

Spring 2021

## A Timeline of Down Syndrome

Christian Guese

*University of Nevada, Las Vegas*

Kristina McInnes

*University of Nevada, Las Vegas*

Kimberly Miranda

*University of Nevada, Las Vegas, mirank6@unlv.nevada.edu*

Follow this and additional works at: [https://digitalscholarship.unlv.edu/durep\\_posters](https://digitalscholarship.unlv.edu/durep_posters)



Part of the [Medical Genetics Commons](#)

---

### 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](https://digitalscholarship.unlv.edu/durep_posters/22)

This Presentation is protected by copyright and/or related rights. It has been brought to you by Digital Scholarship@UNLV with permission from the rights-holder(s). You are free to use this Presentation in any way that is permitted by the copyright and related rights legislation that applies to your use. For other uses you need to obtain permission from the rights-holder(s) directly, unless additional rights are indicated by a Creative Commons license in the record and/or on the work itself.

This Presentation has been accepted for inclusion in Undergraduate Research Symposium Posters by an authorized administrator of Digital Scholarship@UNLV. For more information, please contact [digitalscholarship@unlv.edu](mailto:digitalscholarship@unlv.edu).



# A Timeline of Down Syndrome



Christian Guese, Kristina McInnes, Kimberly Miranda  
School of Life Sciences, University of Nevada Las Vegas

**Today:**

**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.

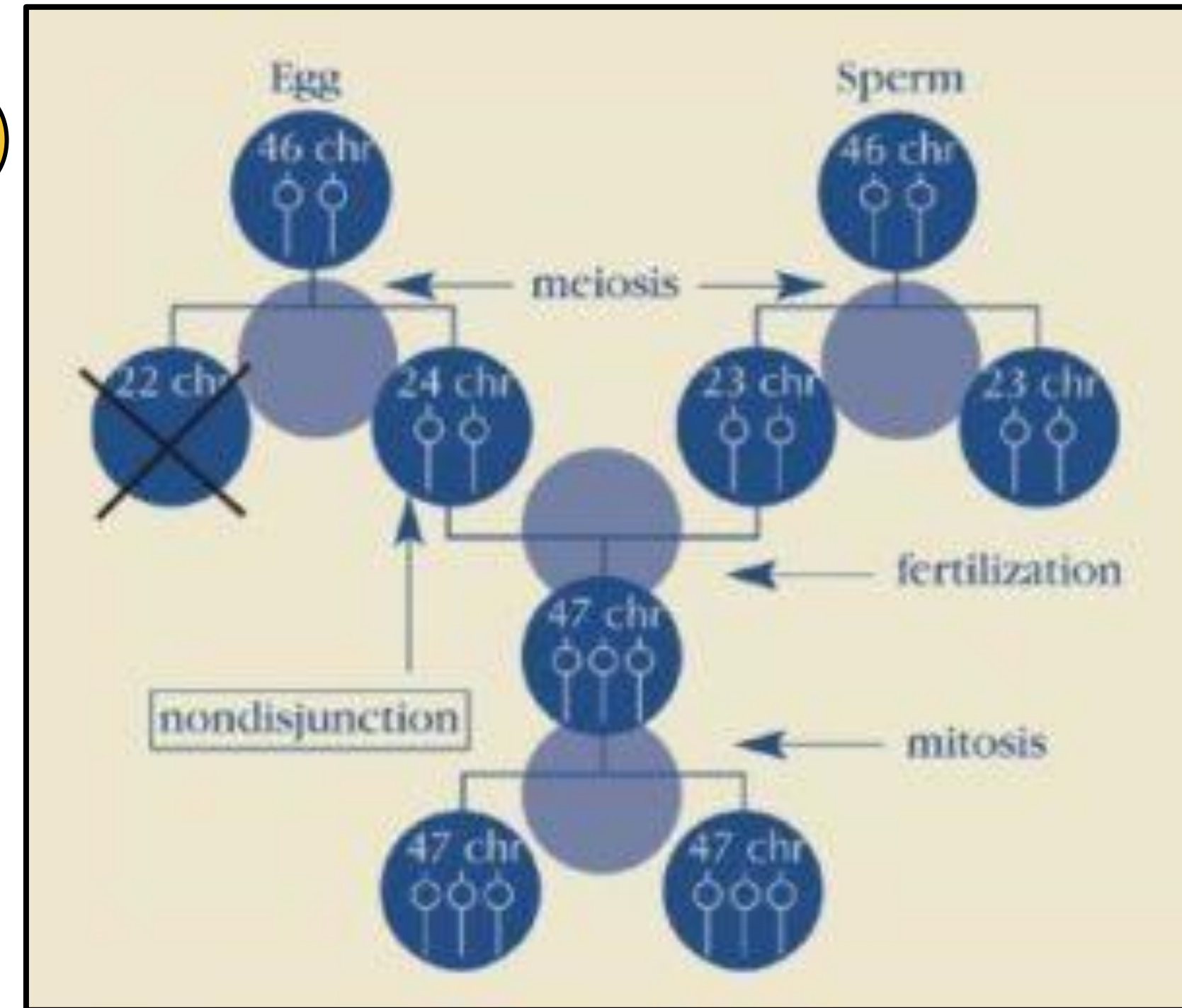
Prenatal screening for DS poses ethical concerns. Lowest DS population in Iceland with an increase in elective abortions. Countries without elective abortions have higher rates of DS.

