ACDA
ALVEOLAR CAPILLARY DYSPLASIA ASSOCIATION
acdassociation.org
WINTER 2022
Issue 78

ACDA NOTES

FROM THE ALVEOLAR CAPILLARY DYSPLASIA ASSOCIATION

What's INSIDE

2021:

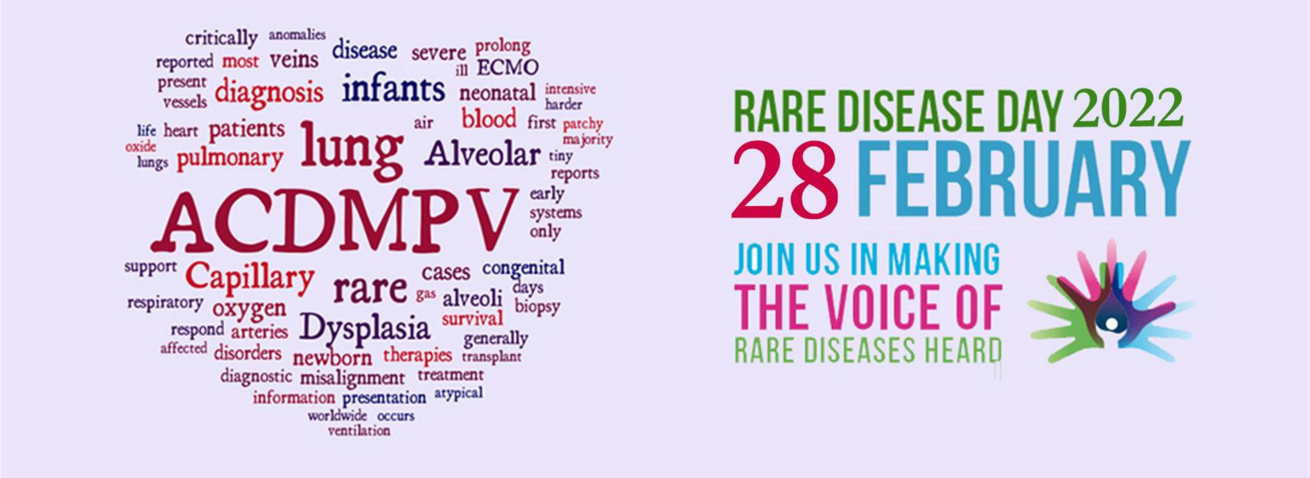
Year in Review

As we take a look back at 2021:

- **One global pandemic, continued.**
- **Sixteen new families registered with the ACDA. These families are from the USA, UK, Ireland, Austria, New Zealand, Poland, The Netherlands and Denmark.**
- **One research grant in the amount of $50,000.**
- **Thirteen rainbow babies born to ACDA registered families.**
- **Multiple attendees at a virtual Rare Disease Day event featuring ACDMPV hosted by the New York City Police Department.**
- **More than twenty journal articles published about ACDMPV.**
- **Numerous successful fundraisers for ACDMPV research and awareness events.**
- **One continued goal – To find the cause of and cure for ACDMPV.**

Regards, Eliza Rista, President

FEBRUARY 28, 2022 IS RARE DISEASE DAY



Rare Disease Day is an international advocacy day to bring widespread recognition of rare diseases as a global health challenge. The day is celebrated on the last day of February every year – February 28 in 2022. The ACDA has signed on once again to partner with NORD to support this awareness campaign and we encourage everyone to participate in some way.

Click to see how the ACDA supported Rare Disease Day in prior years (2021**,** 2020, 2019, 2018, 2017 and 2016)**!** Click HERE for ways to participate in the U.S. in 2022 and click HERE to visit the EURORDIS website for Rare Disease Day 2022 in Europe.

