DNA Testing - Predictive and Diagnostic for Serious Adult Onset Neurogenetic Disorders - Guidelines

Document Number PD2005_303
Publication date 27-Jan-2005
Functional Sub group Clinical/ Patient Services - Medical Treatment
Summary Guidelines for predictive and diagnostic DNA testing for serious adult onset neurogenetic disorders with predictive implications for other family members and which are likely to reduce normal life expectancy.
Author Branch Agency for Clinical Innovation
Branch contact Agency for Clinical Innovation 02 94644606
Distributed to Public Health System, Divisions of General Practice, Health Professional Associations and Related Organisations, NSW Ambulance Service, Ministry of Health, Public Health Units, Public Hospitals, Private Hospitals and Day Procedure Centres
Review date 30-Jun-2017
Policy Manual Not applicable
File No. 98/4807
Previous reference 2003/25
Issue date 04-Apr-2003
Status Active

Director-General

This Policy Directive may be varied, withdrawn or replaced at any time. Compliance with this directive is mandatory for NSW Health and is a condition of subsidy for public health organisations.
DNA Testing - Predictive and Diagnostic for Serious Adult Onset Neurogenetic Disorders - Guidelines

Document Number  PD2005_303
Publication date  27-Jan-2005
Functional Sub group  Clinical/ Patient Services - Medical Treatment
Summary  Guidelines for predictive and diagnostic DNA testing for serious adult onset neurogenetic disorders with predictive implications for other family members and which are likely to reduce normal life expectancy.
Author Branch  NSW Kids and Families
Branch contact  NSW Kids & Families 9391 9503
Distributed to  Public Health System, Divisions of General Practice, Health Professional Associations and Related Organisations, NSW Ambulance Service, Ministry of Health, Public Health Units, Public Hospitals, Private Hospitals and Day Procedure Centres
Review date  27-Sep-2010
Policy Manual  Not applicable
File No.  98/4807
Previous reference  2003/25
Issue date  04-Apr-2003
Status  Active

Director-General

This Policy Directive may be varied, withdrawn or replaced at any time. Compliance with this directive is mandatory for NSW Health and is a condition of subsidy for public health organisations.
Guidelines for predictive and diagnostic DNA testing for serious adult onset neurogenetic disorders with predictive implications for other family members and which are likely to reduce normal life expectancy.

1 INTRODUCTION

This circular replaces circular 2001/87.

Predictive and diagnostic testing using DNA (or sometimes other analytes) is available for a number of adult onset genetic diseases, many of which result in presently incurable illness, dementia and premature death.

Predictive testing refers to testing in an individual who currently does not have symptoms or signs of disease, but who may be at risk due to their family history, and who requests more information about their risk.

Serious adult onset neurogenetic disorders likely to reduce normal life expectancy include Huntington disease, motor neurone disease, spinocerebellar ataxia and pre-senile dementias.

2 PREDICTIVE TESTING

DNA predictive testing for serious adult onset neurogenetic disorders carried out by NSW Health public hospital laboratories shall only be undertaken when requested by certified clinical geneticists.

2.1 Rationale

This requirement is to ensure that patients receive care according to best practice guidelines (Appendix 1). Taking a predictive DNA test is a major life decision and the results are irreversible. Predictive testing raises a number of complex issues outlined below and it is essential that prior to undertaking testing, the patient is fully informed about the implications of testing and is prepared for the results.
Clinical geneticists requesting predictive testing are required to have expertise in the disorder being tested, the complexities of predictive testing, interpretation of results and their implications and follow-up management strategies. Genetics Service staff work in close liaison with referring practitioners to ensure continuity of care.

### 2.2 Offering predictive testing

Predictive testing is best offered through a team which in addition to a clinical geneticist includes a neurologist, psychiatrist, genetic social worker/genetic counsellor, psychologist and laboratory scientists. A list of genetics services is included as Appendix 2.

The diagnosis in the affected relative should be verified. A baseline neurological consultation should be offered for any person undergoing predictive testing.

