



CLINICAL GENETICS SOCIETY  
Clinical Governance Subcommittee

## Paper 1 (Version A, 5/7/2001) Follow-up and recall in

### clinical genetic practice

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1 Clinical geneticists chose to follow those affected individuals where they believe they may  
2 significantly influence diagnosis and management. With the consent of the original  
3 consultant, follow-up in clinical genetic practice may also include extended family members  
4 who may be at risk (or who have concerns about their risk) of developing a disorder or of  
5 transmitting it to their children. Plan for follow-up needs to be explicitly recorded in the  
6 patient chart or letter so that confusion regarding responsibility between various medical  
7 professionals and the family is avoided. Families who have been discharged from follow-up  
8 can be recalled if further information relevant to genetic management becomes available.

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- 10 1. To complete initial assessment e.g. disclosure of diagnosis or of investigation results,  
11 further counselling.
- 12
- 13 2. If no diagnosis is made, selected individual may be seen for re-evaluation given  
14 evolution of phenotype, availability of new investigations, and recognition of new  
15 syndromes.
- 16
- 17 3. Further support and counselling e.g. if poor understanding or if there are other issues  
18 which may be interfering with the communication process.
- 19
- 20 4. Planned staged management as in predictive testing counselling protocols (e.g.  
21 Huntington disease).
- 22
- 23 5. Follow-up initiated by family e.g. further questions, pre-natal testing.
- 24
- 25 6. Active medical management in some specialized circumstances – this activity needs  
26 to be distinguished from the more usual role of providing a clinical genetic service  
27
- 28 7. With the family's consent, follow-up can be arranged in order to facilitate the  
29 geneticist's own learning and for specific research purposes.
- 30
- 31 8. Health surveillance ([see below - a](#))
- 32
- 33 9. Active recall of discharged families because of availability of new genetic knowledge  
34 ([see below - b](#))
- 35
- 36 10. Inclusion on a genetic register ([see below - c](#))
- 37
- 38 11. Contact with relative at risk ([see below - d](#))
- 39
- 40 (a) *Health surveillance* ([8 above](#)) Clinical geneticists strongly support the concept of  
41 health surveillance and some, depending on clinical expertise and interest, may decide

42 to undertake a coordinating role. The decision to participate in aspects of health  
43 surveillance of patients with multi-system genetic disorders usually occurs in  
44 conjunction with other medical specialists. This activity needs to be coordinated  
45 through the establishment of appropriately-funded and supported specialty clinics e.g.  
46 Marfan, muscular dystrophy. ([Back](#))  
47

48 (b) *Is there a duty to recall discharged families? (9 above)* In principle clinical  
49 geneticists feel an obligation to recall discharged families for further counselling  
50 should new information or genetic tests become available through advancing  
51 knowledge. However, in practice, this type of recall or follow-up is becoming  
52 impossible to implement as the speed of availability of new information about the  
53 vast number of genetic disorders has now outstripped the resources available to  
54 implement practice in most, if not all, clinical genetic centres. Consequently, in the  
55 absence of an unequivocal legal opinion re duty to recall in these circumstances, and  
56 the lack of an imperative amongst commissioners of genetic service to prioritise this  
57 issue with increased resources, clinical geneticists and families together must share  
58 the perceived obligation to re-contact. Currently, it appears that practice has evolved  
59 so that most clinical geneticists indicate to families that the information given to them  
60 is the most accurate available at the time of genetic counselling but that new scientific  
61 information becoming available in the future could alter understanding of the disease  
62 process and odds of recurrence amongst family members. Families and referring  
63 physicians therefore should be made aware of possible future changes and encouraged  
64 to re-contact the genetic service if reproductive decision is postponed or if they have  
65 added questions in the future. ([Back](#))  
66

67 (c) *Follow-up as part of inclusion on a genetic register (10 above)* Genetic registers were  
68 first established in the UK in the late 60's in order to achieve complete ascertainment  
69 of particular diseases and, by proactive contact, to have a role in their prevention. In  
70 centres where registers currently exist, they have developed to be a tool that facilitates  
71 review and re-contact particularly focused on the time at which individuals at  
72 potential risk could benefit most from information related to family planning.  
73 Occasionally there is a blurring between the function of a regular clinical chart  
74 review, a follow-up system, organized surveillance and the use of a genetic register. It  
75 is recognized that where registers exist, they must be appropriately resourced so that  
76 data accuracy can be maintained by regular family contact, consent for inclusion and  
77 confidentiality ensured and the process of re-contact made explicit. Decisions about  
78 the financial support of registers currently in use and the addition of new disorders  
79 must be taken in conjunction with commissioners of genetic services. ([Back](#))  
80

80 (d) *Duty to contact relatives at risk (11 above)* In contrast to other tertiary care  
81 physicians, clinical geneticists feel that they have a responsibility to offer services to  
82 extended family members who may be at risk of developing a disorder or of  
83 transmitting it to their children. This responsibility is reflected in the tendency  
84 amongst geneticists to think in terms of “families” rather than “patients”. Contact  
85 with relatives usually is made only with the consent and co-operation of the index  
86 patient / family. In the vast majority of instances, the index patient / family will  
87 undertake the responsibility of informing relatives with whom they are in contact of  
88 their potential increased risk so that they can themselves initiate contact with the  
89 genetic service in their locality for further assessment of their genetic status. Often  
90 letters, which outline the genetic information, are provided to the index patient /  
91 family to pass on to their relatives. Therefore, in most circumstances, the  
92 responsibility to offer genetic information to relatives at risk is shared between the  
93 geneticist and the index family. However, difficulties occasionally arise if the index  
94 patient / family either actively or passively refuses to inform their relatives of their  
95 potential risk of developing and / or transmitting a specific genetic disorder. In many  
96 of these instances the difficulty can be resolved if the situation is handled with  
97 patience and understanding on the part of the counselling geneticist – often reluctance  
98 to contact is based on anxiety about causing worry and distress amongst members of  
99 the extended family or because of perceived inability on the part of the consultant  
100 about how to inform their relatives of potentially distressing information.  
101 What then is the obligation of clinical geneticists when they are left in possession of  
102 information which may have a bearing on the medical and genetic well being of  
103 relatives and where permission to contact has been denied? This issue previously has  
104 been addressed in detail by the Nuffield Council on Bioethics in the 1993 report on  
105 genetic screening. In the Council’s opinion the individual’s desire for confidentiality  
106 may be overridden but the decision can only be made case by case. Whilst litigation  
107 in the USA has been successful against physicians who have not informed relatives of  
108 their genetic risk, there is no precedent for this in UK case law. In current practice,  
109 clinical geneticists and counsellors strongly advise the index patient about risks to the  
110 wider family and this discussion should be recorded in the patient’s notes. At present,  
111 there is no legal duty to follow-up other relatives at risk. ([Back](#))

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114 *References:*

- 115  
116 Role of the clinical geneticist – report from the CGS Council to the Joint Committee on Genetic Services (2000)  
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118 McAbee GN and Sherman J. Physicians duty to warn third parties about the risk of genetic disease. Pediatrics  
119 102:140-142, (1998).  
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