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



**1773:** 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.



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

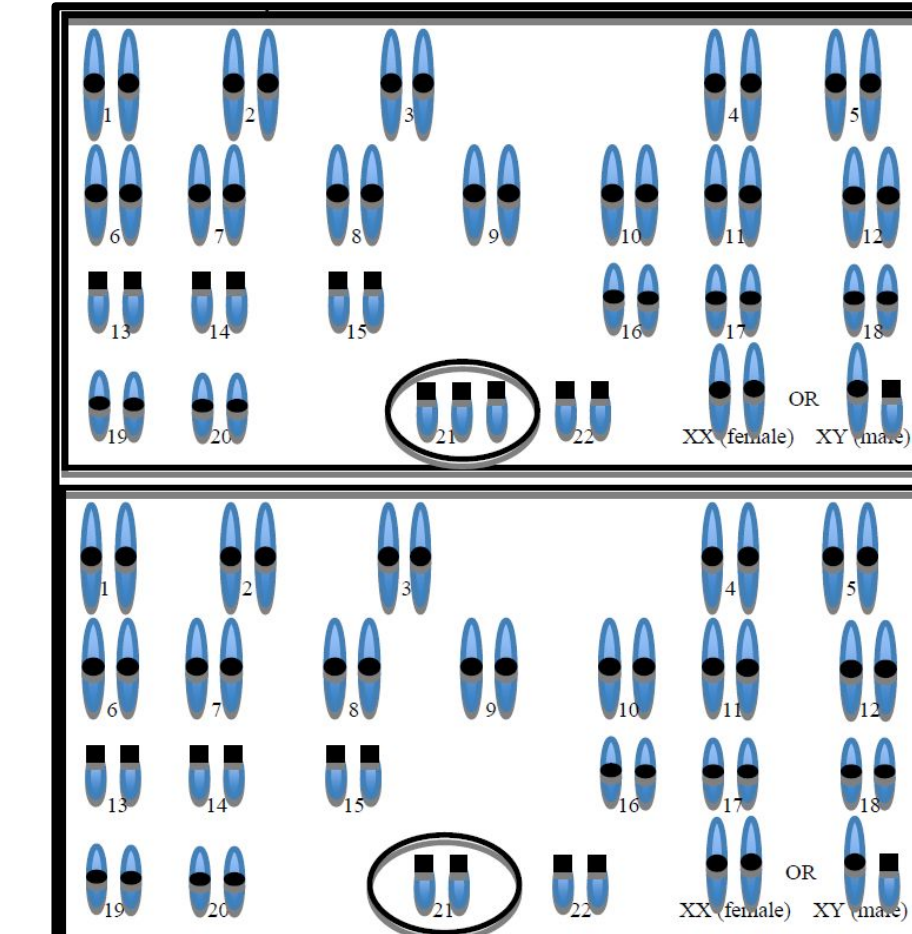
**1933:** Study discovers a link between the age of the mother and the occurrence of DS.<sup>5</sup>

**1932:** Nondisjunction, a novel chromosomal disorder, first suggested cause of Down Syndrome (DS).<sup>4</sup>

**1965:** World Health Organization accepts "Down Syndrome" as the standard term.

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

Karyotype of an individual with mosaic DS



Have an extra chromosome 21 in some cells.

Some cells are normal with two copies of chromosome 21.

**1960:** 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.<sup>8</sup>

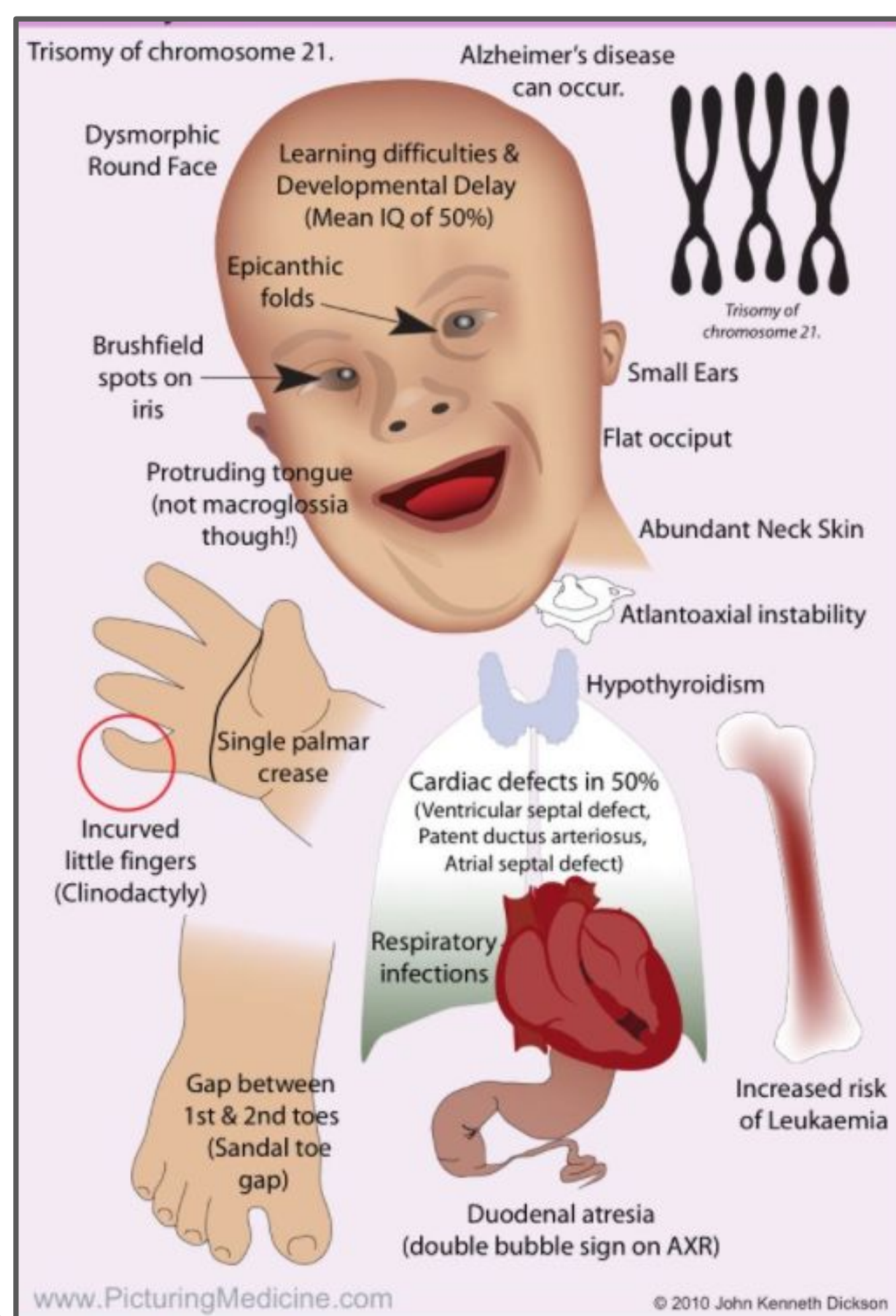
**1981:** DS Preventative Medical Checklist," edited by William I. Cohen, presents the first U.S. medical management suggestions for doctors.