### 2.3 Laboratory requirements

Laboratories will only commence testing on receipt of all information required:

- Information as indicated in the attached request form (Appendix 3)
- A photocopy of the completed consent form (Appendix 4). Completion of the consent form provides the opportunity to address relevant issues in predictive testing.

Laboratories are to keep a list of certified clinical geneticists.

### 2.4 Results and their implications

A **positive test result** will indicate that the individual is at high risk of being affected by the disease, although the actual risk varies from disorder to disorder. For example, for Huntington disease a positive test result means that the person will almost definitely develop the condition if they live long enough. The test result may not accurately predict age of onset or severity of the condition. In the case of a positive result, the person should be offered follow-up with the neurologist. Some results are indeterminate or in the intermediate range and require very specialised interpretation. Such results may raise complex psychosocial issues for the individual being tested and their family, and on-going support is often necessary. A positive result may have implications for future reproductive decisions and possible adverse consequences for employment and insurance. The individual should be informed that routine check-ups with an appropriate specialist may be an alternative to a predictive genetic test.

**There may be implications for other family members** because genetic disorders are inherited. For example, for Huntington disease, a positive predictive test result means that the individual's children and siblings have a 50% chance of inheriting the mutation. Privacy and confidentiality issues need consideration, particularly the need to balance the right to privacy and confidentiality of the person being tested, with their
responsibility to inform other family members, who potentially may suffer harm if their risk status is not disclosed to them.

3 DIAGNOSTIC TESTING

Neurologists and specialists requesting DNA diagnostic testing for adult onset neurogenetic disorders undertaken by NSW Health Public Hospital Laboratories are reminded that a positive result in diagnostic testing in clinically affected patients will have the same implications for family members as those outlined under predictive testing above. In the case of a positive diagnostic test result it is strongly recommended, that the patient’s family members are offered counselling support and the opportunity for follow-up discussion of their risk with a clinical geneticist (Appendix 2), according to the above guidelines for predictive testing (Appendix 1).

4 CHARGING POLICY

Most of these specialised DNA tests are non Medicare Benefits Schedule Items funded by the NSW Health system. Public or privately referred non-inpatients accessing predictive testing services through a public sector clinical genetics service (Appendix 2), or diagnostic testing through a public sector specialist clinic will be treated as public patients without charge. Private patients will be responsible for their own test costs.

Robyn Kruk
Director-General
1 BEST PRACTICE GUIDELINES FOR PREDICTIVE AND PRESYMPTOMATIC DNA TESTING

1.1 Guidelines for Testing for Genetic Disorders, Circular 97/48  

1.2 Specialised Testing for Genetic Disorders, State Health Publication No (SWS) 000061, ISBN: 0 7347 3165 5  

1.3 Guidelines for predictive testing for genetic disorders; Human Genetics Society of Australasia, available at  
www.hgsa.com.au

1.4 Predictive testing in children and adolescents, Human Genetics Society of Australasia, available at  
www.hgsa.com.au

1.5 Accreditation Standards for Nucleic Acid Detection Techniques (Section 1.2)  

1.6 Ethical Code Governing the Provision of Genetics Services  
State Health Publication No (SWS) 980068, ISBN: 0 7313 4036 1  

2 DISORDER SPECIFIC GUIDELINES

2.1 Huntington Disease  
Guidelines for the molecular genetics predictive test in Huntington disease,  
http://www.huntington-assoc.com/guidel.htm
Appendix 2

Clinical and Genetic Counselling Service Locations
Clinical and Counselling Services

CAMPERDOWN
Department of Molecular and Clinical Genetics
Royal Prince Alfred Hospital
CAMPERDOWN NSW 2050
Tel: 02 9515 5080
Fax: 02 9515 7595

LIVERPOOL
Department of Clinical Genetics
Health Services Building
Cnr Campbell and Goulburn Sts
LIVERPOOL NSW 2170
Tel: 02 9828 4665
Fax: 02 9828 4650