Below are a few suggested ideas:

- **Fill your home with color!** In an effort of global solidarity, you are invited to light or decorate your home with the Rare Disease Day colors at **7:00 p.m. your local time on February 28, 2022**. See the flyer below for additional details!
- Click **HERE** ($), **HERE** (£) or **HERE** (€) to print a flyer for anyone to use at their workplace that encourages employees to donate $5 / £5 / €5 to the ACDA and wear jeans to work on Monday, February 28 for Rare Disease Day. **#JeansForGenes**
- Organize a fundraiser (Facebook or direct give) to raise money for the next ACDMPV research grant.
- Contact your local newspaper to write an article about what Rare Disease Day means to you. Click HERE to read various articles previously written about our ACDA families.

(continued on following pages)

1:00 p.m. EST / 6:00 p.m. GMT (UK)

Coffee hour

February 27, 2022

You're Invited

Zoom details to be issued privately via email and parent group
(ACDA registered parents only)

- If you are a parent of a child affected by ACDMPV, please join us for a **Coffee Hour** on **Sunday, February 27, 2022** at **1:00 p.m. EST (6:00 GMT (UK))** to meet and interact with other ACDA registered parents. The Zoom details will be provided separately via email and in a closed group for parents.

LIGHT UP FOR RARE

THIS RARE DISEASE DAY FEBRUARY 28TH

The Rare Disease Day colors are blue, pink, green and purple.

Fill your home with color! In an effort of global solidarity, the ACDA invites you to light or decorate your home with the Rare Disease Day colors at 7:00 p.m. your local time on February 28, 2022. #LightUpForRare #ACDA

(continued on following page)

HOW YOU CAN GET INVOLVED WITH RARE DISEASE DAY

Rare Disease Day is the globally-coordinated movement on rare diseases, working towards equity in social opportunity, healthcare, and access to diagnosis and therapies for people living with a rare disease. Since its creation in 2008, Rare Disease Day has played a critical part in building an international rare disease community that is multi-disease, global, and diverse– but united in purpose.

Join the Global Chain of Lights this Rare Disease Day. Together we will light up our monuments, our public spaces, our buildings, homes, hospitals and more. These lights show our resilience and strength towards a brighter future. Collectively, we can improve the lives of the 300 million people worldwide living with a rare disease.

SCREEN THE VIDEO

Stream to your TV or laptop, fill your home with colour, **28 February, 7pm**. Share your story online with the hashtag **#LightUpforRare**

GET CREATIVE

You could create your own window display - with coloured lights, painted pictures, tissue paper collages or your drawings - however you like to get creative, share your colours!

WANT TO DO MORE?

Start a conversation with the people in your community to help spread awareness of people living with a rare disease. **Share your story**, or share the story of people who inspire you. The more voices that join us, the louder we can be in our demand for equity.

Check out all the **events and activities** happening around the world. Here you can find and join patient organisations, and **join your community**. You can even create your own event and post it to the website, to raise awareness or raise donations.

Download a whole host of **campaign materials** - from social media graphics, videos, posters, badges, banners and toolkits. We've got everything you need to help raise awareness, **your way**.

RARE DISEASE DAY®

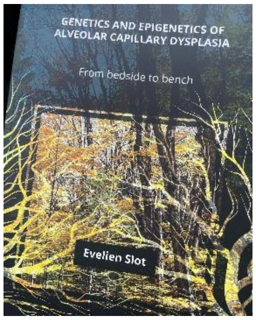
#LightUpforRare this #RareDiseaseDay

RESEARCH NEWS

2022 Research Grant:

The 2022 grant process is underway, with partnerships and announcements to follow in early spring. Stay tuned for further information!

PhD Thesis (Erasmus University Rotterdam):



Dr. E. Heuberger-Slot at **Erasmus MC** in Rotterdam, Netherlands recently had her PhD dissertation entitled, "*Genetics and Epigenetics of Alveolar Capillary Dysplasia: From bedside to bench*" published by Erasmus University Rotterdam, which can be found HERE.

Abstract: "This thesis includes studies into the clinical, histological and molecular aspects of the congenital lung disorder alveolar capillary dysplasia (ACD). This thesis provides an important basis for future research that is needed to increase our understanding of the pathogenesis of ACD and for improvement of disease management."

