



CLINICAL GENETICS SOCIETY
Clinical Governance Subcommittee

Paper 2 (Version A, 5/7/2001) Pedigree Drawing.

Replies to: Dr Helen Hughes, Institute of Medical Genetics, Cardiff CF14 4XW

Tel: 01745 534447 Fax: 01745 534431 e-mail: Helen.Hughes@cd-tr.wales.nhs.uk

Guidelines for Pedigree Drawing

An accurate family pedigree is an important part of the clinical genetics record. The following guidelines are intended to help minimise the risk of errors being made in recording pedigrees or in the interpretation of pedigree data.

1. Using a pre-printed pedigree sheet helps with laying out the pedigree. This can be printed on coloured paper for ease of location in the notes.
2. Using standard pedigree symbols such as those recommended by The Pedigree Standardization Task Force (*Am J Hum Genet* 1995;56:745-52) helps avoid misinterpretation of the pedigree data.
3. Record on the pedigree:
 - a unique family identifier such as the Family Pedigree number.
 - name of person providing the family history information.
 - name and title of person recording the information.
 - date pedigree drawn.
4. For significant additions or amendments to the pedigree, record:
 - name of person providing the additional family history information.
 - name and title of person recording the information.
 - date additions or amendments made.
5. Use ink or biro to draw the pedigree.
6. Consider using a template to keep the pedigree tidy.
7. The usual convention is to place the male partner on the left.
8. The usual convention is to place siblings in birth order with the firstborn on the left.
9. Record the names of family members where appropriate.
Reasons for recording names of relatives include (a) needed for chasing up diagnostic information on affected relative (b) likely to be offering them screening (c) likely they may seek counselling. It is particularly useful to record names of the affected members of the family, even if deceased. Suggest as a minimum taking the names and dates of birth of all first-degree relatives of index case.
10. Record maiden names of women where appropriate.

11. It is good practice to check the spelling of names including first names because there can be unusual spellings of even common first names.
12. Record dates of birth rather than ages. When reviewing the family at a later date, current ages can be more reliably determined since no assumption has to be made about when the pedigree was drawn.
13. If approximate age is all that is known, record as approximate year of birth.
e.g. *b. ~1996* or *b. 1996ish*
14. Where appropriate, ask about the state of health of relatives and if deceased record their cause of death and age at death.
15. Use appropriate symbols for disease status and **provide a key to disease symbol(s) used**. This is particularly important if there are several diagnoses in the family denoted by different symbols, as in the case of familial cancer syndromes.
16. Typical pedigrees will cover three generations. However, the extent and level of detail of a pedigree depends on the nature of the disorder.
e.g. For a couple who have had a pregnancy affected by trisomy 21, it would probably be sufficient to stop at their first degree relatives. In a colon cancer family close to fulfilling Amsterdam criteria, will want all the available information about the family history. For an X-linked lethal condition, need a detailed maternal family history, particularly if distant affected males would influence carrier status.
17. Ask specifically about 'any other branches of the family that haven't been included.'
18. Ask if have children from other relationships.
19. Enquire specifically about children who died, stillbirths, and miscarriages.
20. Ask specifically about family history of miscarriages, stillbirths, childhood deaths, and learning disability.
21. Ask specifically about consanguinity.
22. Indicate proband(s) with an arrow labelled with letter P.
The proband or index case is the affected individual through whom the family is ascertained. There may be more than one.
23. Indicate consultand(s) with an unlabelled arrow.
The consultand is the person seeking advice, who may not be affected.
24. Additional information that can be recorded on the pedigree includes:
 - brief summary information about key clinical findings, but avoid cluttering the pedigree.
 - results of key investigations.
 - results of genetic testing, but bear in mind issues of confidentiality (e.g. HD predictive testing).
 - storage of DNA samples.If abbreviations or symbols are used, make sure these are defined in the key.

Additional pedigree information		
Name, DoB and/or location in pedigree	Clinical or other information	
		Male, female, unknown sex
		Clinically affected (specify condition in key)
		Clinically affected, several diagnoses Some examples (define symbols in key)
		Multiple siblings (in this case 5) If number not known, put <i>n</i>
		Deceased individual (with age died)
		Proband
		Consultand
		Carrier who will never manifest disease (half shaded symbol is not recommended)
		Presymptomatic carrier (may manifest disease later)
		Twins (MZ, DZ and uncertain zygosity)
		Ongoing pregnancy (consider using pencil) <i>LMP 2-12-99 EDD Mar 2000</i>
		Miscarriage (sex specified) <i>male</i>
		Miscarriage, affected (with gestation) <i>16 wk</i>
		Termination, unaffected and affected
		Stillbirth (with gestation) <i>SB 30 wk</i>
		No offspring (if want to specifically indicate this)
		No offspring due to infertility
		Partners now separated
		Consanguinity
		Adopted child (if want to include them)
		Ref: <i>Am J Hum Genet</i> 1995;56:745-52 JRWY Mar 2000

Hospital/Dept: STRICTLY CONFIDENTIAL	Consanguinity yes / no	Family No:
I		
II		
III		
IV		
V		
KEY	Source: Date: Drawn by:	Source: Date: Updated by: