors for their support and assistance:

- Rhonda Runtz and Tim Shea
- Jody and Lawrence Smith
- Angie and John Greig
- Barb and Corey Crozier
- Michael Boutilier and Suzanne Lemke
- Bob and Leslie Cross

Upcoming: Plans are underway for a fall benefit dance to be held this September. We'd love to see you there – to dance, network and bid on the silent auction. Watch the website for upcoming details.