Medical Pedigrees: Typography and Interface
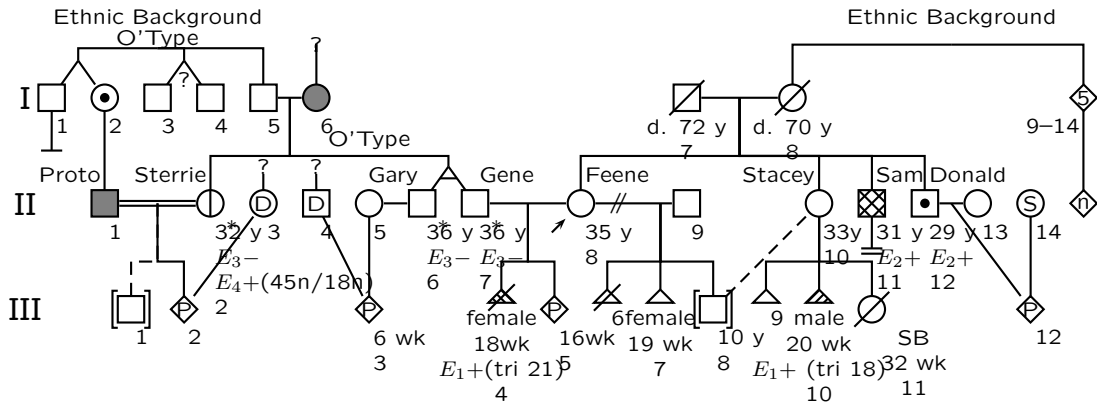
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TUG 2009, Notre Dame, IN, USA

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1. Introduction

A Complex Pedigree[1]

[1] Robin L. Bennett et al., "Recommendations for Standardized Human Pedigree Nomenclature," *Am. J. Hum. Genet.* 56/3 (1995).

Our package for pedigree drawing was described in several papers[2]:

1. A T_EX-PSTricks package to draw diagrams as complex as they can get[3].

2. A Perl program to automatically generate T_EX input for 90% cases[4].

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[2]Boris Veytsman and Leila Akhmadeeva, "Drawing Medical Pedigree Trees with T_EX and PSTricks," *TUGboat* 28/1 (2007); Boris Veytsman and Leila Akhmadeeva, "Medical Pedigrees with T_EX and PSTricks: New Advances and Challenges," *TUGboat* 29/3 (2008)

[3]Boris Veytsman and Leila Akhmadeeva, Creating Medical Pedigrees with PSTricks and LaT_EX, 2007.

[4]Boris Veytsman and Leila Akhmadeeva, A Program For Automatic Pedigree Construction With pst-pdgr. User Manual and Algorithm Description, 2007.

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].

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

[2]Boris Veytsman and Leila Akhmadeeva, "Drawing Medical Pedigree Trees with TeX and PSTricks," *TUGboat* 28/1 (2007); Boris Veytsman and Leila Akhmadeeva, "Medical Pedigrees with TeX and PSTricks: New Advances and Challenges," *TUGboat* 29/3 (2008)

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

[4]Boris Veytsman and Leila Akhmadeeva, A Program For Automatic Pedigree Construction With pst-pdgr. User Manual and Algorithm Description, 2007.

2. History of Pedigrees

This is based on the great work by Resta[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.
>
> *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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[5]Robert G. Resta, "The Crane's Foot: The Rise of the Pedigree in The Human Genetics," *J. Genetic Couns.* 2/4 (1993).

2.1.   Musical Notes for Symbols

Pliny Earle, 1845[6]:

Incidentally, these are females of the author's family!

Generations are marked on margins. No marking for individuals.
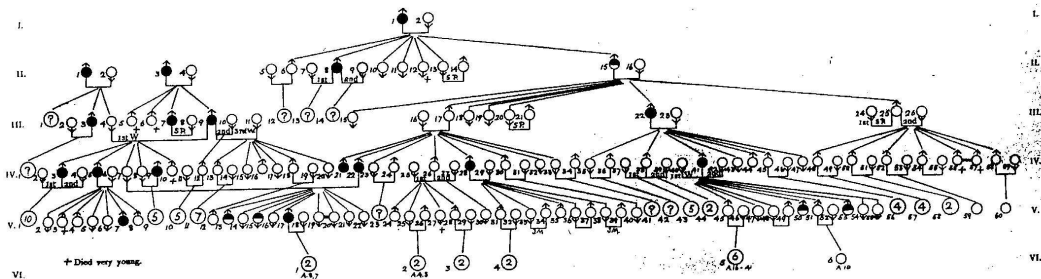
---