**1979:** Life Expectancy increased to 25 years.

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



**1866:** 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.



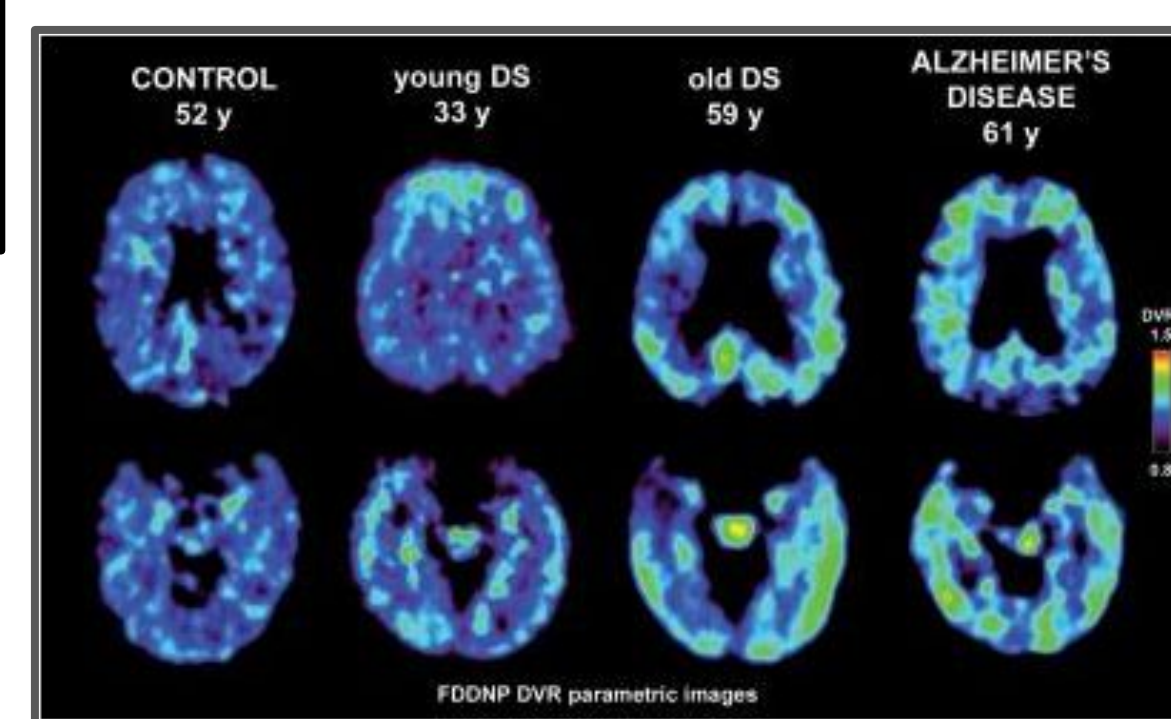
**1929:** Life Expectancy is only 9 years old



