Epidemiology of Turner syndrome in Iceland 1968-2012

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Introduction:

Turner syndrome (TS) is a common genetic disorder with an estimated range of occuring in 25-210 per 100,000 liveborn females. TS has been extensively studied in Denmark with an estimated prevalence of 40 per 100,000 liveborn females.[1] Our aim was to study the epidemiology of TS in Iceland for the period of 1968 – 2012.

Methods:

Primary source of data were hospital records and records from all pediatric endocrinologists in Iceland. To validate the data the karyotypes were obtained from the chromosomal laboratory which is the only cytogenetic laboratory in Iceland, serving all hospitals and private physicians since 1968.

Results:

A total of 51 females were diagnosed with TS during the 45 year period. Cases diagnosed in the first year of life were 16 (31%). The median age of diagnosis was seven years. Five were diagnosed after the age of 16, the oldest at 59 years. The incidence of TS, computed for five year periods, was on average 1 per 1876 liveborn females and the prevalence 53 per 100,000 females. Induced abortions on TS diagnosed fetuses were 19.

Almost half of the TS females (49%) had the classical karyotype 45,X, whereas in fetuses the 45,X karyotype was found in nearly 80% of cases. Various mosaic karyotypes were seen, most commonly 45,X/46,XX (11%) and 45,X/46,X,i(Xq) (isochromosome q) (10%).

Period	No. diagnosed	Live born	TS per liveborn girls
1968-1972	8	10407	1 per 1300
1973-1977	5	10426	1 per 2085
1978-1982	9	10591	1 per 1177
1983-1987	6	9980	1 per 1663
1988-1992	4	11261	1 per 2815
1993-1997	4	10619	1 per 2655
1998-2002	3	10219	1 per 3406
2003-2007	7	10553	1 per 1508
2008-2012	5	11596	1 per 2319
Total	***51***	95652	*1 per 1876*

Table I *Incidence calculated per five year interval.*

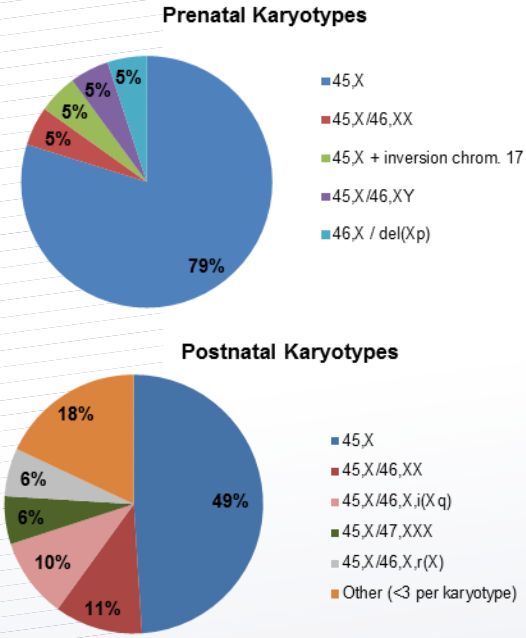

Figure I Above: *The genetic background for TS phenotype is highly variable, with 45,X being the predominant karyotype in both pre- and postnatal TS.*

Conclusion:

The prevalence of TS in Iceland is higher than that reported from Denmark.
A higher percentage of the classical 45,X karyotype was seen in the aborted fetuses. For the last 2 decades most of the girls have been diagnosed in the first year of life or at a relatively young age. A diagnostic delay was seen in many cases in the earlier years as some females were not diagnosed until adulthood.
Clinical vigilence is important for early diagnosis of Turner syndrome.

Ref. 1: Gravholt CH. Epidemiological, endocrine and metabolic features in Turner syndrome. Eur J Endocrinol. 2004 Dec; 151(6):657-87.