[6] *Idem*

2.2. Tabular System

Galton, 1889[7]:

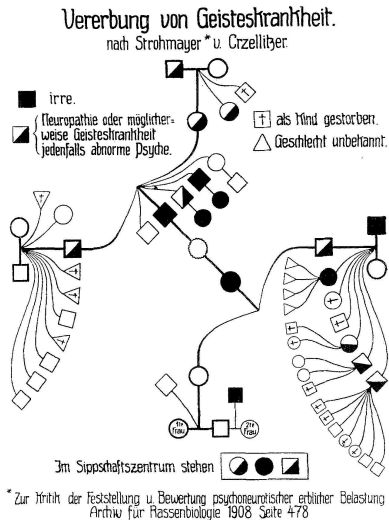[7] *Idem*

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.
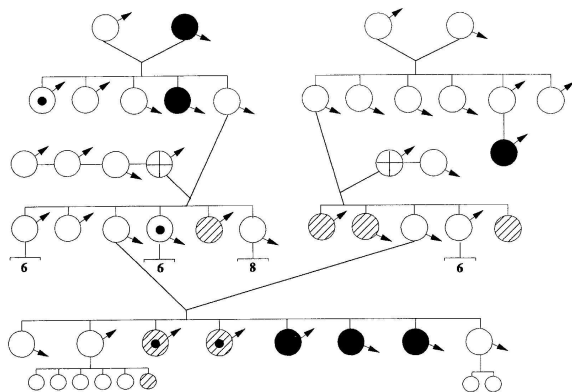
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[8]*Idem*

2.4. German Style

Rüdin, 1910[9]:

Proband in center, radial composition.

---

[9]*Idem*

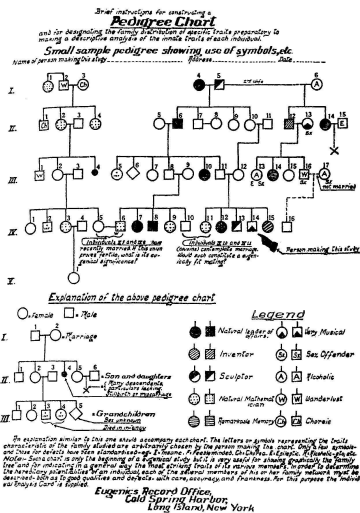## 2.5. Galton-Pearson Style

Mott, 1910[10]:

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

Was used in some journals up to 1970s!

[10]*Idem*

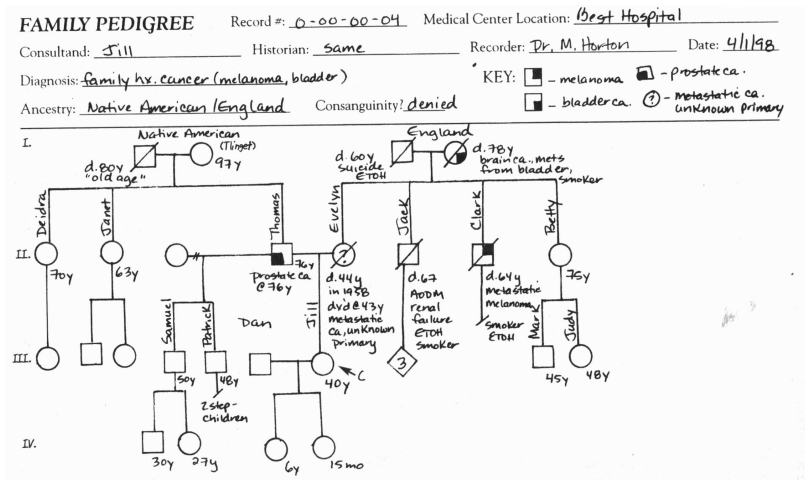## 2.6. Davenport-Goddard Style

Goddard, 1911[11]:

This is almost modern! Note interesting proband notation.

