



Libyan International Medical University Faculty of Basic Medical Science

Rates of Down Syndrome in Libya

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Abstract:

This report shall be discussing a rather common genetic disorder known as down syndrome, along with some of the risk factors that may increase the chances of a baby being born with the disease. The report will also aim to compare the different rates of down syndrome in different countries with a main focus being the rates of the disorder in Libya, comprehending the reasons for the abnormally high levels of the disease in Libya shall also be an aim of study for this report. The factors involved shall include both medical and socioeconomic factors that may help explain the rate.

Introduction:

Down syndrome is a genetic disorder caused by the presence of all or part of a 3rd copy of chromosome 21^[1] but 95% of those with the disease have a full 3rd chromosome, 4% show extra chromosomal material translocation from the long arm of chromosome 21 to chromosome 14 or 22 and 1% are mosaics having a mixture of the two^[4]. This occurs by chance, and the disorder is known as the most common genetic disorder of our time having average rates of up to 1/700, and the number seems to be on the rise, this can be shown by the increase of rates of the disorder by 30% between 1979 and 2003^[2]. The disease that accounts for 8% of all congenital anomalies in Europe^[3] cannot be cured and usually results from normal parents. Down syndrome may lead to many complications such as mental retardation and increased risks of leukemia. Maternal age has strong influence on the rates of down syndrome and may be the only known risk factor of the disease, this can be seen when we compare the average rates for 20 year old woman which is around 1/1550 to the frequency of the disease in woman older than 45 which is as high as 1/25. The correlation with maternal age suggests that in most cases the meiotic nondisjunction of chromosome 21 occurs in the ovum. Indeed, in 95% of cases the extra chromosome is of maternal origin. The reason for the increased susceptibility of the ovum to nondisjunction is not fully understood. No effect of paternal age has been found in those cases in which the extra chromosome is derived from the father^[4]. The best treatment of the disease can be providing extensive medical support with educational support and a sheltered environment helping growth and survival with all these factors provided life expectancy can be as high as 50 to 60 years^[5].

Rates of Down Syndrome vary greatly from country to country depending on overall lifestyle, but also deaths due to the disorder vary greatly, and this is heavily due to differences in the treatment and care provided to them.

Discussion:

The genetic disorder known as Down Syndrome has one known risk factor and that is maternity age, and the increase in rates with increased age can be clearly seen when comparing the frequency of down syndrome births of mothers of different ages.

Rates of Down Syndrome average 1/700 worldwide^[2], but like stated the number varies from country to country, rates in the USA are around 1/714^[6] while rates in European countries such as Norway are as low as 1/909^[7]. Both these countries are considered to have low rates of the disease especially when compared to Libya, with its 1/516 birth frequency for children with Down Syndrome^[8].

When taking into consideration the main risk factor (maternal age) we can compare the age of mothers using different methods. The average age of mothers at first pregnancy in Denmark is 29.1^[9] which is relatively very high, hence it's safe to assume that most births are to females of over 30 years old, while in Libya only 36%^[8] of births are to mothers over 30 years old. When we compare the rates in Denmark to rates in the USA we can see that even though the USA has a lower average age of mothers at first birth (25.6)^[9] the rates of down syndrome are significantly higher than those in Denmark.

When this is taken into consideration the high rates of Down syndrome in Libya when compared to the average worldwide frequency may be due to a more cultural reason, after the introduction of prenatal screening the rates of down syndrome and the overall pattern of frequency was heavily affected. Simply put countries in which abortion was allowed saw a steady decrease in the rates of live births (even though the number of people living with down syndrome increased as a result of better care) while the average age of birth of the mother steadily increased^[3].

It was found that 67% of Down Syndrome pregnancies where terminated in the USA^[10] and up to 92% in Europe^[11], hence this social factor may also heavily influence the world average making the rates in countries where abortion does not occur seem abnormally high due to other medical reasons. Indeed when the rates in the USA back in 1950 were compared to modern rates it was found that the sharp decrease was directly correlated with the prevalence of parental screening (back in 1950 rates of Down Syndrome in the USA were as high as 1/500 even though woman were marrying younger back then)^[6].

So when Libya was compared to other countries with a similar cultural environment such as the UAE it was found that the rates were relatively similar, it being higher in the UAE $1/449^{[12]}$, a country which is considered as a 1^{st} world country with high standards of medical care.

Even when we compare the frequency of the disease to more western countries such as Ireland where abortion is also illegal rates were also higher than normal average being at 1/546^[13].

Conclusion:

While the rates of Down Syndrome in Libya may be considered high when compared to the worldwide average we can safely assume that the most likely cause to the abnormal number is more of a cultural cause than a medical cause. So prevalence of Down Syndrome in Libya is due to Socioeconomic reasons, slightly due to the relatively old age of females at marriage and age at child birth, and mainly due to the lack of abortion in Libya.

I do however recommend more research into the matter to allow a more accurate comparison to other countries with more reliable results.

References:

- 1)Patterson, David. "Molecular genetic analysis of Down syndrome." Human genetics 126.1 (2009): 195-214.
- 2)https://www.cdc.gov/ncbddd/birthdefects/downsyndrome/data.html
- 3) Weijerman, Michel E., and J. Peter De Winter. "Clinical practice." European journal of pediatrics 169.12 (2010): 1445-1452.
- 4)basic patho 8th
- 5) Kliegma, Robert M. (2011). "Down Syndrome and Other Abnormalities of Chromosome Number". Nelson textbook of pediatrics. (19th ed.). Philadelphia: Saunders. pp. Chapter 76.2.
- 6) child neurology 7th edition page 228
- 7)Malt, E. A., et al. "Helse og sykdom hos voksne med downs syndrom." Tidsskrift for den Norske Laegeforening 133.3 (2013): 290-294.
- 8) Verma, I. C., et al. "Cytogenetic analysis of Down syndrome in Libya." The Indian Journal of Pediatrics 57.2 (1990): 245-248.
- 9)https://www.cia.gov/library/publications/the-world-factbook/fields/2256.html
- 10)Natoli, Jaime L., et al. "Prenatal diagnosis of Down syndrome: a systematic review of termination rates (1995–2011)." Prenatal diagnosis 32.2 (2012): 142-153.

- 11) Mansfield, Caroline, Suellen Hopfer, and Theresa M. Marteau. "Termination rates after prenatal diagnosis of Down syndrome, spina bifida, anencephaly, and Turner and Klinefelter syndromes: a systematic literature review." Prenatal diagnosis 19.9 (1999): 808-812.
- 12) Murthy, Sabita K., et al. "Incidence of Down syndrome in Dubai, UAE." Medical Principles and Practice 16.1 (2007): 25-28.
- 13)http://www.irishhealth.com/article.html?con=64