

Longevity of a Woman With Down Syndrome: A Case Study

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Abstract: A case of a woman who is among the longest surviving people with Down Syndrome was described. The life expectancy of persons with Down syndrome has increased more than six-fold to 56 years since the turn of the century. The literature regarding life expectancy for persons with Down syndrome was reviewed, and the implications regarding Down syndrome and Alzheimer's disease were discussed.

Down syndrome is the most frequent genetic or chromosomal cause of mental retardation, occurring in approximately 1 per 800 to 1 per 1,000 live births in developed countries (Baird & Sadovnick, 1989). The syndrome is characterized by trisomy 21, particular morphologic features, mental retardation, and associated health problems. The average life expectancy for persons with Down syndrome has increased from 9 years in 1929 (Penrose, 1949) to 56 years today (Baird & Sadovnick, 1988, 1989; Dupont, Vaeth, & Videbech, 1986). In this paper we have described one of the longest surviving people with Down syndrome.

Case Presentation

In November 1993, an 83-year old woman with Down syndrome, "Ann," was brought to our attention by the National Association for Down syndrome. Two staff members from that organization had been visiting her socially since 1988. Her date of birth, November 3, 1910, was confirmed by her family and records provided by the nursing home where she resided. She was the youngest of six children, and her siblings and parents were all deceased. Her oldest sister died at age 45 years from breast cancer. The next sister died at the age 84, apparently from complications of colon cancer 10 years after the original diagnosis and placement of a colostomy. The next sibling, a brother, died at age 85 from heart disease. The next brother died when he was in his 30s from complications of diabetes mellitus. The next oldest sibling died in her 50s from heart disease. Her father died when he was in his 60s. We were unable to determine the cause of his death or that of her mother. No member of her family had been noted to have Alzheimer's disease or other form of dementia.

Ann moved to the nursing home several years prior when her last surviving sibling became ill and could no longer care for himself or her. The woman had good self-help skills, and although she never attended school, she could print neatly, had some reading skills, and knew her numbers. Although her speech, characterized by dysarthria,

was somewhat difficult to understand for those who did not know her, she communicated quite well.

Ann was hospitalized once in 1991 for pneumonia. In early 1993, she was diagnosed with hypothyroidism and started on levothyroxine 0.025 mg daily. She had moderate hearing loss, particularly in her left ear, and had osteoporosis. Her family members, the National Association for Down Syndrome staff, and the nursing home staff reported that she had no physical deterioration, no memory loss, and no loss of skills. She had been relatively healthy all her life; however, early in 1994, several months after our meeting, she suffered a fractured hip. After the hospitalization for surgery, she declined to eat adequate amounts, and her family and her physician decided against alternative feeding methods. The patient subsequently died.

The first author, a physician, visited her in November 1993. Her physical exam revealed an elderly woman with very short stature. Characteristic features of Down syndrome were noted, including flattened occiput, eyelids that slanted upward, prominent epicanthal folds, Brushfield's spots of the eyes, and a small palate. Ann's extremities had characteristic features, including bilateral valgus-curving fifth finger, small hands and feet, bilateral complete palmar crease, and enlarged space between the first and second toes and a plantar crease between the first and second toes. In addition, she had the following health problems that are found commonly in persons with Down syndrome: bilateral hallux valgus, bilateral cataracts, and dystrophic toenails consistent with onychomycoses. No heart murmur or other cardiac abnormality was found.

During the exam Ann showed the first author greeting cards and the corresponding photographs of the family member who had sent the card. Her stuffed animal collection was neatly displayed on her bed, and she placed them all back in their proper place after showing them to the first author.

At the time the first author visited, blood was drawn, and her thyroid function blood tests were normal (corrected with the levothyroxine). A chromosome analysis was abnormal, with a female karyotype consistent with mosaic Down syndrome: 75% of the cells had trisomy 21 and 25% had the normal 46 chromosome karyotype.

Discussion

At the time of Ann's death, the previously reported oldest persons with Down syndrome have been reported to be 63 (Forssman & Akesson 1965), 69 (Jancar, 1989), and 70 years old (Dupont et al., 1986). Demise, Ayres, and Briggs (1988) described a patient who was 75 years old in 1986. However, she was deteriorating rapidly at that time, and therefore, it is unlikely she lived much longer. Adlin (1993) noted that there are some reported cases of individuals with Down syndrome who had lived to the age of 85 but gave no further details. Therefore, to our knowledge , the woman we have described

in this article is the longest surviving person with Down syndrome documented in the literature.

In the past 85 years, the life expectancy for persons with Down syndrome has increased from less than 10 years to 56 years. Better treatment of congenital heart disease and infectious disease are a few of the recognized factors in the increased life expectancy (Chaney, Eyman, & Miller, 1985).

Forty to 50% of children with Down syndrome are born with congenital heart disease, and, as of 1987, 43% of those with this disease died before age 10 compared to only 15% of those without the disease (Baird & Sadovnick, 1987). Thase (1981) reported similar findings of 50% and 20%, respectively. The improvement of surgical techniques to correct congenital heart defects has contributed significantly to the increased life expectancy and should continue to decrease the discrepancy between the two groups (Stein & Susser, 1971).