Journal Article (Hum Genomics):

The genetic research team at **Baylor College of Medicine** in Houston, Texas, USA recently collaborated with the clinical lab, **Baylor Genetics**, and an international team in Poland to publish a manuscript entitled, "*Detection of low-level parental somatic mosaicism for clinically relevant SNVs and indels identified in a large exome sequencing dataset*" in the Human Genomics journal, which can be found HERE. Partial Abstract:

"**Background:** Due to the limitations of the current routine diagnostic methods, low-level somatic mosaicism with variant allele fraction (VAF) < 10% is often undetected in clinical settings. To date, only a few studies have attempted to analyze tissue distribution of low-level parental mosaicism in a large clinical exome sequencing (ES) cohort.

Methods: Using a customized bioinformatics pipeline, we analyzed apparent de novo single-nucleotide variants or indels identified in the affected probands in ES trio data at Baylor Genetics clinical laboratories. Clinically relevant variants with VAFs between 30 and 70% in probands and lower than 10% in one parent were studied. DNA samples extracted from saliva, buccal cells, redrawn peripheral blood, urine, hair follicles, and nail, representing all three germ layers, were tested using PCR amplicon next-generation sequencing (amplicon NGS) and droplet digital PCR (ddPCR). […]

Conclusions: Our study illustrates the importance of analyzing ES data using sensitive computational and molecular methods for low-level parental somatic mosaicism for clinically relevant variants previously diagnosed in routine clinical diagnostics as apparent de novo.

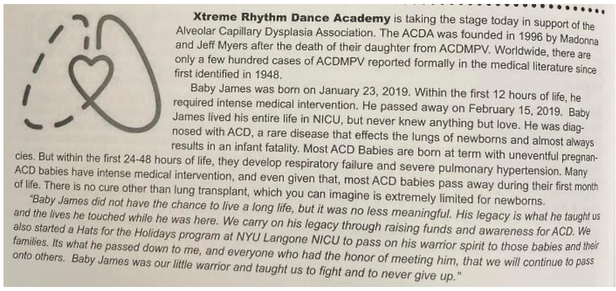
ACDA COMMITTEE POSITIONS

Please check our website for a full listing of Board and Committee members and let us know if you would like to get involved.

http://acdassociation.org/board-members

AWARENESS NEWS

Dance Competition (James Perrella):



Xtreme Rhythm Dance Academy is taking the stage today in support of the Alveolar Capillary Dysplasia Association. The ACDA was founded in 1996 by Madonna and Jeff Myers after the death of their daughter from ACDMPV. Worldwide, there are only a few hundred cases of ACDMPV reported formally in the medical literature since first identified in 1948.

Baby James was born on January 23, 2019. Within the first 12 hours of life, he required intense medical intervention. He passed away on February 15, 2019. Baby James lived his entire life in NICU, but never knew anything but love. He was diagnosed with ACD, a rare disease that effects the lungs of newborns and almost always results in an infant fatality. Most ACD Babies are born at term with uneventful pregnancies. But within the first 24-48 hours of life, they develop respiratory failure and severe pulmonary hypertension. Many ACD babies have intense medical intervention, and even given that, most ACD babies pass away during their first month of life. There is no cure other than lung transplant, which you can imagine is extremely limited for newborns.

"Baby James did not have the chance to live a long life, but it was no less meaningful. His legacy is what he taught us and the lives he touched while he was here. We carry on his legacy through raising funds and awareness for ACD. We also started a Hats for the Holidays program at NYU Langone NICU to pass on his warrior spirit to those babies and their families. Its what he passed down to me, and everyone who had the honor of meeting him, that we will continue to pass onto others. Baby James was our little warrior and taught us to fight and to never give up."

We are so proud of you, Frankie! Thank you for raising funds and ACDMPV awareness by selecting the ACDA in honor of your baby brother, James.

In February 2022, **Maria and Jimmy Perrella**, ACDA parents to James (January 23, 2019 – February 15, 2019), helped coordinate an awareness and fundraising event for **ACDMPV research in honor of Baby James Perrella**. In Maria's own words, "My daughter Frankie dances competitively. She participated in 'The Power of Dance' competition yesterday. **Each dance team was to choose a charity to dance for. Frankie's school chose the ACDA.** The competition will be making a donation to the ACDA. We were truly humbled by this. We are truly grateful that Frankie belongs to such a phenomenal dance academy. **Frankie was so proud to dance for her baby brother.**"

Baseball Helmet (Bella Blehr):

Way to go, Austin! Thank you for honoring your sister, Bella, while raising ACDMPV awareness.

