From the ‘Founder’ to all of you

• The World is full of Wonderful, Good, and Charitable people.
• You all are here because you are among them!
• Thank you for all you have done and now do for SADS

Founding the SADS Foundation

• Background and Beginnings
• Salt Lake City to the World

Background: Key concepts.

• Long QT syndrome (LQTS) came from Denmark to Salt Lake City in the mid 1850s
• Intermountain Healthcare Inc., a multi-hospital and clinics entity and the University of Utah, through shared faculty and facilities, collaborated in LQTS research and clinical care
• Together we set the stage for the SADS Foundation
The beginning of LQTS research in Utah

- 1972: I joined the U of U faculty and the University CVRTI.
- 1973: I identified a Utah family of Danish origin to have LQTS, following the SCD of a member.
- Two brothers and families came to Utah in 1856 as members of the Church of Jesus Christ of Latter Day Saints (The Mormons) and they brought family genealogy records (a gold mine for inherited disease researchers).

LQTS research at the U of U and LDS Hospital

- 1975: I moved my office 2 miles from the University to the IHC LDS Hospital to become the Chair of Medicine, and at the same time the Vice-Chair of Medicine at the University of Utah School of Medicine. “The faculty collaboration” began.

The beginning of LQTS research in Utah

- Between 1856 and 1972, their posterity grew large.
- Many remained in Utah, so a large number of members, including affected members, created a “founders (the two brothers) effect” (many in the area had the disease).
- With their genealogy we could find and study them easily and that dramatically helped our study of the family and of inherited LQTS.

Laying the background for SADS

- 1975-1988: With the wonderful work of Katherine Timothy and Jolene Fox and several other great team members we developed a busy clinical care and aggressive clinical research program, with goals to:
  - Expand our family pedigrees as much as possible
Laying the background for SADS

- Prospectively contact as many members of each family as possible to get resting and exercise ECGs and a history of symptoms, to see who was affected.
- With this program we saw many family members and identified many affected members of these families.

An average example of the pedigrees we developed

- An example of the large pedigrees we developed.

There are 21 gene carriers: 80% were asymptomatic.

Laying the background for SADS

- Discovering who had the disease before they had any symptoms allowed us to treat them with prophylactic medications.
- This pre-symptomatic treatment was very effective, and is the key to preventing sudden death! Early recognition and early treatment!

Additional help arrived in 1990

- Li Zhang MD joined the team and Katherine and she plus the others really advanced the clinical care and research of the LQTS and other inherited arrhythmias programs.
The program was very successful

- I had weekly to twice weekly LQTS clinics during this time, often seeing a number of family members at the same time.
- By the mid 80's we had an 11 generation pedigree on the Danish family with around 1100 members identified!
- We had cared for and developed large pedigrees on many other LQTS families as well.

Serendipity or?

- Mid 1980's: Ray White PhD and Jean-Marc Lalouel PhD join the University faculty in Human Genetics.
- Jean-Marc had just developed the "linkage analysis" program for molecular genetic studies in families.
- 1988: I asked Ray, Jean-Marc, and Mark Leppert to collaborate and apply the linkage tool to the large Danish family pedigree and others; they agreed with enthusiasm.

More Serendipity, or?

- 1989: Mark Keating, MD, Ph.D., newly minted molecular cardiologist researcher, joined the University faculty and chose to pursue the linkage studies in the Danish and other families as his primary project.
- He recruited several outstanding molecular genetics post-docs who made great contributions to the projects.

First FRUITS OF OUR LABORS

- 1991: Keating et al report the first genetic linkage results in LQTS, using my Danish and two other families. Affected members linked to the Harvey ras-1 locus on chromosome 11 (later named KCNQ1): the LQT1 form.
Second FRUITS OF OUR LABORS

1992: Vincent, et al report the first genotype-phenotype studies in LQTS (comparison of the genetic findings to the clinical findings- symptoms, ECG), based on the H-ras-1 linkage study.


