Profile

Felicia Bliss Axelrod: the dysautonomia pioneer

Almost 50 years ago, Felicia Bliss Axelrod established the Dysautonomia Center at the New York University School of Medicine (NY, USA). On arrival at the 9th floor of the New York University Langone Health Center, which is the location of the Dysautonomia Center, visitors are greeted with the view of a large wall full of photographs of babies, young children, and teenagers, all of them smiling. These photographs are of former patients, who never were considered mere clinical cases by the healthcare professionals at the Center. The staff not only attend to patients' clinical needs but also to their personal needs and desires. The extraordinary atmosphere in the Center reflects the soul of its founder, as Axelrod's positive energy continues to influence the place to the present day.

Felicia Axelrod was born in New York on Feb 3, 1941. Her father, Leon, was an engineer and a well known sculptor. Axelrod was married to Robert F Porges, a prominent obstetrician–gynaecologist and a professor at the New York University School of Medicine. Porges passed away in 2012. They had a son, John, and a daughter, Vicki.

For almost half a century, until her retirement as a professor of paediatrics in 2015, Axelrod was affiliated with New York University, where her focus was on providing care for children with autonomic and sensory disorders. She is now recognised as an outstanding pioneer in the treatment of these children. After completing her residency in 1969, Axelrod founded the Dysautonomia Center, where she combined clinical care with clinical research on the autonomic and sensory nervous systems, which at that time were in their infancy. Her prime interest was familial dysautonomia, also known as Riley–Day syndrome or hereditary sensory and autonomic neuropathy type III, a rare genetic disorder that was first described in 1949 by Conrad Ridley and Richard Day in five Ashkenazi Jewish children. The current patient registry at the Dysautonomia Center, started more than 40 years ago by Axelrod, now includes more than 700 individuals with familial dysautonomia and is the only comprehensive registry worldwide.

Patients with familial dysautonomia are a genetically homogeneous group, with lesions that essentially affect unmyelinated sensory fibres and the sympathetic nervous system. In addition to the characteristic inability to produce overflow emotional tearing, patients have difficulties with swallowing and walking, and are prone to centrally induced nausea and vomiting; they can also have large and unexpected swings in blood pressure and heart rate, as well as respiratory disorders, such as sleep apnoea. As Axelrod described, individuals with familial dysautonomia have reduced sensitivity to pain and temperature, and disturbed balance. Many patients also have scoliosis and can experience other orthopaedic conditions that heighten their risk of breaking bones.

While focusing on improving the quality of life of the patients and their families, Axelrod was determined to accelerate the pace of familial dysautonomia research to better understand the physiopathology of this disabling condition. Her research included fine descriptions of the symptomatology of the disease, natural history studies, and the investigation of therapeutic strategies. She also worked in partnership with geneticists and, together with Susan Slaugenhaupt and James Gusella (at Massachusetts General Research Institute and Harvard Medical School, Boston, MA, USA), reported the association of *IKBKAP*—now called *ELP1* (elongator protein 1 gene), which is located on the long arm of chromosome 9Q31—with the disorder. This finding led to the implementation of genetic screening, prenatal diagnosis, and further research into strategies to modify gene expression.

As the years went by, Axelrod became recognised worldwide as a top researcher in the field, and she encouraged international collaboration between clinical and preclinical research groups. She was also committed to offering the best quality of life to her patients by enhancing a holistic care programme. Axelrod developed a protocol in which the treatment of each patient was coordinated with their family and the various specialists who might be required for managing the protean symptoms, such as cardiologists, gastroenterologists, intensivists, ophthalmologists, pulmonologists, and sleep specialists. She said: "I can't give them a cure, but I can give them a team".

Among her many achievements, Axelrod was also an early member of the American Autonomic Society, and she was its president for several years. Now that she is retired, Axelrod enjoys other activities, such as spending time with her four grandchildren and other family members. She is a keen ballroom dancer and has participated in several dancing contests. Furthermore, Axelrod has co-written an inspiring children's book entitled *Why Won't Willy Fly?* This optimistic book reflects her own experiences working with children with dysautonomia, and it highlights the importance of friendship and resilience.

With such joy and harmony, determination, enthusiasm, hard work, perseverance, and rigor, it is easy to guess how Axelrod has improved life expectancy and quality of life for so many patients with dysautonomia, and why so many smiling faces are pictured on the wall of the Dysautonomia Center.

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For more on the **Dysautonomia Center** see https://nyulangone.org/care-services/dysautonomia-center

For more on **Axelrod's description of patients with autonomic neuropathy** see *Orphanet J Rare Dis* 2007; **2:** 39

For more on the **association of IKBKAP with familial dysautonomia** see *Am J Hum Genet* 2001; **68:** 598–605

For more on the **American Autonomic Society** see https://americanautonomicsociety.org/