Maryann Blehr, ACDA mother to Bella Blehr (December 21, 2009 – January 12, 2010), shared another heartwarming story about the impact of ACDMPV on siblings. One of her sons, born after Bella, **asked on his own accord if he could wear the ACDA logo on his team's baseball helmet**. What a beautiful story of love between siblings.

School Assemblies (David Ashwell):

Amelia Ashwell, ACDA mother to David Ashwell (March 4, 2011 – March 19, 2011), recently **spoke at two school assemblies in advance of Rare Disease Day 2022** on behalf of The David Ashwell Foundation (see further TDAF updates on page 13 of this newsletter.)

ACDA Tattoo (Cassie Carrigan):

In February 2022, Cassie Carrigan, ACDA mom to Caleb (August 9, 2019 - September 4, 2019) got a tattoo of the ACDA logo. In Cassie's own words, "It was a powerful, heart-warming, sad and honoring experience. While I was getting it, I thought about each of you and your precious babies. This is for Caleb and all of our babies! I can't wait for people to ask me about it! I will share our stories and our babies."

The ACDA is now aware of a total of **fourteen tattoos featuring the ACDA logo!**

LUNG TRANSPLANT UPDATES

Lung Transplant Update (Boston Children's Hospital):

In February 2022, an ACDA registered family shared the good news that their (now seven-month old) son **received a bilateral lung transplantation in January 2022 after a continous six months admittance at Boston Children's Hospital in Boston, Massachusetts, USA**. We look forward to sharing more details about this ACDMPV fighter once the challenges of being immediately post-transplant have settled. In the meantime, please keep this family in your thoughts.

Five Year Anniversary (Luca Palmisano):

We introduced you to Luca Palmisano in Issues #57-59, #61, #66-67, #69 and #75 of ACDA Notes. In November 2016, Luca received a lung transplant at 4.5 months old at Children's Hospital of Philadelphia in the USA.

On November 8, 2021, Luca celebrated five years post bilateral lung transplantation. In his parent's own words, "Forever indebted to the family who made the ultimate sacrifice for our dying son. We remember you today, and with every breath Luca takes. You gave him (us) the greatest gift mankind can ever give, the gift of life. For all those involved in this journey, on behalf of #TeamLuca, thank you. Thank you for the care, the prayers and the continued support. It's amazing what can happen when you keep the faith. 5 years, Wow. They said you wouldn't make it, Bubba…And LOOK AT YOU NOW!!! **Happy five-year Transplant Anniversary.**"

You can follow Luca's Lung Transplant Story HERE. #teamluca #GiftofLife #acdwarriors

FUNDRAISING NEWS

Donations:

To make a secure tax deductible donation to the ACDA or the NORD Research Fund (ACD), please visit our website for full instructions.

acdassociation.org/donate

The ACDA is a 501(c)(3) non-profit, tax-exempt organization as designated by the Internal Revenue Code of the United States.

Balance of ACDA account:

The balance of the ACDA bank accounts as of February 26, 2022 is [redacted] (which includes the remaining funding allocated to the 2021 grant recipient in the secondary payment amount of $25,000, together with $50,000 originally deferred due to the COVID-19 emergency to be reapplied to 2022 grant issuance).

Update from AmazonSmile:

The ACDA was issued a [redacted] donation from the AmazonSmile Foundation as a result of AmazonSmile program activity between July 1 and September 30, 2021. To designate the ACDA as your charity, please follow the link below so that all of your eligible shopping will benefit the ACDA:

http://smile.amazon.com/ch/46-2915711

Update from Spreadshirt:

Items with the ACDA logo are available for purchase in our Spreadshirt store HERE. The accrued commission payment from Spreadshirt between October 14, 2021 and February 24, 2022 is [redacted]. Please continue to shop at our store as new items and new features are added regularly. You have the option to customize your products by choosing "Create," including adding your child's name or picture to most items featuring the ACDA logo. Don't forget the ACDA earns a commission equal to **20% of every product sold**! **Look for 15% off from March 4-9, 2022!**