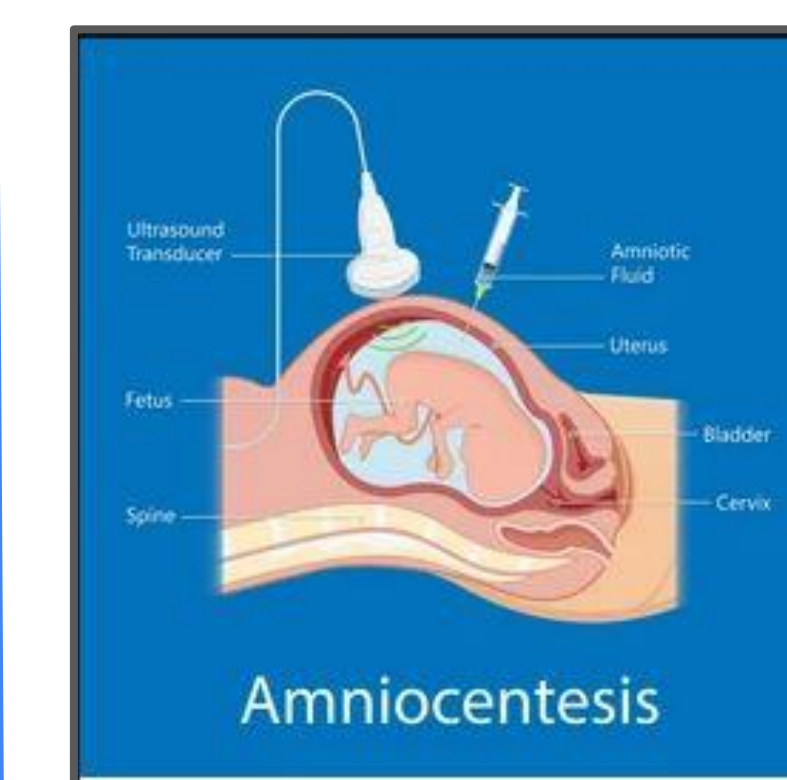
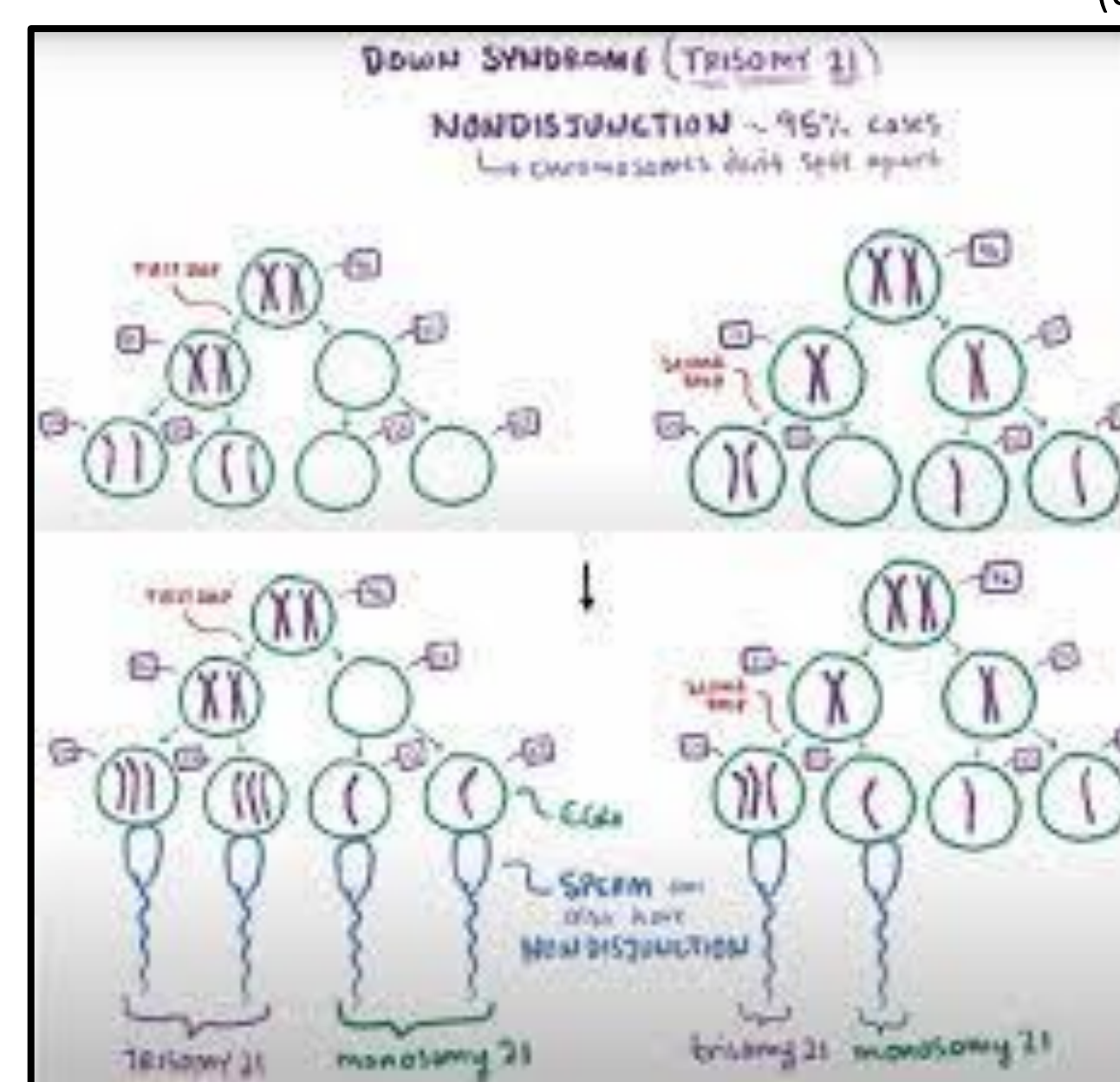
**1946:** Life Expectancy is 12 years old.

