

“Individuals Seeking Cures” Showcase

The Chromosome 18 Registry & Research Society: Overcoming Chromosome Abnormalities

Our mission is to help individuals with chromosome 18 abnormalities overcome the obstacles they face so they might lead healthy, happy and productive lives. When the Chromosome 18 Registry & Research Society was formed in 1990, most of the families were told there were only about 60 other families in the world with the same condition. There was limited information on the physical appearance and virtually no information on abilities, disabilities and outcomes. We set out to not only understand the medical and educational issues, but to define the genetic components and to use this information to create a treatment design. The abnormalities of chromosome 18 are rare, and the affected individuals have a complex phenotype involving many body systems. For these reasons, we decided to establish a single comprehensive research center. The Chromosome 18 Clinical Research Center is located in the Department of Pediatrics at the University of Texas Health Science Center at San Antonio. A single center means that all the information on a relatively small group of individuals is in one place and includes as many individuals as possible. The comprehensive clinical assessment is performed by one team so information is consistent. There is a single point of contact for the families to find out about research results and progress. To date, The Chromosome 18 Registry has 1700 families as members, and the Chromosome 18 Clinical Research Center has evaluated 233 individuals with chromosome 18 abnormalities; some as many as 5 times over 12 years. The study had produced 21 peer reviewed publications. This work was initiated and sustained primarily by funding from the families of the Chromosome 18 Registry who have provided almost 3 million dollars to the Center over the last 12 years. Fundraising has involved several annual organizational projects such as a Phantom Tea, Run for Research, Birdies for Charity and golf tournaments. Individual families have also held personal fundraisers including golf tournaments and comedy nights.

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