Donations Received:

Thank you to the following families and friends that have made donations to the ACDA since the last ACDA Notes:

Donor	In Memory Of
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**We are sorry we do not know the child for whom the memorial contribution was made. Please contact us to let us know.*

#GivingTuesday Update:

WOW. The ACDA's fundraising goal for #GivingTuesday on November 30, 2021 was $1,000 and, with your help, we doubled our goal and were able to bring in **$2,105 (with a total of 12 different honorees, see the list below)**!!

One day, one mission, one BIG thank you. We give our thanks to those who donated to the ACDA on #GivingTuesday. The money raised will be contributed directly to ACDMPV research and supports the efforts of the medical community to find the cause of and cure for ACDMPV.

Our gratitude extends not only to our amazing donors but to each and every person who shared a post or email to spread awareness, asked their family and friends to consider a donation or helped in any possible way to make our #GivingTuesday campaign a huge success in 2021.

Thank you for the generous #GivingTuesday donations in honor of *Xavier K., Amelia, Caleb, Callie, Dahlia, Dean, Madison, Chelsea, Johnny R., Finley, Landen and Brian.*

October 15 Fundraiser (James Perrella):

[redacted]

ACDA fundraiser in memory of Baby James

$3,634 raised of $5,000

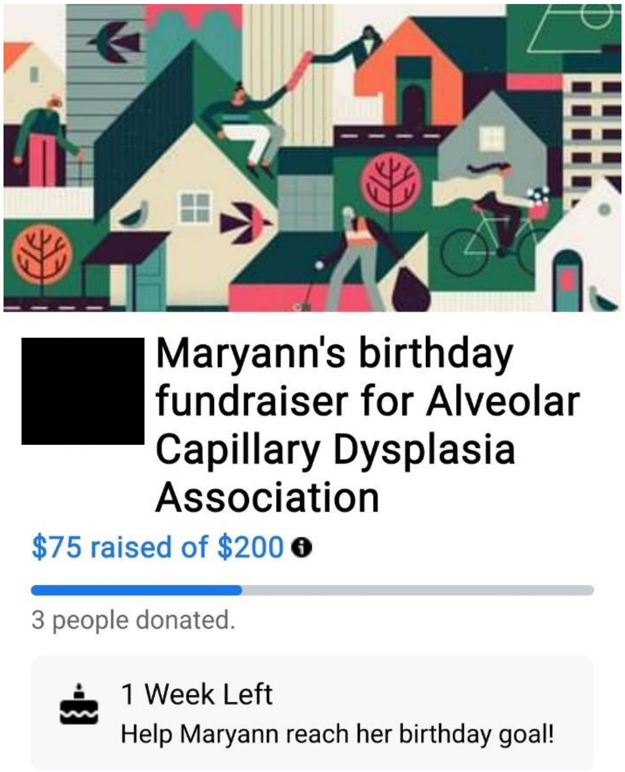
Fundraiser ended

Maria and Jimmy raised $3,634 through a Facebook fundraiser in honor of baby James Perrella, with 60 individual donors! Amazing!!

In October 2022, **Maria and Jimmy Perrella**, ACDA parents to James (January 23, 2019 – February 15, 2019), organized a fundraiser for **ACDMPV research in honor of Baby James Perrella**. In Maria’s own words, “October 15 is Pregnancy and Infant Loss Awareness day. In honor of our son, Baby James, we are again fundraising on behalf of the ACDA. Our son was born in January, 2019 and passed away February 2019 from a rare disease called Alveolar Capillary Dysplasia. **This is our third year holding this fundraiser and with your support we have previously raised almost $18,000.** We would like to sincerely thank everyone for their love and support in the past with Baby James, and we couldn’t be more blessed having friends and family like all of you. We hope that our combined donations can help the Association work towards discovering more information about ACD, and assist in the development of treatment methods in the future.