Stein and Susser (1971) also reported that some of the increased life expectancy could be attributed to the decline in deaths due to infectious diseases. Respiratory infections are reported to be the first or second most common cause of death, with congenital heart disease being the other (Deaton, 1973; Forssman & Akesson, 1965; Oster, Mikkelsen, & Nielsen, 1975). Improved antibiotic therapy is hypothesized to contribute to the reduced death from infectious disease.

More recently, Eyman, Call, and White (1991) found another risk factor for early death of individuals with Down syndrome. Lack of mobility or poor feeding skills were better predictors for early death than were medical problems associated with congenital heart disease. Eyman, Grossman, Chaney, and Call (1990) showed that individuals with severe mental retardation would be more likely to have lack of mobility or feeding skills and lower survival rates. Perhaps as our ability to repair congenital heart defects and treat infections has improved, lack of mobility or feeding skills has played a greater role in early death.

Of particular interest about the patient reported in this article is that she was reported to have no decline in mental function and performance of activities of daily living. As indicated, we interviewed several people who knew her and reviewed videotape taken of her several years prior. There was no obvious decline noted. By family choice, no autopsy was performed. However, results of previous studies suggest that neuropathologic changes consistent with Alzheimer's disease would have likely been found in our patient because they appear to be universal in persons with Down syndrome after age 40 (Malamud, 1966). Recent findings, however, have suggested that clinical Alzheimer's disease is not found in all persons with Down syndrome (Chicoine, McGuire, Hebein, & Gilly, 1994). An 83-year-old woman with Down syndrome, without indication of decline in function, is further evidence (albeit anecdotal) that investigators should reassess previous reports that all persons with Down syndrome develop clinical Alzheimer's disease.

The longevity of our 83 year-old patient can probably be attributed to the absence of congenital heart disease and the fact that she was not institutionalized at a time many years ago when conditions in institutions tended to be poor. Although more difficult to prove, a contributing factor may have been her warm, supportive family, who helped provide her with a meaningful life. Life expectancy is on the rise for persons with Down syndrome. Further increases should be expected as there is less reliance on congregate care facilities and better access to good medical care. As the population of older persons with Down syndrome and other developmental disabilities increases, families and careproviders will have to continue to evaluate and adjust for the changing needs of this population.

References

- Adlin, M. (1993). Health care issues. In E. Sutton, A. Factor, B. Hawkins, T. Heller, & G. Seltzer (Eds.), *Older adults with developmental disabilities* (pp. 48-60). Baltimore: Brookes.
- Baird, P.A., & Sadovnick, A.D. (1987). Life expectancy in Down syndrome. *Journal of Pediatrics*, 110, 849-854.
- Baird, P.A., & Sadovnick, A.D. (1988). Life expectancy in Down syndrome adults. *Lancet*, II, 1354-1356.
- Baird, P.A., & Sadovnick, A.D. (1989). Life tables for Down syndrome. *Human Genetics*, 82, 291-292.
- Chaney, R.H., Eigheten, R.K., & Miller, C.R. (1985). The relationship of congenital heart disease and respiratory infection mortality in patients with Down's syndrome. *Journal of Medical Deficiency Research*, 29, 23-27.
- Chicoine, B., McGuire, D., Hebein, S., & Gilly, D. (1994). Development of a clinic for adults with Down syndrome. *Mental Retardation*, 32, 100-106.
- Deaton, J.G. (1973). The mortality rate and causes of death among institutionalized mongols in Texas. *Journal of Mental Deficiency Research*, 17, 117-122.
- Demise, A., Ayres, R.C., & Briggs, R. (1988). Old age in Down's syndrome. *Journal of the Royal Society of Medicine*, 81, 740.
- Dupont, A., Vaeth, M., & Videbech, P. (1986). Mortality and life expectancy of Down's syndrome in Denmark. *Journal of Mental Deficiency Research*,

30, 111-120.

- Eyman, R.K., Call, T.L., & White, J.F. (1991) Life expectancy of persons with Down syndrome. *American Journal of Mental Retardation*, 95, 603-612.
- Eyman, R.K., Grossman, H. J., Chaney, R.H., & Call, T.L. (1990). The life expectancy of profoundly handicapped people with mental retardation. *New England Journal of Medicine*, 323, 584-589.
- Frossman, H., & Akesson, H.O. (1965). Mortality in patient's with Down syndrome. *Journal of Mental Deficiency Research*, 9, 146-149.
- Jancar, J. (1989). Old age in Down's syndrome (letter). *Journal of the Royal Society of Medicine*, 82, 317-318.
- Malamud, N. (1966). The neuropathology of mental retardation. In I. Philips (Ed.), *Prevention and treatment of mental retardation* (pp. 24-32). New York: Basic Books.
- Oster, J., Mikkelsen, M., & Nielsen, A. (1975). Mortality and life-table in Down's syndrome. *Acta Paediatrica Scandanavica*, 64, 322-326.
- Penrose. L. S. (1949). The incidence of mongolism in the general population. *Journal of Mental Science*, 95, 685-688.
- Stein, Z.A., & Susser, M.(1971). The preventability of Down's syndrome. *HSMHA Health Reports* , 86, 650-658.
- Thase, M. E. (1981). Longevity and mortality in Down's syndrome. *Journal of Mental Deficiency Research*, 26, 177-192.
- Thomas, P. (1986, February 24). Special adults: New challenges to primary care MDs. *Med World News*, 68-81.

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