

In Memoriam **John H. Edwards, MD**

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It was with a deep sense of sadness that I heard the name of John H. Edwards read at the annual business meeting of the American Society of Human Genetics in San Diego last October. The reading of names of members who died in the previous year is always a touching event, but this past year the announcement was particularly poignant for me. I first met John in Baltimore at the ASHG meeting in 1989, when I had received direction from the Board of Directors of the Support Organization for Trisomy 18, 13, and Related Disorders (SOFT) to invite the esteemed Oxford professor to be “our guest” at the annual conference in St. Louis for July 1990. John graciously accepted the invitation, and during a particularly humid and hot summer week along the Mississippi River, joined the 75 or so families of children with trisomy 18 (and related chromosome syndromes). SOFT is unique in that the members include families who have lost their children from these serious chromosome disorders and families who are coping with the challenges of rearing a child with chronic disease and disability. John and his wife, Felicity, also a physician, participated in every event of the Conference, including the Ryan Cantrell Balloon Release. In addition to the photo included here (Fig. 1) of John, Felicity, and his newly made friend, Kyle, I have a wonderful snapshot of John holding a balloon for release that honored the first-known patient with trisomy 18, described in his seminal paper [Edwards et al., 1960].

Professor John H. Edwards (1928–2007) made many significant contributions to the fields of Human and Medical Genetics during his illustrious career [McKusick, 2007], but the one that bears his name comes from the observation and documentation of the first patient with trisomy 18, or “Edwards syndrome.” John evaluated this 9-week-old infant at Birmingham Children’s Hospital in the summer of 1959. The girl showed all of the features that have come to be known as the Edwards syndrome due to trisomy 18. John was able to obtain postmortem tissue from the infant after she had died at 5 months of age, and in collaboration with Dr. Harnden, who was the second author on the paper and worked at

the research unit in Harwell, made the discovery of 47 chromosomes with an extra E group. John and his coauthors titled the paper “A New Trisomic Syndrome” because they appropriately recognized that they could not distinguish for certain whether the chromosome was the number 17 or 18. Later, newer technology and consensus on nomenclature allowed distinction between them, and we have come to refer to this as trisomy 18 syndrome.

On inviting Dr. Edwards to speak at the SOFT Conference, I asked him to relate the story of his first meeting and evaluation of this young girl at Birmingham Children’s Hospital. When he began his keynote address at the Conference, neither I nor the audience anticipated the unique account that we would receive. John’s first five minutes of the talk summarized the history of the Children’s Hospital and the adjoining region. Most of us—I suspect Felicity included—were not sure where the opening of his talk was going, but he proceeded to detail the narrative of his first examination and his astute hypothesis that she had a new aneuploidy syndrome.

During the Conference, I had the honor and privilege of joining Professor Edwards for an in-hotel clinic for many of the families of older children with trisomy 18 who were at the St. Louis meeting. (Since that time, SOFT has hosted a clinic for the families who attend the annual conference, usually at a nearby children’s hospital in the city of the conference. This past summer the 2008 SOFT clinic was held at Denver Children’s.) I have very fond memories of introducing Professor Edwards to many older children with trisomy 18 and trisomy 13 during the course of our clinic. One of the interesting side notes of John at that conference was his continual

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FIG. 1. Dr. Edwards and wife, Felicity, with a new friend at the SOFT picnic, St. Louis, 1990.

scholarship: He took advantage of being in Missouri and was reading as much of Mark Twain as he could.

In fact, he quoted Twain's "Life on the Mississippi" during the course of his talk.

Professor Edwards had several academic appointments during his years, and spent the last three decades of his productive career at Oxford University. While his many contributions to Human Genetics, including the famous Oxford Grid and his avid participation in the Human Genome Mapping workshops, are part of his scientific legacy, John's mentorship to me and his kindness to the families that had the chance to meet him in St. Louis live on in the history of SOFT. His connection to the families of children with the syndrome that bears his name is now part of his legacy as well. Professor John H. Edwards is survived by Felicity, his wife of 54 years, and their four talented offspring.

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