Thank you all for helping to honor and remember Baby James, and for continuing to play such a profound role in the funding of research into ACD, and working towards a future where effective treatment is a reality. A life need not be long to be meaningful _unknown.”

Birthday Fundraiser (Bella Blehr):



[redacted]

Maryann's birthday fundraiser for Alveolar Capillary Dysplasia Association

$75 raised of $200

3 people donated.

1 Week Left

Help Maryann reach her birthday goal!

Maryann is on her way to her goal (donate here!) through a Facebook birthday fundraiser in honor of her daughter, Bella Blehr. Thank you, Maryann!

In February 2022, **Maryann Blehr**, ACDA mother to Bella (December 21, 2009 – January 12, 2010, organized a birthday fundraiser for **ACDMPV research in honor of Bella Blehr.** In Maryann’s own words, “For my birthday this year, I'm asking for donations to Alveolar Capillary Dysplasia Association. I've chosen this nonprofit because their mission means a lot to me, and I hope you'll consider contributing as a way to celebrate with me. Every little bit will help me reach my goal.”

Here’s how to set up a birthday fundraiser on Facebook!

REPORT FROM STICHTING ACD (NETHERLANDS):

Mathijs Lourens, ACDA father to Myla (November 20, 2015 - December 17, 2015), continues his amazing work in support of his Dutch ACDMPV foundation, "**Stichting ACD**." Mathijs plans to continue running marathons for ACDMPV awareness in Europe and then worldwide! Stichting ACD previously completed the Rotterdam Marathon (April 2017, 2018 and 2019, October 2021), Amsterdam Marathon (October 2016 (*half*) and 2017), Amsterdam Kids Run (October 2018, 2019 and 2021), Berlin Marathon (September 2019), Stichting Marathon #1 (October 2020), Stichting Marathon #2 (April 2021) and various other running events. **Please contact Mathijs if you are interested in participating or cheering on his upcoming plans to raise ACDMPV awareness in the Netherlands or beyond!** For additional information about the upcoming runs highlighted below, please visit Run4ACD on Stichting ACD's website.

NN Marathon Rotterdam (April 12, 2022):

Upcoming Event: Mathijs and Stichting ACD will run the **Rotterdam marathon on April 10, 2022**. The ACDA sends our very best to Mathijs and the team of Stichting ACD runners in Rotterdam. We look forward to providing pictures and an update in the next newsletter!

TCS Amsterdam Marathon Kids Run (October 17, 2021):

Mathijs and **kids** from team Stichting ACD ran the **TCS Amsterdam Marathon Kids Run on October 17, 2021** to raise ACDMPV awareness! The Kids Run is a children's run that takes place during the TCS Amsterdam Marathon. Children aged 4 to 12 can participate in the 700-metre run, which starts behind the stadium and finishes in the stadium itself. A particularly proud moment for Mathijs was watching **his young son, Alec, and daughter, Liva, participate in the event in honor of their sister, Myla.**

NN Marathon Rotterdam (October 24, 2021):

Team Stichting ACD ran the Rotterdam marathon on October 24, 2021 for the fourth time in a row (canceled in 2020 due to COVID-19). In Mathijs' own words, "So very proud and grateful for these champs who do this! For us and all other ACD families around the world."

International Children's Rights Day:

On November 20, 2021, in addition to being Myla Lourens birthday, it was International Children’s Rights Day, as established in 1989. In Mathijs’ own words, “Yesterday, the SP Landsmeer therefore distributed ribbons to children who are nominated, because they have selflessly done something for someone else. And we signed up our Team Myla kids from Landsmeer for this. We are so proud of them. They didn't have to think about running with us to support our cause. They are great! And now champs with ribbons.”


SP Landsmeer reikt 23 kinderlintjes uit

PhD dissertation (Erasmus MC):

On December 1, 2021, Dr. E. Heuberger-Slot defended her PhD dissertation, entitled: 'Genetics and Epigenetics of Alveolar Capillary Dysplasia' at Erasmus MC in Rotterdam, Netherlands. In Mathijs’ own words, “Today I had the honor to witness the defense of researcher Evelien Slot's thesis! She has spent four years researching ACD with a team which has produced great results, but also answers and new questions. Myla has also officially passed the contents of the thesis and therefore another special milestone today. This is how the commission cited Myla's booklet, which helped ACD get a face in addition to all the laboratory data. This has motivated several people to keep going. Today has also directly led to new motivation to meet new team members soon and tell them about Myla. The road may still be long, but step by step we get closer!”