Key New Findings

1. LQTS has significant reduced penetrance (diminished expression) of the prolonged QT interval and symptoms phenotypes. About 25% of genetically affected members had normal-borderline QT intervals, and 50+ % had not had symptoms.

2. There is considerable variability of QT duration and frequency of symptoms, even within single families, indicating that genetic factors other than the specific mutation influence the expression of the QT interval and symptoms in LQTS patients.

3. The rate of SCD and other symptoms is much lower than previously thought, because of all the asymptomatic members, often lifelong, who were not previously recognized and thus, not included in earlier studies.
**Founding the SADS Foundation**

- These and other subsequent publications and our many talks given about these findings increased the public interest in the “rare disease” Long QT syndrome, but physicians were generally unaware of it and, therefore, did not look for it.

- Up to that time our program had seen many tragic, unexpected sudden deaths or aborted sudden deaths in children and young persons as the presenting findings in a family, a terrible experience for a family, which we hoped could be avoided by education of the public and physicians.

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**Some of the emotions after such a tragedy**

- Grief (inconsolable)
- Horror
- Fear
- Who’s next?
- Despair
- Depression

William Blake, c. 1800. “Plagues of Egypt”

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**Founding the SADS Foundation**

- So, with increased interest in the lay and scientific press, we thought there “had to be a better way to inform medical personnel and families about the disease and it’s warning signs and symptoms” in order to identify affected families before a tragic event occurred. A Charitable Foundation was the answer.
The Founding

- In 1992 I contacted an attorney friend, David E Salisbury, to draw up the papers for a 501(c)(3) charitable organization.
- He was Vice President, Katherine Timothy was Secretary, and I was the President, Chair of the Board, and Chief Medical Officer. We were the founding members.

The Beginning: The Office Personnel got the Foundation up and running, and growing

Charlene Holmstrom, Exec. Director
Lynn Godfrey, Asst. Director,
Nancy Duitch and Doris Goldman,
Katie Roberts, Kathy Vincent,
and several others were terrific in developing and directing the fledgling foundation

Lesson learned: Power of the Media

- A Reader’s Digest Article by Peter Michelmore. “Michael Vincent’s Good Cause”. June 1996. 27 million copies sold
- And many subsequent media articles on TV, and radio, in newspapers, magazines, etc.

Into the World

- SADS UK, The Canadian SADS Foundation
- SADS Israel, Italy, Argentina, China
- Working with the SADS International Scientific Advisors and their outstanding research and care programs
- Patients found and treated, lives saved, all over the world by the efforts of SADS, other similar wonderful organizations, and the many physicians and volunteers involved in these efforts
To the Current SADS Officers, Staff, Board of Trustees, and the Scientific Advisors

Thank you so much for the great work you are doing and have done, in your own programs and with SADS. I am so very grateful to you for your role in bringing my hopes and dreams for the SADS Foundation to such fruition!

Founding the SADS Foundation

• That in a nutshell is the background and beginnings of the SADS Foundation.
• “Appreciation is a wonderful thing: It makes what is excellent in others belong to us as well.” Voltaire French author, humanist, rationalist, & satirist (1694 - 1778)
• In the next few slides I'll show pictures of some of the “excellent others”. My apologies to those for whom I don’t have pictures or time.

Thanks to a few of the Key players in this story


Some researchers, and Lynn from SADS

The Founders

The two SADS Executive Directors, R to L, Charlene and Alice, the beginning and the present, and from their works, the future.

Alice Lara me Charlene Holmstrom
And to Laura, Sarah, Christine, Anne, Adrienne, Amy and Brandon
To Mike Ackerman MD, PhD, and to Susan Etheridge MD, for taking over the President and Vice-President of the Board roles, to keep SADS medical research and education efforts going.

My Apologies, and thanks to all others – my presentation time is gone

But, I need to thank my wife for her support, patience and advice. Thanks too to our daughter Kathy who worked with Charlene and the others in the early SADS years.

My wife D., and our daughter Kathy Salisbury

Questions, comments?