[11]*Idem*

3. Modern Pedigrees

A hand drawn pedigree[12]:

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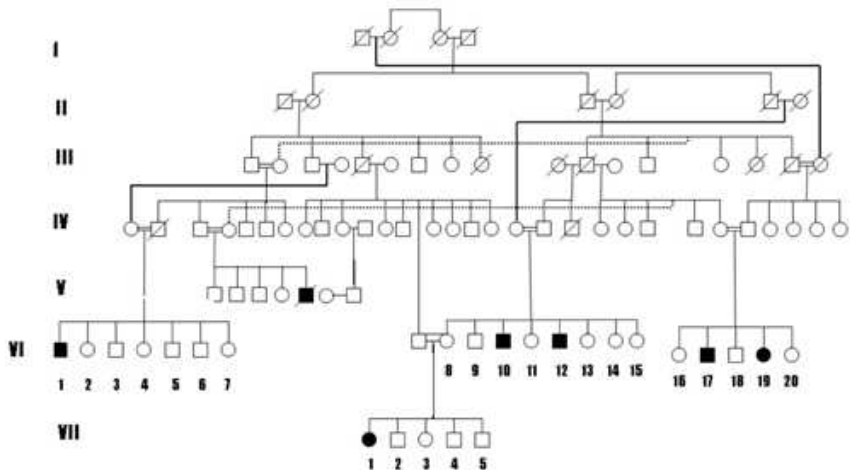
A typical pedigree[13]:

Figure 1. Gynealogy of the family living in a small village in the Southern Turkey.

[13]Üner Tan et al., "Unertan Syndrome: A Case Series Demonstrating Human Devolution," *Int. J. Neurosci.* 118/1 (2008), http://search.ebscohost.com/login.aspx?direct=true&db=pbh&AN=27625823&site=ehost-live.
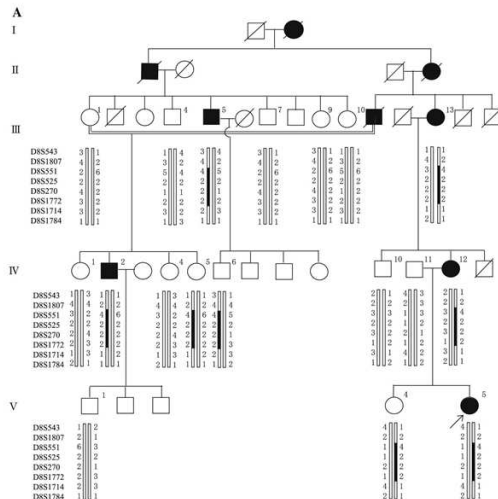
Use of pedigree for conveying additional information[14]:

Fig. 1 Family tree with haplotypes at **a** 8q21.11-q22.2, **b** 12q24.32-qter, and **c** 14q21.1-q23.2 regions. *Closed, open and slash symbols* indicate affected, unaffected, and deceased individuals, respectively. *Double horizontal line* depicts consanguineous marriage, and *short bar* above individual symbols indicates individuals examined clinically. *Thick columns* depict disease-associated haplotypes

[14]Mitsuko Nakashima et al., "Genome-wide linkage analysis and mutation analysis of hereditary congenital blepharoptosis in a Japanese family.," *J. of Hum. Gen.* 53/1 (2008), http://search.ebscohost.com/login.aspx?direct=true&db=a9h&AN=27978113&site=ehost-live.
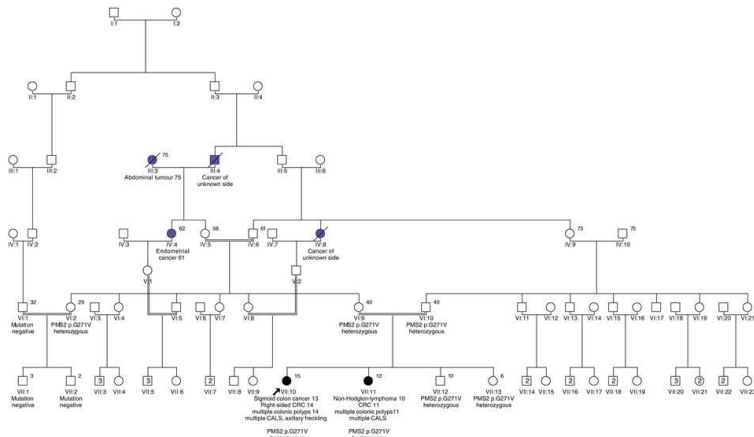
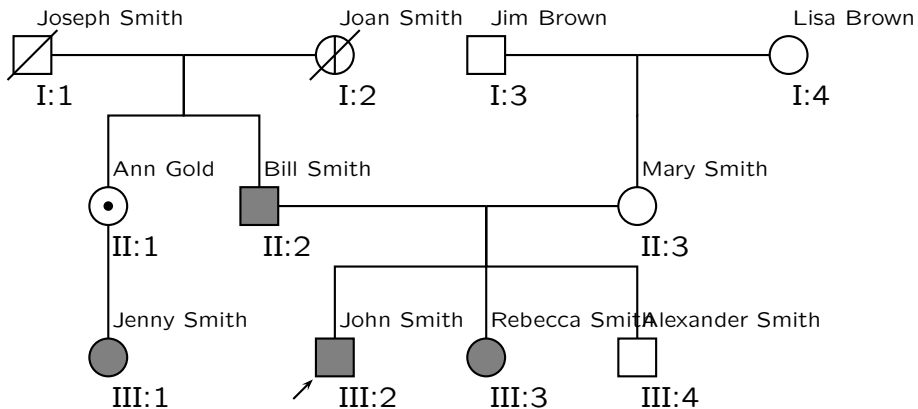A complex situation with self-intersections[15]:

Figure 2 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.

[15]Stefan Krüger et al., "Homozygous PMS2 Germline Mutations in Two Families With Early-Onset Haematological Malignancy, Brain Tumours, HNPCC-Associated Tumours, and Signs of Neurofibromatosis Type 1," *Eur. J. Hum. Gen.* 16/1 (2008), http://search.ebscohost.com/login.aspx?direct=true&db=a9h&AN=27879615&site=ehost-live.

4.   Samples of Our Output

Jane
I:1

Nancy
II:1

Samantha
II:2

Ann
II:3

John
II:4

Jane
III:1

Paul
III:2

Mary
III:3

Laura
IV:1

Jack
IV:2
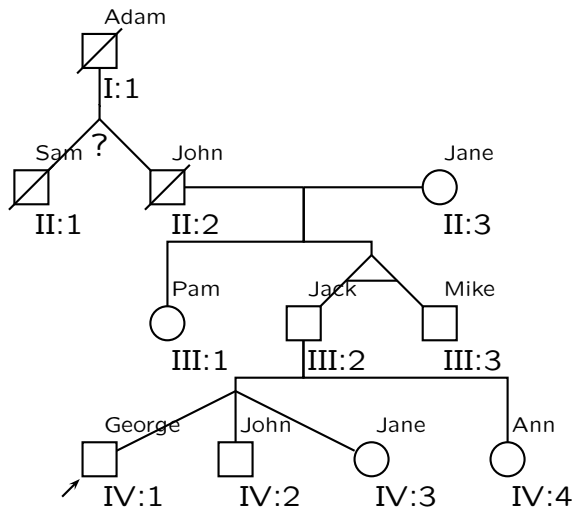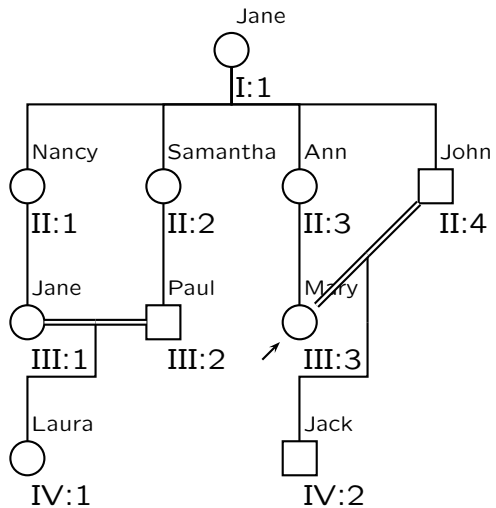
5.   Interfaces and User-Friendliness

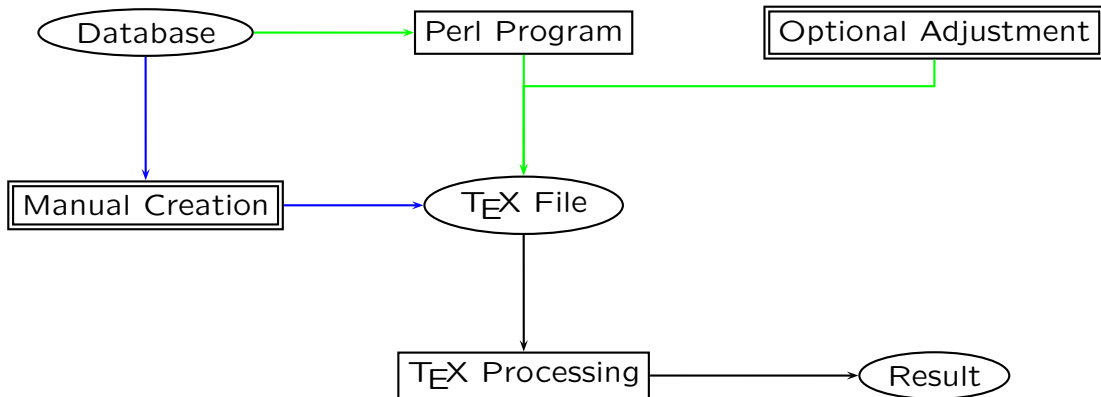   "Sure, Unix is a user-friendly operating system. It's just picky with whom it chooses to be friends."
Anonymous