Benjamin Spock suggests that babies with DS should be institutionalized.

**1948:** First publication about association between Alzheimer's Disease (AD) and DS.<sup>6</sup>



**1959:** 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.



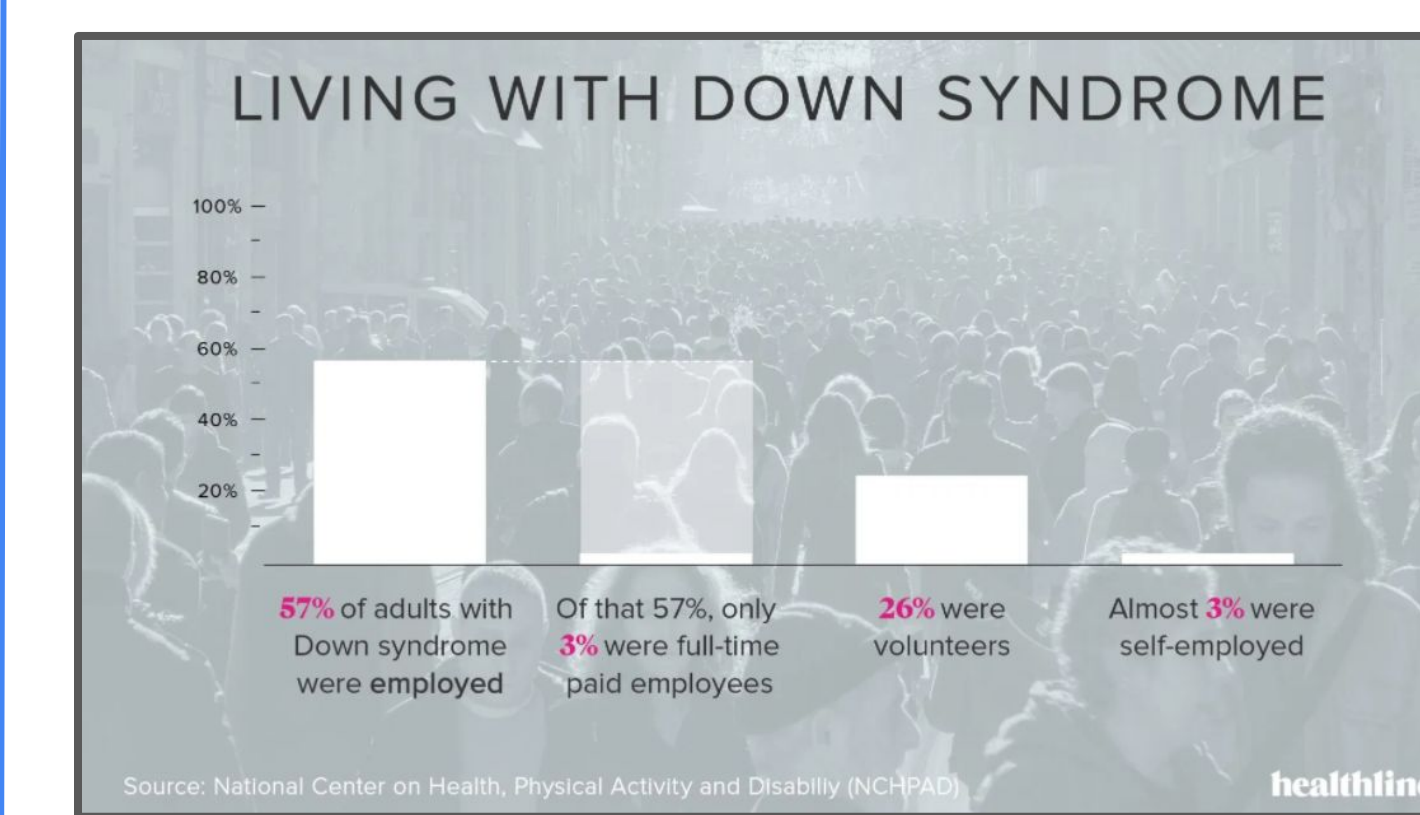
**1968:** DS is first detected with amniocentesis, a study to diagnose fetal abnormalities.

