AUTOSOMAL RECESSIVE INHERITANCE – Traditional Patterns of Inheritance 1

FACT SHEET 8

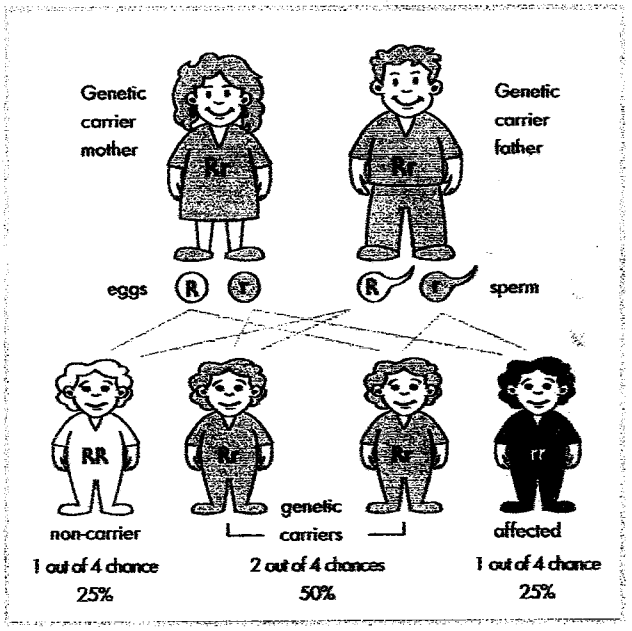

Figure 8.1: Autosomal recessive inheritance when both parents are unaffected genetic carriers for the condition. The faulty copy of the gene containing a recessive mutation is represented by 'r'; the working copy of the gene by 'R'.

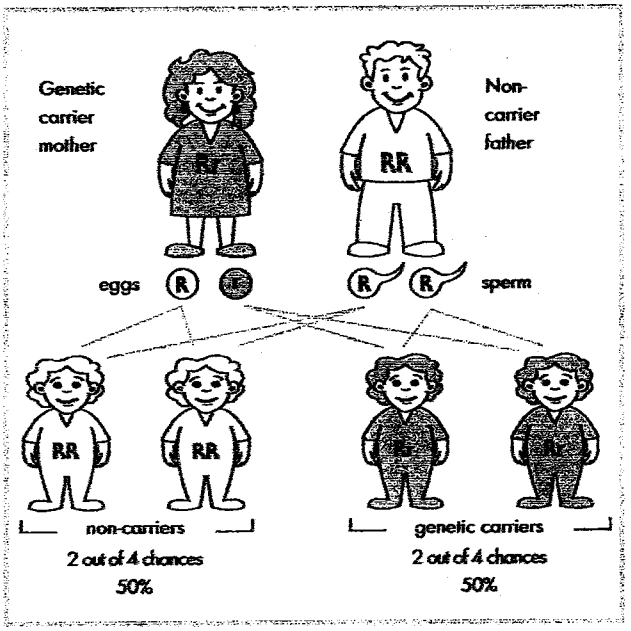

Figure 8.2: Autosomal recessive inheritance when only one of the parents is an unaffected genetic carrier for the condition. The faulty copy of the gene containing a recessive mutation is represented by 'r'; the working copy of the gene by 'R'.

AUTOSOMAL DOMINANT INHERITANCE – Traditional patterns of inheritance 2

FACT SHEET 9

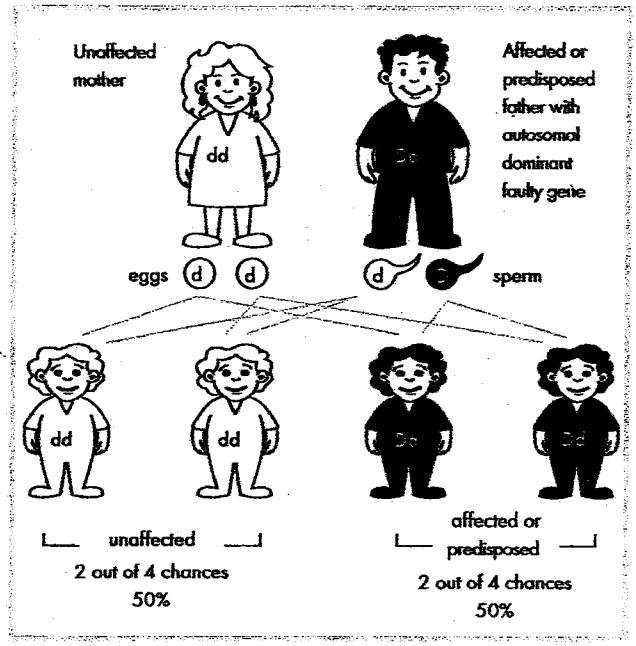

Figure 9.1: Autosomal dominant inheritance when one parent carries the autosomal dominant faulty gene copy. The autosomal dominant faulty gene copy is represented by 'D'; the working copy of the gene by 'd'.