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

5.1. T_EX Part

```
\rput(-6, 2){\pstPerson[male, condition=normal, belowtext={\small
    I:1}, deceased, abovetext={\scriptsize Joseph Smith}]{GF1}}
\rput(0, 2){\pstPerson[male, condition=normal, belowtext={\small I:3},
 abovetext={\scriptsize Jim Brown}]{GF2}}
\rput(4, 2){\pstPerson[female, condition=normal, belowtext={\small
    I:4}, abovetext={\scriptsize Lisa Brown}]{GM2}}
\rput(2, 2){\pnode{GF2_m_GM2}}
\rput(-4, 2){\pnode{GF1_m_GM1}}
\rput(-2, 2){\pstPerson[female, asymptomatic, belowtext={\small I:2},
 deceased, abovetext={\scriptsize Joan Smith}]{GM1}}
\rput(0, 0){\pnode{F1_m_M1}}
...
```

This is not user-friendly[16]—and is not intended to be!

5.2. Perl Part

This is supposed to be user-friendly—and it is[17]!

```
Id |Name        |Sex    |DoB       | DoD       |Mother|Father|Proband
P  |John Smith  |male   |1970/02/05|           |M1    |F1    | yes
M1 |Mary Smith  |female |1940/02/05|           |GM2   |GF2   |
F1 |Bill Smith  |male   |1938/04/03|           |GM1   | GF1  |
GM1|Joan Smith  |female |1902/07/01|1975/12/13||      |      |
...
```

---

[17]If you like spreadsheets. . .

How do we change the parameters?

How do we change the parameters?

```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: specialliy designed Makefile. Definitely not for everybody.

6. Conclusions

We started a journey to check our typography and our programs.

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An unexpected result: our typography is good. . .

But our interfaces needs changes. . .

References

Bennett, Robin L. *The Practical Guide to the Genetic Family History*. New York; Chichester; Weinheim; Brisbane; Singapore; Toronto: Wiley-Liss, Inc., 1999.

Bennett, Robin L. et al. "Recommendations for Standardized Human Pedigree Nomenclature." *Am. J. Hum. Genet.* 56/3 (1995): 745–752.

Krüger, Stefan et al. "Homozygous PMS2 Germline Mutations in Two Families With Early-Onset Haematological Malignancy, Brain Tumours, HNPCC-Associated Tumours, and Signs of Neurofibromatosis Type 1." *Eur. J. Hum. Gen.* 16/1 (2008): 62–72. http://search.ebscohost.com/login.aspx?direct=true&db=a9h&AN=27879615&site=ehost-live.

Nakashima, Mitsuko et al. "Genome-wide linkage analysis and mutation analysis of hereditary congenital blepharoptosis in a Japanese family.." *J. of Hum. Gen.* 53/1 (2008): 34–41. http://search.ebscohost.com/login.aspx?direct=true&db=a9h&AN=27978113&site=ehost-live.

Resta, Robert G. "The Crane's Foot: The Rise of the Pedigree in The Human Genetics." *J. Genetic Couns.* 2/4 (1993): 235–260.

Tan, Üner et al. "Unertan Syndrome: A Case Series Demonstrating Human Devolution." *Int. J. Neurosci.* 118/1 (2008): 1–25. http://search.ebscohost.com/login.aspx?direct=true&db=pbh&AN=27625823&site=ehost-live.

Veytsman, Boris and Leila Akhmadeeva. Creating Medical Pedigrees with PSTricks and LaTeX. July 2007. http://ctan.tug.org/tex-archive/graphics/pstricks/contrib/pedigree/pst-pdgr.

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———. A Program For Automatic Pedigree Construction With pst-pdgr. User Manual and Algorithm Description. July 2007. http://ctan.tug.org/tex-archive/graphics/pstricks/contrib/pedigree/pedigree-perl.

———. "Medical Pedigrees with TeX and PSTricks: New Advances and Challenges." *TUGboat* 29/3 (2008): 484. http://www.tug.org/TUGboat/Articles/tb29-3/tb93abstracts.pdf.