**1970s:** Public schools were required provide a free and appropriate education to children with DS.

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



**1990:** George H. W. Bush signs The Americans with Disabilities Act.



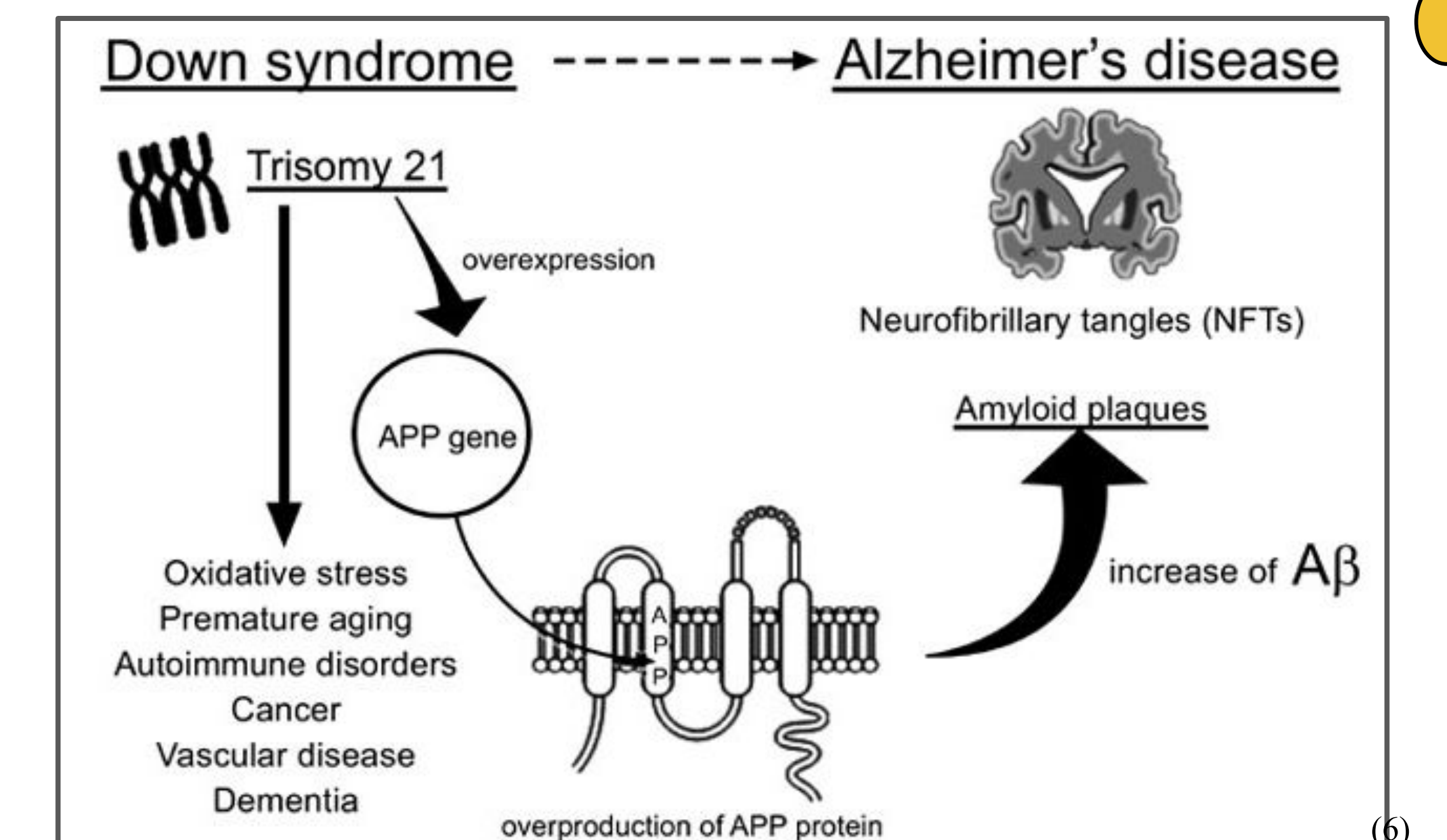
**1995:** Link between miscarriages and chromosomal abnormalities. Discovered that 32% of DS pregnancies miscarried before 16 weeks, and a total of 54% were lost by term.<sup>9</sup>

