Medical Pedigrees: Typography and Interface

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1. Introduction

A Complex Pedigree

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Our package for pedigree drawing was described in several papers:\(^2\):

1. A \TeX-PSTricks package to draw diagrams as complex as they can get\(^3\).

2. A Perl program to automatically generate \TeX input for 90\% cases\(^4\).


\(^3\)Boris Veytsman and Leila Akhmadeeva, Creating Medical Pedigrees with PSTricks and \LaTeX, 2007.

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New challenges:
How to make things beautiful & user-friendly?

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3Boris Veytsman and Leila Akhmadeeva, Creating Medical Pedigrees with PSTricks and \LaTeX, 2007.

2. History of Pedigrees

This is based on the great work by Resta\textsuperscript{5}. A quotation from there:

A complete pedigree is often a work of great labour, and its finished form is frequently a work of art.

\textit{Karl Pearson, 1912}

Pedigrees vs. genealogical trees: traits instead of individuals. Pedigrees are relatively modern: since the middle of 19 century.

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2.1. Musical Notes for Symbols

Pliny Earle, 1845\textsuperscript{6}:

Incidentally, these are females of the author’s family!
Generations are marked on margins. No marking for individuals.

\textsuperscript{6}Idem
2.2. Tabular System

Galton, 1889\textsuperscript{7}:

\begin{center}
\includegraphics[width=\textwidth]{tabular_system.png}
\end{center}

\textsuperscript{7}Idem
2.3. Mars and Venus

Pearson, 1912\(^8\):

This chart shows inheritance of "commercial and legal ability". The numbering is almost modern. Symbols are quite different. Marriage lines are *under* the symbols. Peculiar marking for twins.

\(^8\)Idem
2.4. German Style

Rüdin, 1910\(^9\):

Proband in center, radial composition.

\(^9\)Idem
2.5. Galton-Pearson Style

Mott, 1910\textsuperscript{10}:

Stylized Mars and Venus for males and females. Note marriage lines.

Was used in some journals up to 1970s!

\textsuperscript{10}Idem
2.6. Davenport-Goddard Style

Goddard, 1911\textsuperscript{11}:

This is almost modern! Note interesting proband notation.

\textsuperscript{11}Idem
3. Modern Pedigrees

A hand drawn pedigree\textsuperscript{12}:

\textsuperscript{12}Robin L. Bennett, \textit{The Practical Guide to the Genetic Family History} (New York; Chichester; Weinheim; Brisbane; Singapore; Toronto: Wiley-Liss, Inc., 1999).
A typical pedigree\textsuperscript{13}:

\begin{figure}[h]
\centering
\includegraphics[width=\textwidth]{pedigree.png}
\caption{Gynealogy of the family living in a small village in the Southern Turkey.}
\end{figure}

Use of pedigree for conveying additional information\textsuperscript{14}:

A complex situation with self-intersections:\(^\text{15}\):

\begin{figure}
\centering
\includegraphics[width=\textwidth]{figure2}
\caption{Pedigree of family 2. Tumours and age of onset are reported. Black symbols indicate CCS-related tumours (CRC, colorectal cancer). Blue symbols indicate other tumours. Numbers on right-top give the current age or age of death. The index patient is indicated by an arrow. In addition, signs of NF1 and mutational status are given.}
\end{figure}

4. Samples of Our Output

Joseph Smith
I:1

I:2
Ann Gold
II:1
Jenny Smith
III:1

Bill Smith
II:2
John Smith
III:2

Jim Brown
I:3

Mary Smith
II:3
Rebecca Smith
Alexander Smith
III:3

Lisa Brown
I:4

Joan Smith

Joan Smith
I:2

Bill Smith
II:2
John Smith
III:2

Jim Brown
I:3

Mary Smith
II:3
Rebecca Smith
Alexander Smith
III:3

Lisa Brown
I:4

Joan Smith
I:2

Bill Smith
II:2
John Smith
III:2

Jim Brown
I:3

Mary Smith
II:3
Rebecca Smith
Alexander Smith
III:3

Lisa Brown
I:4

Joan Smith
I:2

Bill Smith
II:2
John Smith
III:2

Jim Brown
I:3

Mary Smith
II:3
Rebecca Smith
Alexander Smith
III:3

Lisa Brown
I:4
5. Interfaces and User-Friendliness

“Sure, Unix is a user-friendly operating system. It’s just picky with whom it chooses to be friends.”

Anonymous
“Sure, Unix is a user-friendly operating system. It’s just picky with whom it chooses to be friends.”

*Anonymous*
This is not user-friendly\textsuperscript{16}—and is not intended to be!

\textsuperscript{16}At least for some users
This is supposed to be user-friendly—and it is\(^\text{17}\)!

<table>
<thead>
<tr>
<th>Id</th>
<th>Name</th>
<th>Sex</th>
<th>DoB</th>
<th>DoD</th>
<th>Mother</th>
<th>Father</th>
<th>Proband</th>
</tr>
</thead>
<tbody>
<tr>
<td>P</td>
<td>John Smith</td>
<td>male</td>
<td>1970/02/05</td>
<td></td>
<td>M1</td>
<td>F1</td>
<td>yes</td>
</tr>
<tr>
<td>M1</td>
<td>Mary Smith</td>
<td>female</td>
<td>1940/02/05</td>
<td></td>
<td>GM2</td>
<td>GF2</td>
<td></td>
</tr>
<tr>
<td>F1</td>
<td>Bill Smith</td>
<td>male</td>
<td>1938/04/03</td>
<td></td>
<td>GM1</td>
<td>GF1</td>
<td></td>
</tr>
<tr>
<td>GM1</td>
<td>Joan Smith</td>
<td>female</td>
<td>1902/07/01</td>
<td>1975/12/13</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

\(^{17}\)If you like spreadsheets...
How do we change the parameters?
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```perl
# Do we want to print a legend?
#
$printlegend=1;

# Fields to include in the legend. Delete Name for privacy protection.
#
@fieldsforlegend = qw(Name DoB AgeAtDeath Comment);

# Fields to put at the node. Delete Name for privacy protection.
#
@fieldsforchart = qw(Name);
```

This is not user-friendly...
Changes: paper size, fonts, scaling...
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Ad hoc solution: specially designed Makefile. Definitely not for everybody.
6. **Conclusions**

We started a journey to check our typography and our programs.
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We started a journey to check our typography and our programs.
An unexpected result: our typography is good.
But our interfaces needs changes.
References