Angel Gallery:

You are invited to view the “[Angel Gallery]” on Stichting ACD’s website for a touching image display featuring many of our ACDA babies. If you would like your child to be included in the gallery, please contact Mathijs at contact@stichtingacd.nl and send him a picture with full name, birth date and angel date.

REPORT FROM THE DAVID ASHWELL FOUNDATION (UK):

Important announcement, The David Ashwell Foundation is excited to introduce our NEW system for fundraising!! Please visit our new link below:

https://wonderful.org/charities/1121783

Thank you to those families who have set up direct debits for the charity and whose families fundraise at work for us. Looking back since March 2011 over **£200,000** has been raised for ACDMPV Research by families in the UK and Europe. This has contributed to the funding raised internationally to the ACDA. We are always very enthusiastic to raise more money for ACDMPV research and would appreciate any help!

Please use **The David Ashwell Foundation "Wonderful" donation system!** We are here to help UK families fundraise for ACDMPV research so that we can help to find answers to the condition that has become such a major part of our lives.

At the moment not many people are using The Giving Machine. Please, UK families can you use The Giving Machine (GO HERE) to generate income from online shopping. Everyone in the UK can do this – it is **so easy to register and is a way of earning easy and free money**. Please also encourage your friends and family too! Watch HERE how it works! I have the giving machine app on my phone and it makes giving as I shop so easy. It's very quick and easy to sign up!

If you live in the UK (and elsewhere), there are a number of options available for funding ACDMPV research through The David Ashwell Foundation.

1. UK families you can set up direct debits to The David Ashwell Bank Account (we have a number of people who do this – just ask me for bank details.)
2. You can make a donation directly, using the Wonderful website.
3. Through the Wonderful account you can set up a page in memory of your child or for special occasions (e.g. birthdays or Christmas).
4. Fundraise while you online shop (The Giving Machine) (a percentage of what you spend is donated)
5. Collect postage stamps

All UK & Europe based families are welcome to use The David Ashwell Foundation as a means of fundraising for ACDMPV Research. Please raise awareness of our small charity via social media including Twitter and Facebook. We are more than happy to hear from other families who would like to use the charity to raise money for ACDMPV Research. Simon and Amelia run The David Ashwell Foundation on a voluntary basis. **All** money raised will be transferred to fund ACDMPV research. Thank you to all who have used The David Ashwell Foundation to fundraise in memory of their precious child. For additional information, contact Simon and Amelia.

Website: https://wonderful.org/charities/1121783
Email: davidashwellfoundation@yahoo.co.uk
Twitter: @TDavidAshwellF
Mobile (Amelia): 07855473686

REMEMBERING OUR BABIES

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[redacted]

[redacted]

[redacted]

Welcome to New Families

A sad but warm welcome to the following newly registered families:

- [redacted]

[redacted]

[redacted]

[redacted]

Safe Arrivals!

Congratulations on the birth of the following little siblings in our ACDA registered families:

- [redacted]

[redacted]

[redacted]

[redacted]

[redacted]

CONNECT WITH US

Facebook:

- Official ACDA Public Page
- Parent Group (private)
- Family Group (private)

Read about the private groups with information on how to join:

http://acdassociation.org/support-groups/

Twitter:

- Follow us @acdassociation

Website:

- acdassociation.org

Email:

President@acdassociation.org (Eliza Rista)
Secretary@acdassociation.org (Renee Murray)
Treasurer@acdassociation.org (John Rista)

A note from the President: We absolutely want to hear from you as to how we can best meet your needs with respect to information about ACDMPV and also grief support. We are here to help in any way we can. Please know we always want to hear your ideas and we love community involvement on any level. Please never hesitate to contact me at President@acdassociation.org.

Regards, Eliza Rista, mom to Johnny
(February 20, 2013 – March 4, 2013)