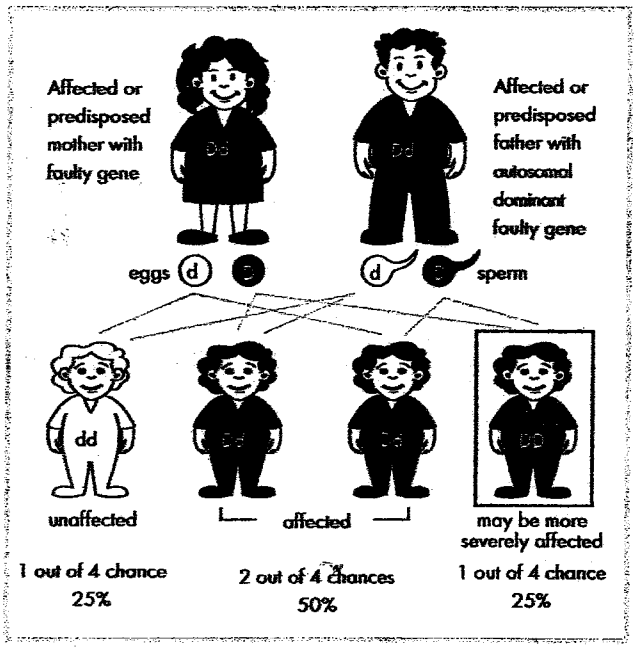

Figure 9.2: Autosomal dominant inheritance when both parents carry the autosomal dominant faulty gene copy. The autosomal dominant faulty gene copy is represented by 'D'; the working copy of the gene by 'd'.

AUTOSOMAL RECESSIVE

Dor Yeshorim Programme

Gene Frequency

Tay Sachs	1:30
Cystic Fibrosis	1:26
Familial Dyautonomia	1:30
Glycogen Storage	1:90
Mucolipidosis [Type IV]	1:90
Niemann-Pick	1:90
Bloom Syndrome	1:100
Gaucher's Disease	1:15

AUTOSOMAL DOMINANT

BRCA 1
BRCA 2 2.5% Ashkenazi Jews

Risk Estimates

High risk families [BRCA 1 or BRCA 2 positive]

Breast cancer 85%
Ovarian cancer 60%
Prostate cancer 25%
Male breast cancer 6%

Screened population [BRCA 1 or BRCA 2 positive]

Breast 55%
Ovarian 16%
Prostate 16%

BRCA 1 or BRCA 2 gene frequency

Ashkenazi women with ovarian cancer 30%
High risk breast cancer gene families 44%

France must say how genetic data are passed to patients' relatives

Paul Benkimoun PARIS

France's Biomedicine Agency has called for the government to draw up guidance on giving genetic information to relatives after a patient has been given a diagnosis of a serious genetic abnormality.

The regulatory agency, which was set up under the 2004 Bioethics Act, was presenting evidence to an inquiry to review the working of the act. The inquiry was established at the request of the French health minister, Roselyne Bachelot-Narquin.

Although the agency concluded that overall the act was working well, it expressed regret that the government had not yet drawn up practical guidance to implement one section of it relating to the question of how information can be passed to relatives of someone with a genetic disorder without a doctor breaching the patient's confidentiality.

French health minister, Roselyne Bachelot-Narquin

The act says that when a severe genetic abnormality is diagnosed, the doctor must give the patient a written summary of the risks to his or her relatives, so that the patient can pass on this information, if he or she wishes, thereby enabling the relatives to seek help and advice.

It specifies how patients can pass on this information if they don't want to tell their relatives directly. It lays down a specially devised "medical information procedure," whereby patients give their doctor the names of relatives and their whereabouts. The doctor then passes these details on to the Biomedicine Agency, which uses another doctor to tell the relatives that it has information that may concern them.

The law also lays down that patients can't be sued for not having passed on the information about their abnormality under this procedure.

However, the agency is still awaiting the government decree that will put these step into effect and clarify how they will work.

Anneke Lucassen, professor of clinical genetics at the University of Southampton, said that the situation in the United Kingdom differed from that in France. France had dealt with the issue of deciding when, and if, genetic information about one person should be passed to relatives by making the patient's confidentiality absolute in such circumstances.

She said, "A doctor [in France] may not disclose any information to relatives without an individual's consent. In the UK, however, despite the importance of confidentiality... a concern for harm to others places a limitation on this duty.

"Both professional guidelines and UK case law recognise that confidentiality can be breached legitimately if [this is] considered [to be] in the public interest. Genetic tests have rarely been mentioned as reasons to breach confidentiality.

"But this is changing," she added. "The General Medical Council is currently updating its guidance on confidentiality."

Bilan d'Application de la Loi de Bioéthique du 6 Aoî 2004, Rapport Remis à la Ministre Chargée de la Santé can be found at www.agence-biomedecine.fr.

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