

# InfoFact

# Feb 2002

## Familial MND/ALS

### Introduction

FOLLOWING diagnosis of a relative with MND/ALS, other family members may express understandable anxiety and ask: “Will I get it too?”

This INFOFACT leaflet from the Scottish Motor Neurone Disease Association attempts to assist families and individuals affected by MND/ALS – and health care professionals – with some answers to that question.

- Most cases of MND/ALS – between 90-95% - are what is known as “sporadic MND/ALS”. The term “sporadic MND/ALS” means that the disease has occurred in isolation and no other family member is known to have or have had MND/ALS.
- Medical scientists suggest that “familial MND/ALS” – often abbreviated to FMND/FALS – accounts for only 5-10% of all cases.
- Familial MND/ALS is defined as MND/ALS affecting at least two members of one family in the preceding or succeeding generation – or as MND/ALS in which a genetic abnormality such as mutation of a gene known as SOD 1 is identified in an individual even when his or her family history is not available.
- Medical scientists refer to FMND/FALS as “rare”. At the time of production of this infoFACT leaflet (March 1999) there appears to be agreement among medical scientists that for all practical purposes FMND/FALS is clinically indistinguishable from much more common sporadic MND/ALS. In both forms of MND/ALS, progression of the disease varies from individual to individual.

### IS THERE A GENETIC COMPONENT IN FMND/FALS?

- Medical scientists have identified the SOD 1 gene as a “candidate gene” in FMND/FALS. This means it may only be a **possible** factor in FMND/FALS. But it has also been shown that perhaps only 10-20% of families in which FMND/FALS has been identified have members with one of the many possible mutations of the SOD 1 gene.
- Accordingly, most people diagnosed as having FMND/FALS have no currently known “genetic markers”. As with the much more common sporadic MND/ALS, the causes of the disease remain unknown – although there is increasing research worldwide on the subject.

## WHAT IS THE POSSIBLE ROLE OF SOD 1 GENES IN MND/ALS?

- Medical scientists suggest that mutated SOD 1 genes may impair removal from motor neurone cells of so-called “free radicals”.
- Free radicals are molecules, which are normally “mopped up” within cells by substances known as “antioxidants”. Free radicals are damaging to DNA, protein and fat in cell membranes and are implicated in a wide range of diseases.

## WHERE CAN I FIND OUT MORE ABOUT FMND/FALS?

- For people who have concerns about FMND/FALS, genetic counselling is available throughout Scotland. Some travel may be required, although, increasingly, NHS genetic counsellors are holding “peripheral clinics” in local communities.
- NHS genetic counselling is confidential.
- In the first instance, advice on FMND/FALS or genetic counselling should be obtained from general practitioners. Scotland’s network of MND/ALS care advisers/clinical specialists can also provide information on genetic counselling or FMND/FALS.
- MND Care Advisers can be contacted:

**If you live in Greater Glasgow, Renfrewshire, Inverclyde, Dumbartonshire, Lanarkshire, Dumfries & Galloway, Ayrshire & Arran, Argyll & Bute, Western Isles, Highlands please contact:**

MND Clinical Specialist  
First Floor- Block B, Southern General Hospital  
South University Hospital NHS Trust  
1345 Govan Road, Glasgow, G51 4TF  
Telephone: 0141 201 2380 or 0141 201 2381  
[ferguson@mndscot.sol.co.uk](mailto:ferguson@mndscot.sol.co.uk)

**If you live in Aberdeen, Aberdeenshire, Highland, Moray, Orkney, Shetland, Western Isles, please contact:**

MND Clinical Specialist  
Aberdeen Royal Infirmary, Cornhill Road  
Aberdeen, AB25 2ZD  
Telephone: 01224 5543  
[dianne.fraser@arh.grampian.scot.nhs.uk](mailto:dianne.fraser@arh.grampian.scot.nhs.uk)

**If you live in Angus, Borders, Fife, Midlothian, Edinburgh, Falkirk, Perth and Kinross, please contact:**

MND Clinical Specialist  
Dept. Clinical Neurosciences, Western General Hospital  
Crewe Road, Edinburgh, EH4 2XU  
Telephone: 0131 315 2203, [newton@smnda.sol.co.uk](mailto:newton@smnda.sol.co.uk)

**If you live in Tayside and Dundee, please contact:**

MND Clinical Specialist  
Neurology Department, Level 6/South Block  
Ninewells Hospital, Dundee, DD1 9SY  
Telephone: 01382 425726, [shuna.colville@tuht.scot.nhs.uk](mailto:shuna.colville@tuht.scot.nhs.uk)

The Scottish MND Association maintains a professionally administered MND/ALS Library and Information Centre at its head office: Contact Sandra Wilson, Information Officer, Scottish MND Association, 76 Firhill Road, Glasgow, G20 7BA.  
Tel: 0141-945-1077, Fax: 0141-945-2578,  
Email: [info@scotmnd.sol.co.uk](mailto:info@scotmnd.sol.co.uk) Web: [www.scotmnd.org.uk](http://www.scotmnd.org.uk)

The information in this Infocart leaflet is believed to be accurate at the time of going to press. However, the Scottish MND Association cannot give detailed medical advice – and this Infocart leaflet should be regarded only as general background information.