Brian's Story

Brian was born in 1989 to affluent parents; Hugh and Gladys who were in their early forties. When Gladys found out she was pregnant in her forties, her doctor suggested that Gladys be tested for Down syndrome. Gladys refused to have an amniocentesis done. He had an older brother, Paul who was away at boarding school when he was born. Brian was diagnosed with Trisomy 21. There are three types of Down syndrome: Trisomy 21; Translocation Down syndrome; Mosaic Down syndrome. The diagnosis was suspected based on Brian's physical appearance at birth. It was confirmed by analysis of his chromosomes.

Gladys and Hugh were devastated by the news that their child was "less than perfect". They could not possibly bring this child home, what would their friends and business associates think? This child would require too much time and care for their lifestyle. Their view evolved and changed after reading about Down syndrome, learning the benefits of therapies like early intervention, and long discussions with peer groups (e.g. other parents of children with Down syndrome).

Brian had difficulty breathing and the physician noticed Brian had blue-tinged skin, a heart murmur (an abnormal whooshing sound caused by turbulent blood flow). Several tests were ordered:

- Echocardiogram
- Electrocardiogram
- Chest x-ray
- Oxygen level measurement
- Cardiac catheterization

Brian was diagnosed with Tetralogy of Fallot, a rare condition caused by a combination of four defects that are present at birth (congenital). These defects cause oxygen-poor blood to flow out of the heart and to the rest of the body.

Tetralogy of Fallot occurs during fetal growth, when the baby's heart is developing. While factors such as poor maternal nutrition, viral illness or genetic disorders might increase the risk of this condition, in most cases the cause of tetralogy of Fallot is unknown.

The lungs of children with Down syndrome do not develop as fully as in the general population. Consequently, the growth of blood vessels throughout the lungs is limited. The narrowed arteries of the lungs hold potential for lasting consequences due to the increased pressure and flow of blood through the lungs.

Treatment

Surgery is the only effective treatment for Tetralogy of Fallot. Brian was two weeks old when he required temporary surgery due to his underdeveloped pulmonary arteries (hypoplastic). A bypass (shunt) was created between a large artery that branches off from the aorta and the pulmonary artery.

After six months, the cardiologist deemed Brian strong enough to undergo ‘intracardiac repair’. This is an open-heart surgery that involves several repairs:

- Removal of the shunt
- Patch over the ventricular septal defect to close the hole between the ventricles
- Repair or replace the narrowed pulmonary valve and widens the pulmonary arteries to increase blood flow to the lungs

The surgery was a success and Brian was eventually discharged home.
He had around the clock care which included:

- Nursing
- Physiotherapist
- Occupational therapist
- Respiratory therapist
- Play workers
- Tutor

Brian required regular medical follow-up to maintain good health.

- Routine follow-up care – regular check-ups with a cardiologist, primary physician routine exams, medications that are prescribed, routine dental care
- Heart-healthy lifestyle – heart-healthy eating, physical activity, maintaining healthy weight
- Emotional health – may feel isolated, sadness, and frustration

Brian’s only interactions were with the “hired help”. As a pre-teen and teen, he formed strong bonds with his workers and struggled with changes in staff and routine. This led to frustration and anger.

Results of Prenatal Screening for Down's Syndrome in Maine between 1980 and 1993.

Variable	1980-1985	1986-1990	1991-1993	Total
Live births • Total no. • Maternal age <20 yr-% • Maternal age ≥35 yr-%	96,287 13.4 4.1	82,335 11.2 6.8	46,934 10.5 8.7	225,556 12.0 6.1
Cases of Down's syndrome Expected* • No. of cases • % with maternal age < 35 yr Ascertained± • No. of cases • % with maternal age < 35 yr	97.4 80 85.2 79	95.1 72 99.3 61	60.3 65 61.5 75	252.9 73 245.3 71
Cases of Down's syndrome identified By amniocentesis alone • No. of cases • No. of pregnancies terminated By serum screening • No. of cases • No. of pregnancies terminated • After birth-no. of cases	9 8 0 0 78	20 19 15 10 71	10 10 30 27 30	39 37 45 37 179
Reduction in prevalence of Down's syndrome among live births-%¥	7.2	22.5	46.3	23.2

*The expected number of cases is based on the maternal age distribution and the maternal-age-specific prevalence of Down's syndrome among live births.

±The ascertained number of live-born infants with Down's syndrome, in the absence of prenatal diagnosis and selective termination of pregnancy, was calculated as the number of live-born infants with Down's syndrome that was not detected prenatally + the number of live-born infants with Down's syndrome that was detected prenatally + (the number of terminated pregnancies x 0.77), which takes into account spontaneous loss.

¥The prevalence of Down's syndrome among live births was calculated as 1 – (the number of live-born infants with Down's syndrome not detected prenatally + the number of live-born infants with Down's syndrome detected prenatally)/the total number of ascertained cases of Down's syndrome.

- **Prenatal Screening for Down's Syndrome in Maine, 1980 to 1993**
  Palomaki G.E., Haddow J.E. and Beauregard L.J. | N Engl J Med 1996; 334:1409-1410