**1997:** Life Expectancy increased to 47 years.

**2012:** 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.<sup>3</sup>

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



References:

1. Esquirol, J.E.D. (1838). Des maladies mentales considérées sous les rapports médical, hygiénique et médico-légal (Brussels, J.B. Tischer).
2. Down, J. L. H. Observations on an ethnic classification of idiots. Lond. Hosp. Rep. 3, 259–262 (1866); reprinted in Ment. Retard. 33, 54–56 (1995).
3. Liu, Yansheng et al. "Systematic proteome and proteostasis profiling in human Trisomy 21 fibroblast cells." Nature communications vol. 8, 1 1212. 31 Oct. 2017, doi:10.1038/s41467-017-01422-6
4. Davenport, C. B. in Sixth International Congress of Genetics Vol. 1. (ed. Jones, D. F.) 135–140 (Brooklyn Botanic Garden, New York, 1932).
5. Penrose L.S. The relative aetiological importance of birth order and maternal age in Mongolism. Proc R Soc B Biol Sci. 1934;115:431–50
6. Jervis, G.A. Early senile dementia in mongoloid idiocy. Am. J. Psychiatry 105, 102–106 (1948).
7. Gardiner K. (2004). Gene-dosage effects in Down syndrome and trisomic mouse models. Genome biology, 5(10), 244. doi: 10.1186
8. Lister TJ, Frola-Pesson O. Recurrence risks for Down syndrome. Hum Genet. 1980;55(2):203-8. doi: 10.1007/BF00291768. PMID: 6450157.
9. Macintosh MC, Wald NJ, Chard T, Hansen J, Mikkelsen M, Therkelsen AJ, Petersen GB, Lundsteen C. Selective miscarriage of Down's syndrome fetuses in women aged 35 years and older. Br J Obstet Gynaecol. 1995 Oct;102(10):798-801. doi:10.1111/j.1471-0528.1995.tb10845.x. PMID: 7547736.
10. J.F. Bram, L.L. Talib, H.G. Joaquin, C.L. Carvalho, W.F. Gattaz and O.V. Forlenza\*. "Alzheimer's Disease-related Biomarkers in Aging Adults with Down Syndrome: Systematic Review", Current Psychiatry Research and Reviews (2019) 15: 49. doi:10.2174