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Nationwide study of Turner syndrome in Ukraine

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Abstract

Turner Syndrome (TS) is one of the most common genetic disorders associated with abnormalities of chromosome X that occurs in different populations with a frequency of 25-210 per 100 000 female live births [1].

Objective and hypotheses

The aim of this study was to investigate the prevalence of TS in Ukrainian children, frequency of different variations of karyotype and age of primary diagnosis of TS.

Methods

We analyzed the database of the Ukrainian Registry of children with dwarfism (2005-2015 y.y.), that include 453 girls with TS 11 month-18.2 y.o. The Register contains information on the age of first diagnostic of TS, the age at the start of GH therapy, the results of basic clinical examination: height, weight, pubertal stage by Tanner, karyotype, the results of hormonal, instrumental and radiological studies, the results of rGH therapy.

Results

Prevalence of TS in Ukraine is 0.04 per 1 000 child population 0-17 y.o. or 77.5 per 100 000 live births females. The percentage of children with TS among all children with dwarfism, that includes in Registry, is 24,8%.

Annually are diagnosed 17-25 new cases of TS.

The average age of diagnosis of new cases – 9,4±4,9 y.o.

Age of primary diagnosis of TS in children with karyotype:

- **45,X – 9,1±5,2 y.o.,**
- **mosaic – 9,5±4,4 y.o.**
- **structural anomalies of chromosome X – 10,4±3,9 y.o.**

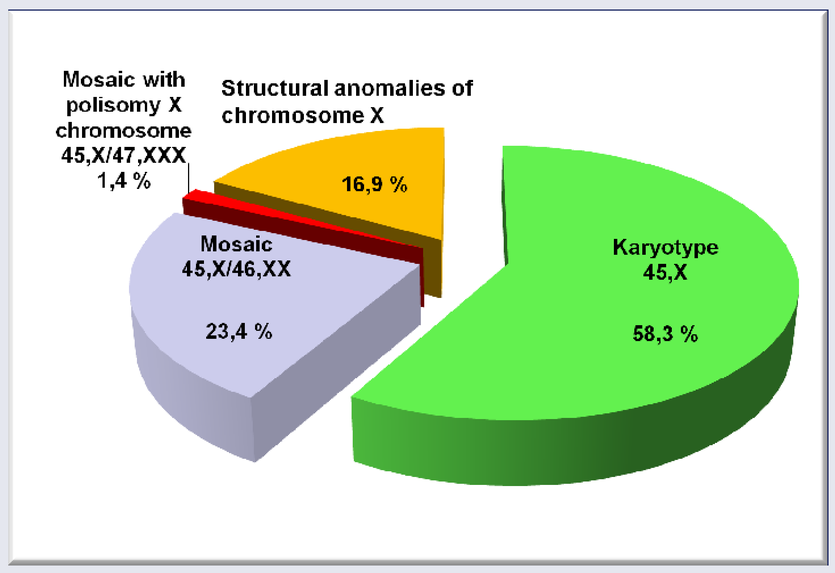

Fig 1. Variants karyotype in children with TS in Ukraine (%)

By the Registry girls with TS:

45,X - 58.3%;

45,X/46,XX – 23.4%;

mosaic with polisomy X (45,X/47,XXX) – 1.4%,

structural anomalies of chromosome X – 16.9%:

46,Xi(Xq) – 4.8%;
45,X/46,Xi(Xq) – 6.3%;
45,X/46,X+mar – 3.1%,
46,X,del(X)(Xq) – 1,9%
45,X/46,X,del – 0.8%

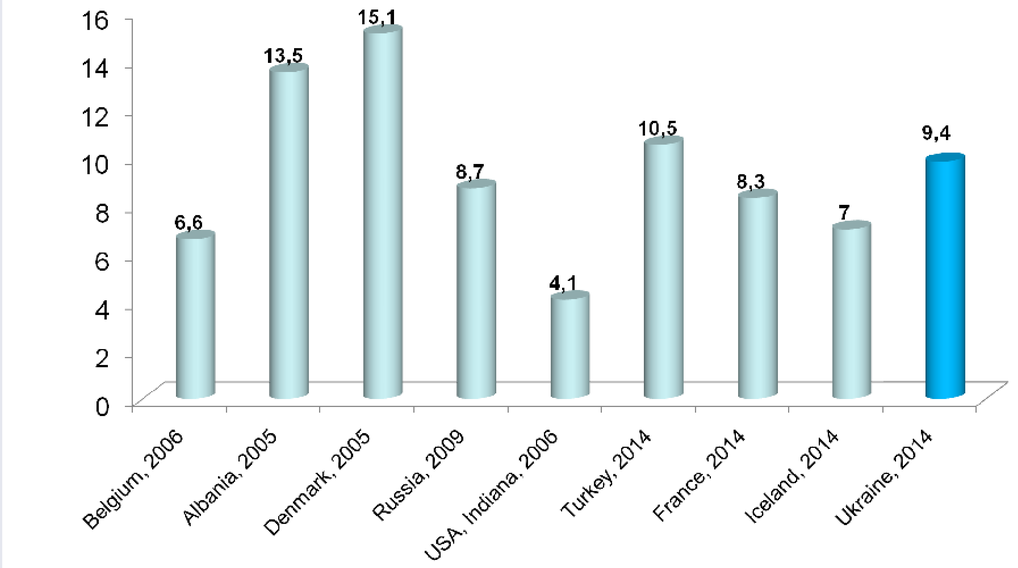

Fig 3. The average age of diagnosis of the TS [2-10]

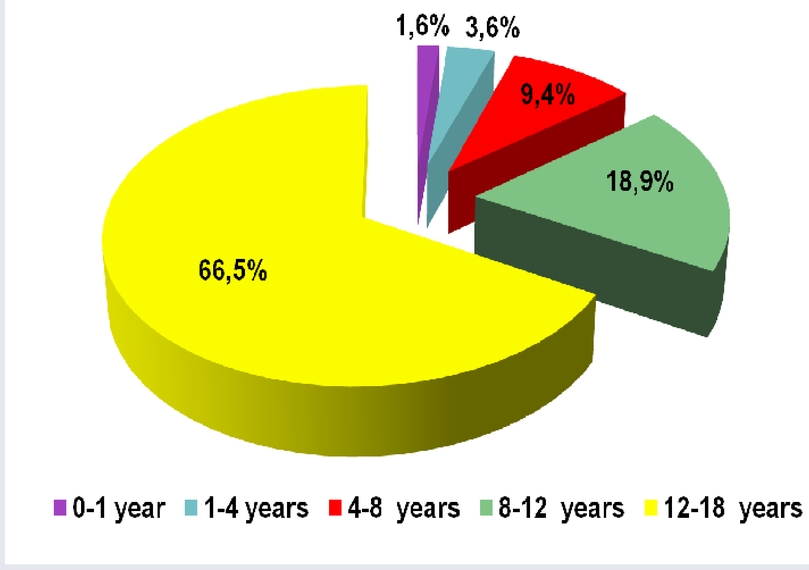

Fig 2. Number of newly diagnostic cases of TS at different age group (%)

The age of first diagnostic:

1 y.o. - 1.6%

1-4 .o. - 3.6%

4-8 y.o. - 9.4%

8-12 y.o. - 18.9%

12-17 y.o. - 66.5%

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Conclusions

In Ukraine the highest incidence of primary diagnosis of TS - at the age after 12 y.o. and in girls with karyotype 45,X.

By Registry the largest proportion of girls with identified TS has a karyotype 45,X.

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Misc 1

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