

INTRODUCTION OF FOUNDERS

2015 CHARGE SYNDROME FOUNDATION CONFERENCE

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by Steve Sorkin

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I am truly honored to be asked to introduce to you the four founders of the CHARGE Syndrome Foundation. And what a joy it is to have them together with us today! Those of us who were here back in the '90s, and have seen the Foundation itself, and more generally, awareness of and knowledge about CHARGE Syndrome, grow exponentially, owe so much to the work and foresight of these four pioneers.

There are few occasions in history when fate has placed together in one place and one time a select group of people who all have vision, knowledge, courage, and determination, and who go on to actually accomplish wonderful things that have a positive impact and better the lives of everyone. As was the case with our Founders, Washington, Jefferson, Franklin and Adams. Or maybe the Justice League of America with Superman, Batman, Green Lantern, and Wonder Woman.

We are fortunate that Sandy Davenport, Meg Hefner, Jim Thelin, and Marion Norbury all happened to be in Columbia, Missouri in the early and mid '80s. And we are all here today because of their work.

When you think about it, it is amazing how far we have come in just a little over 30 years. It is hard to imagine now, but back in those days only a handful of people, even medical professionals, knew anything about CHARGE or were even making the connections between all of the symptoms that we know today make up the syndrome. There were a few, among them Drs. Bryan Hall and Bonnie Pagon, who played the leading roles in discovering and naming CHARGE. But our four friends here today paved the way not only for more identification and research, but just as importantly for most of us in this room, for providing information and other resources to the individuals, parents, and families affected, and literally founded this Foundation. This fantastic foursome worked for ten years before the first CHARGE conference even happened.

Dr. Sandy Davenport specializes in sensory deficits, especially genetics and blindness, and the impact on neurological development. She had worked with Dr. Pagon in Seattle, and took notice of the similarities she was seeing in people with deaf-blindness who came into the clinic in Columbia. Multiple issues with sensory

deficits, that were more than just the sum of the individual disabilities, affected them all. That may seem obvious to us now, but it was Sandy that made the connections. Through her strong conviction that it is all about the families, Sandy led the way for lifting CHARGE Syndrome out of the pages of medical journals and in to our doctor's offices, schools, and homes.

Sandy was a mentor to **Meg Hefner**. Meg has been a genetic counselor for 35 years, and has worked on CHARGE for 32 of them. She was working in Columbia after getting degrees in Toronto and Ann Arbor. She and Sandy were beginning to identify a number of people with CHARGE and their families. The families were wonderful and interested and involved and eager to learn and help, and Meg quickly realized that there was absolutely nothing to give them. No pamphlet, no booklet, no printed information, period. Working with Sandy and Jim, Meg took the lead on writing the first booklet on CHARGE. She helped organize the Foundation, organized the first CHARGE conference, in 1993 in St. Louis, and has continued to serve as a valued advisor ever since.

Whereas Sandy worked on blindness and Meg on genetics, **Dr. Jim Thelin** brought expertise on deafness to the group. He was a pioneer in a more holistic view of deafness that we all take for granted now. He has since expanded his research to include the development of communication and the links between language development and mobility in people with CHARGE. His commitment to CHARGE has been demonstrated through the mountain of research he has directed with specialists in other fields and his own graduate students. He was an editor of the book on CHARGE Syndrome as well. With his reassuring manner and ability to give both frank advice and gentle encouragement, he has been a model for many CHARGE professionals.

As if that is not enough, Jim also is responsible for perhaps the most valuable achievement of all. His work with children who are deaf brought him in to contact with a service group called Quota International, a major part of whose mission was and is to provide assistance to families that have been affected by deafness. And working for the Quota group in Columbia was **Marion Norbury**.

Marion became, quite simply, the backbone of the Foundation. Remember we are talking before computer databases, desktop publishing, voice mail, and cell phones. It was the dawn of personal computers and, of course, before the internet. As the sole staff person, Marion did all of the typing, the mailing, coordinated the printing, maintained the ever growing lists, and answered the phone—her home phone number was the CHARGE Syndrome Foundation number—all from her home, which became the international headquarters for all things CHARGE Syndrome. When the other three moved to other cities, she coordinated communications between

Tennessee, Minnesota, and Missouri. As the Foundation grew after incorporating in 1993, she was a board member, treasurer, secretary, executive director, and director of family services. Many, many families in this room benefited from Marion's warmth, concern, outreach, and personal touch that always came shining through over the phone, in person, and even in the mail.

Meg always says that these kinds of groups are built on a triad foundation: medical, educational, and families. The CHARGE Syndrome Foundation was a pioneer in its commitment to put families first, whether it be providing child care at conferences, encouraging the professionals to spend time with and learn from the families, or seeking family input in to research topics and design.

Over the past twenty-plus years, our board members, volunteers, and other staff have been instrumental in helping the Foundation reach this point. And for sure there have been many other medical and educational professionals who have done invaluable work from which we have greatly benefited, many of whom are in this room [*Kim Blake, Jeremy Kirk, Rod Last, David Brown, Conny van Ravenswaaij, and CHARGE parents Tim and Nancy Hartshorne*]. But the framework for making it all possible traces back to these four visionary, caring, committed people who set us on our path.