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Spring 2021

A Timeline of Down Syndrome

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Recommended Citation

Guese, Christian; McInnes, Kristina; and Miranda, Kimberly, "A Timeline of Down Syndrome" (2021). *Undergraduate Research Symposium Posters*. 22. https://digitalscholarship.unlv.edu/durep_posters/22

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<u>1773</u>: Eastern State Hospital in Williamsburg, VA is the first institution for the mentally ill in The United States

1883: Americans embrace "Eugenics," coined by Sir Francis Galton, which advocated for selective mating, "breeding out" those with disease, disabilities, and "undesirable" traits.

meiosis ---> fertilization nondisjunction mitosis

770-1870 1871-1913 1914-1929

<u>1866</u>: John Langdon Down – **English Physician and** advocate for patients with intellectual disabilities. Used the term "Mongoloid" to describe phenotype, which varies in severity and in penetrance, with no predilection to any population.

isomy of chromosome 21 Alzheimer's disease Dysmorphic Learning difficulties & **Round Face Developmental Delay** (Mean IQ of 50%) ****** 10 Brushfield Small Ears spots on Flat occiput Protruding tongu (not macrogloss Abundant Neck Skin though! Atlantoaxial instability Hypothyroidism Single palr crease Cardiac defects in 50% (Ventricular septal defect Incurved Patent ductus arteriosus little fingers Atrial septal defect (Clinodactyly) Respirator nfection Gap between Increased risk 1st & 2nd toes of Leukaemia (Sandal to gap)

ww.PicturingMedicine.com

uble bubble sign on AXR)

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<u> 1929</u>: Life Expectancy is only 9 years old



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A Timeline of Down Syndrome

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Maternal Age	Incidence of Down Syndrome
20	1 in 2,000
30	1 in 900
40	1 in 100
49	1 in 10



<u>1933</u>: Study discovers a link between the age of the mother and the occurrence of DS.⁵

<u>1932</u>: Nondisjunction, a novel chromosomal disorder, first suggested cause of Down Syndrome $(DS).^{4}$

<u>1965</u>: World Health Organization accepts "Down Syndrome" as the standard term.

<u>1961</u>: Mosaicism, a third genetic variation, was discovered. Individuals may not display all common phenotypic traits in DS.(8)



<u>1960</u>: Translocation, a second genetic variation of DS, was discovered. This has the only form of inheritance with a risk of 3 in 1000 non-karyotyped births.⁸

<u> 1946:</u>

Life Expectancy is 12 years old.

<u>1930-1939</u> <u>1940-1952</u>

Benjamin Spock suggests that babies with DS should be institutionalized.

1948: First publication about association between Alzheimer's Disease (AD) and DS.⁶



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Biomarkers in Aging Adults with Down Syndrome: Systematic Review", Current Psychiatry Research and Reviews

<u>1959</u>: Dr. Jerome Lejeune discovered Trisomy 21 (T21). Causes 95% of DS cases across the US. Risk increases with maternal age. This genetic variation has no environmental influences.



<u>1961</u>: John F. Kennedy established National Institute of Child Health and Human Development to conduct and support research on intellectual disabilities.



Have an extra 21 in some

Some cells are normal with two copies of chromosome

<u>1981</u>:

DS Preventative Medical Checklist," edited by William I. Cohen, presents the first U.S. medical management suggestions for doctors.

<u>1979</u>: Life Expectancy increased to 25 years.

<u>2004</u>: Down Syndrome linked to gene dosage, due to duplication of part/all of chromosome, increasing dosage of any single gene.⁷

Life Expectancy increased to 60 years. Early intervention and specialized therapies, help improve a child with DS motor and cognitive development. There is still potential to improve with clinical care and health care disparities.

Dr. Thessa Hilgenkamp, at UNLV, is conducting a NIH-funded research to better understand the cause of fatigue and exercise intolerance for individuals with DS.

Prenatal screening for DS poses ethical concerns. Lowest DS population in Iceland with an increase in elective abortions. Countries without elective abortions have higher rates of DS.

<u>1968</u>: DS is first detected with amniocentesis, a study to diagnose fetal abnormalities.

Amniocentesis

<u>1970s:</u> Public schools were required provide a free and appropriate education to children with DS.

1976: Abortions increased at 3rd trimester after parents were informed of DS diagnosis.

<u>1990</u>: George H. W. Bush signs The Americans with Disabilities Act.



<u>1995</u>: Link between miscarriages and chromosomal abnormalities. Discovered that 32% of DS pregnancies miscarried before 16 weeks, and a total of 54% were lost by term.⁹

<u>1997</u>: Life Expectancy increased to 47 years.





Today:

<u>2010</u>: Rosa's Law, signed by Barack Obama, removes words such as "retardation" from legislation.





<u>2012</u>: Global Down Syndrome Foundation and National Down Syndrome Congress published a patient education pamphlet for prenatal testing.

2017: T21 linked to an overdose of mRNA and proteins, dysregulating cellular functions.³

<u>2019</u>: AD study connecting over production of APP gene on T21.¹⁰

overproduction of APP protein