

Trisomy 13 and 18 and Quality of Life: Treading “Softly”

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Not too long ago I was privileged to speak at an annual international conference of the Support Organization For Trisomy 18, 13 and Related Disorders (SOFT). I learned much more than I taught. There were over 200 families there. Approximately half of the families were bereaved parents of a child with Trisomy 13 or 18. But then there was the other half, the half with surviving children with a trisomy. The oldest child I met was a young lady with trisomy 18 who was celebrating her 30th birthday. She was beautiful. Well dressed and impeccably groomed. She was non-ambulatory, non-verbal, and had a gastrostomy. But she surely knew her parents, she smiled at them and touched them with affection. There were many others with a wide distribution of ages. I met 16-year-old Karah and her siblings 11-year-old Olivia and 9-year-old Spencer. Karah’s smile as she hugs her siblings is contagious (Fig. 1). Olivia had written a wonderful tribute to Karah in which she wished that others could see Karah’s heart in the same way she did. She loved her deeply and wanted others to do the same. I met a family with an adorable 3-year-old with trisomy 13. She was walking and squealing and smiling and relating to her parents. Must be a mosaic I thought but then her physician mom told me that chromosomes were done at Boston Children’s and she was a full trisomy. I saw a room full of families with children with trisomies and yes, they had profound disabilities. But the room was filled with smiles and laughter and all of the sounds of people enjoying one another. A room full of long-term survivors. After nearly 40 years as a neonatologist and now

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several years as a pediatric palliative care physician, I considered long-term trisomy survivors a statistical rarity. Forget the statistics and the rarity. This was a room full of families having fun with their children. Each one was different, each with a distinct personality, each with the dignity of full personhood. I do not want to minimize the struggles these families go through. One mom said that most were just inches away from losing it completely on many days and that sometimes it was just hard to get out of bed to face changing a diaper on her 20-year-old. But, these parents would not have it any other way.

I learned very quickly that the three words these families never wanted to hear from physicians and others who provide care were “Quality of Life.” I was somewhat taken aback because as a palliative care physician I frequently use these words to describe what we do in terms of helping families cope, helping children deal with pain and other symptoms so that life may be as good as it can be up to and including the time of dying. Good palliative care is much more about living than dying. Apparently for these families, the phrase “quality of life” had been used by the medical establishment in judgmental ways perhaps in times of giving advice with regard to treatment or just general commentary. A remark soon after birth that sounds like, “I’m afraid your child won’t have a very good quality of life” may seem innocent but it is not. How easy it is to assume we know what a good quality of life is for anyone other than ourselves. We assess the burdens of care, the impact on siblings, the impact on parental relationships, the impact on finances and the utilization of resources. We perform ethical analyses assuming



FIG. 1. Olivia, Karah, and Spencer.

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we truly understand how to apply beneficence or maleficence to a child with trisomy 18 or 13 or any other disorder in which there may be profound disability. As if the child can tell us what he or she is feeling. But we can do our best to assess and treat pain and discomfort. Smiles and laughter need no score pad. We know what they mean. The advice we give may often be centered around our personal notions about quality of life. As a profession we are far from all being on the same page with such advice as was recently well described by Janvier et al. [2011] in an ethical case discussion about a child with trisomy 18. They point out the absence of data on outcome of procedures done on children with trisomy 18.

I write this not to advocate for any particular point of view except to advocate for a willingness to do whatever it takes, however long it takes, however many consults and team members it takes to fully

inform and understand the goals, values and aspirations parents may have for their children. Once we have done that we need to honor the parents by helping them achieve those goals whether it is comfort care alone, a full court press or something in between. Regardless of the choices of the family, they deserve our respect, our affirmation, our support, and part of our own humanity as we try to do our part to make their lives and the lives of their children as good as can be. Is not the privilege of doing that exactly why we are here?

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