Genetics and population analysis

pedigreejs: a web-based graphical pedigree editor

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Abstract

Motivation: The collection, management and visualization of clinical pedigree (family history) data is a core activity in clinical genetics centres. However, clinical pedigree datasets can be difficult to manage, as they are time consuming to capture, and can be difficult to build, manipulate and visualize graphically. Several standalone graphical pedigree editors and drawing applications exist but there are no freely available lightweight graphical pedigree editors that can be easily configured and incorporated into web applications.

Results: We developed ‘pedigreejs’, an interactive graphical pedigree editor written in JavaScript, which uses standard pedigree nomenclature. Pedigreejs provides an easily configurable, extensible and lightweight pedigree editor. It makes use of an open-source Javascript library to define a hierarchical layout and to produce images in scalable vector graphics (SVG) format that can be viewed and edited in web browsers.

Availability and implementation: The software is freely available under GPL licence (https://ccge-boadicea.github.io/pedigreejs/).

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Supplementary information: Supplementary data are available at Bioinformatics online.

1 Introduction

The collection, management and visualization of pedigree (family history) data is a core activity in clinical settings, such as genetic centres and primary care, where healthcare professionals manage patients at risk of disease. Pedigree data are used to identify individuals at risk of familial disease and to inform their clinical management, including referral of individuals at elevated risk for mutation screening and/or recommending enhanced screening. For example, generating a pedigree is a requirement for calculating BRCA1 and BRCA2 mutation carrier probabilities and breast cancer risks in the BOADICEA (Lee et al., 2014) and IBIS (Tyrer et al., 2004) programs recommended in the National Institute for Health and Care Excellence guidelines (NICE, 2013, https://www.nice.org.uk/guidance/cg164).

Clinical pedigree datasets can be difficult to manage for several reasons: (i) they are supplied by humans not machines, and are therefore time consuming to capture in digital form; (ii) they include both family structure (which can be difficult to visualize in the presence of complex structure and still remain clear and comprehensible) and separate information for each family member (which must be linked); (iii) they often require updating over time; (iv) they can include data errors (e.g. broken family structures) and internal...
inconsistencies; and (v) they must be stored securely in order to fulfill data privacy requirements.

Many existing software applications capture and display pedigree data. Some open-source tools (e.g. Kinship2: Sinnwell et al., 2014) generate a vector image file from an input data file, whereas others (e.g. Pelican: Dudbridge et al., 2004 and Phenotips: Gardea et al., 2013) provide a graphical pedigree editor. There are also commercial subscription pedigree drawing services (http://www.progenygenetics.com, http://pedigreedraw.com, http://www.pedigreexp.com). However, after evaluating available tools (see Supplementary Data) we found no freely available lightweight pedigree drawing tools (components implemented solely in JavaScript) that could be easily incorporated into other web applications, and configured to provide a means of capturing and visualising pedigree data. Pedigrees has been developed to fulfill these requirements as part of the ongoing BOADICEA project (http://ccge.medschl.cam.ac.uk/boadicea, Lee et al., 2014) (Fig. 1).

![Image of a pedigree extending to second degree built using pedigrees.](image)

**Fig. 1.** Screenshot showing a pedigree extending to second degree built using pedigrees. The pedigree is annotated in the conventional manner: the proband is identified with an arrow; family members who have developed cancer are shaded; and unaffected family members are unshaded. Users can add or delete family members, and change their properties using widgets, which appear when the individual is selected. Buttons directly above the pedigree are used to ‘undo’ and ‘redo’ actions, ‘reset’ the initial structure, and switch to ‘fullscreen’ mode. The buttons at the top are used to ‘Load’ and ‘Save’ pedigree data files, to print the pedigree, and to export it as a SVG/PNG image solely in JavaScript) and uses D3.js to assist in the rendering and does not send requests to a backend server. Creating and deleting individuals is made easy by clicking on widgets that appear on mouserover of a family member. ‘Undo’, ‘redo’ and ‘reset’ options and the ‘fullscreen’ option add to this being a user friendly editing tool.

Future developments will include further support for other commonly used pedigree data file formats (e.g. Madeline, Progeny). Pedigrees will also be updated to support more complex family structures including consanguineous partners beyond those at the same level in the tree such as cousins. Pedigrees will be used in future releases of the BOADICEA Web Application.

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References


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