Trends in the prenatal diagnostics of the Down syndrome in the Czech Republic: Can we find the association with the maternal age?

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http://www.vrozenevady.cz/
Registry: Population based (whole area of the Czech Republic, 100,000 annual births).

Law: The registration is compulsory, required by the Internal Law of Ministry of Health (nr. 14/2001). The database is run by the Institute of Health Information and Statistics of the Czech Republic. Additional data on prenatally diagnosed cases are collected thanks to the country-wide cooperation.

Cases: All cases in TOPs, live births and stillbirths are reported.

Coding: ICD-10 (international), no verbal description.

Sources: Multiple sources, including departments of medical genetics, genetic laboratories, pediatric and neonatology departments, delivery units, ultrasound diagnostics departments etc.

Termination of Pregnancy: Legal, up to the 24th week of gestation.
Down syndrome – All cases in live births and TOPFAs
Down syndrome

Down syndrome – relative number of TOPFAs (%)
Maternal age – Time trends

Proportion of mothers aged 35 years or over
Down syndrome (2014)

Down syndrome cases (all) according to the maternal age

Mothers aged 35 or over: 63%
Mothers under 35 years: 37%
Time of diagnosis

Average week of gestation at diagnosis of Down syndrome
Down syndrome - Diagnostics

Time trends – most common week of Down syndrome diagnosis
Down syndrome - Diagnostics

Time trends – most common week of Down syndrome diagnosis

Time trends – Early vs. Late diagnosis regarding the maternal age

Early diagnosis
Average diagnosis
Late diagnosis

Maternal age
Down syndrome – 2008-2011

Time trends – Early vs. Late diagnosis regarding the maternal age

Maternal age

Early diagnosis
Average diagnosis
Late diagnosis
What is the significance?

<table>
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<th>Years</th>
<th>EvsA</th>
<th>AvsL</th>
<th>EvsL</th>
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<tbody>
<tr>
<td>1998-1999</td>
<td>0.039</td>
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<td>2008-2009</td>
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<td>2010-2011</td>
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</tbody>
</table>

p values computed using nonparametric Kolmogorov-Smirnov test
The role of screening

Changing role of screening programs in prenatal diagnosis

NIPT?
1. The indication criteria for the invasive prenatal diagnosis are changing.
2. The average maternal age is notably increasing.
3. The early prenatal diagnosis (during the first trimester of gravidity) is becoming much more important.
4. There are no significant changes in the timing of the diagnosis among younger and elder mothers.
5. However, this trend was visible some 10 years ago.
6. May we see it in the future again?
We would like to express our thanks to all helpful professionals (geneticists, neonatologists, pediatricians, gynecologists and many others) who are involved in the registration process and provide the data, necessary for the surveillance of the congenital anomalies in the Czech Republic.
Thank you for your attention!

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http://www.vrozenec-vady.cz/