PENRITH
Nepean Hospital
PENRITH NSW 2750
Tel: 4734 3362
Fax: 4734 2567

RANDWICK
Department of Medical Genetics
Sydney Children's Hospital
RANDWICK NSW 2031
Tel: 02 9382 1708
Fax: 02 9382 1711

WESTMEAD
Department of Clinical Genetics
The New Children's Hospital
WESTMEAD NSW 2145
Tel: 02 9845 3273
Fax: 02 9845 3204

NEWCASTLE
Hunter Genetics
Cnr Turton & Tinonee Sts
WARATAH NSW 2298
Tel: 4985 3100
Fax: 4985 3105
Genetic Counselling Services in conjunction with visiting clinical genetics services

KOGARAH
Women's and Children's Health
2nd Floor Prichard Wing
St George Hospital
Gray Street
KOGARAH NSW 2217
Tel: 02 9350 2315
Fax: 02 9350 3901T

ST LEONARDS
Fetal Medicine Unit
Royal North Shore Hospital
ST LEONARDS NSW 2065
Tel: 02 9926 6478
Fax: 02 9906 1872

BATHURST
Community Health Centre
158 William Street
BATHURST NSW 2795
Tel: 6331 5533
Fax: 6332 2039

BROKEN HILL
Community Health Centre
BROKEN HILL NSW 2880
Tel: 08 8080 1556
Fax: 08 8080 1611

CANBERRA
The Antenatal Clinic
The Canberra Hospital
PO Box 11
CANBERRA ACT 2605
Tel: 6244 4042
Fax: 6244 3422

COFFS HARBOUR
Coffs Harbour Health Campus
Pacific Highway
COFFS HARBOUR 2450
Tel: 6656 7806
Fax: 6656 7817
GOSFORD
Central Coast Health
Public Health Unit
PO Box 361
GOSFORD NSW 2250
Tel: 4337 0207
Fax: 4337 0217

GOULBURN
Child Development Unit
Cnr Albert and Clifford Streets
GOULBURN NSW 2580
Tel: 4827 3951
Fax: 4827 3958

LISMORE
37 Oliver Avenue
GOONELLABAUGH NSW 2480
Tel: 6625 0111
Fax: 6625 0102

MUDGEE/DUBBO
Mudgee Community Health Centre
MUDGEE NSW 2850
Tel: 6372 6455
Fax: 6372 7341

MUSWELLBROOK
Community Health Centre
Brentwood Street
MUSWELLBROOK NSW 2332
Tel: 6542 2083
Fax: 6542 2005

PORT MACQUARIE
Hastings Macleay Community Health
Morton Street
PORT MACQUARIE 2444
Tel: 6588 2882
Fax: 6588 2800

TAMWORTH
Community Health Centre
180 Peel Street
TAMWORTH NSW 2340
Tel: 6766 2555
Fax: 6766 3967
TAREE/FORSTER
Community Health Centre
64 Putney Street
TAREE NSW 2430
Tel: 6592 9315
Fax: 6592 9607

WAGGA WAGGA
Wagga Base Hospital
WAGGA WAGGA NSW 2650
Tel: 6938 6393
Fax: 6921 5632

WOLLONGONG
Maternal and Paediatric Services
Wollongong Hospital
Crown Street
WOLLONGONG NSW 2500
Tel: 4222 5216
Fax: 4222 5477

Mothersafe
Statewide Medications in Pregnancy and Lactation Advisory Service

Royal Hospital for Women
RANDWICK NSW 2031
Tel: 02 9382 6539 (Sydney calls)
Tel: 1800 647 848 (Other calls)

AGSA
Association of Genetic Support of Australasia Inc.
66 Albion Street
SURRY HILLS NSW 2010
Tel: 02 9211 1462
Fax: 02 9211 8077
Email: agsa@ozemail.com.au
Web: www.agsa-geneticsupport.org.au
Prenatal Diagnosis & Counselling
Specialised services:

CAMPERDOWN
Fetal Medicine Unit
King George V Hospital
CAMPERDOWN NSW 2050
Tel: 02 9515 8258
Fax: 02 9515 6579

LIVERPOOL
Fetal Medicine Unit
Liverpool Hospital
Elizabeth Drive
LIVERPOOL NSW 2170
Tel: 02 9828 4145
Fax: 02 9828 4146

RANDWICK
Prenatal Diagnosis
Royal Hospital for Women
RANDWICK NSW 2031
Tel: 9382 6098
Fax: 9382 6706

PENRITH
Fetal Medicine Unit
Nepean Hospital
PENRITH NSW 2750
Tel: 02 4724 3163
Fax: 02 4724 3206

ST LEONARDS
Fetal Medicine Unit
Royal North Shore Hospital
ST LEONARDS NSW 2065
Tel: 02 9926 7280
Fax: 02 9906 1872

WESTMEAD
Fetal Medicine Unit
Westmead Centre
WESTMEAD NSW 2145
Tel: 02 9845 6802
Fax: 02 9845 7793
NEWCASTLE
Prenatal Diagnosis Unit
John Hunter Hospital
NEWCASTLE NSW 2310
Tel: 4921 4694
Fax: 4921 3133
Cancer Genetics

Specialised services:

**DARLINGHURST**
Family Cancer Clinic
Department of Medical Oncology
St Vincent's Hospital
Victoria Street
DARLINGHURST NSW 2010
Tel: 02 8382 3395
Fax: 02 8382 3386

**KOGARAH**
Cancer Care Centre
St George Hospital
Belgrave Street
KOGARAH NSW 2217
Tel: 02 9350 3815
Fax: 02 9350 3958

**LIVERPOOL**
Liverpool Hospital
Elizabeth Drive
LIVERPOOL NSW 2170
Tel: 02 9828 4665
Fax: 02 9828 4650

**RANDWICK**
Hereditary Cancer Clinic
Prince of Wales Hospital
RANDWICK NSW 2031
Tel: 02 9382 2587
Fax: 02 9382 2588

**WESTMEAD**
Familial Cancer Services
Westmead Hospital
WESTMEAD NSW 2145
Tel: 02 9845 5079
Fax 02 9687 2331

**NEWCASTLE**
Hunter Genetics
Cnr Turton & Tinonee Sts
WARATAH NSW 2298
Tel: 4985 3100
Fax: 4985 3105
Further Information:
on services in other areas and newly developed services:

**NSW Genetic Education Program**
PO Box 317,
ST LEONARDS NSW 2065
Tel: 02 9926 7324
Fax: 02 9906 7529
Request Form for Specialised Molecular Genetic/DNA Testing for Genetic Disorders

- Must be used for **non-Medical Benefits Schedule items**
- Before testing is commenced, the laboratory may require the following details
  (see *Guidelines for Specialised DNA Testing for Genetic Disorders*

### Send by courier/express post to:

- Send samples at room temperature
- Same day OR overnight

### Sample Date Drawn ……./……./……. (dd/mm/yyyy)

<table>
<thead>
<tr>
<th>Blood</th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>☐ EDTA</td>
<td>.......... mL (room temp)</td>
<td></td>
</tr>
<tr>
<td>☐ Lithium heparin</td>
<td>.......... mL (room temp)</td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Prenatal</th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>☐ amniotic fluid</td>
<td>.......... mL (room temp)</td>
<td></td>
</tr>
<tr>
<td>☐ cultured amniocytes</td>
<td>xT25 Flask(s) (room temp)</td>
<td></td>
</tr>
<tr>
<td>☐ CVS sample</td>
<td>.......... mg</td>
<td>☐ on ice</td>
</tr>
<tr>
<td>☐ cleaned</td>
<td>☐ uncleaned</td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Other</th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>☐ DNA</td>
<td>.......... g</td>
<td></td>
</tr>
</tbody>
</table>

### Test requested

- PLEASE ATTACH FAMILY/PEDIGREE INFORMATION

### Patient ID

<table>
<thead>
<tr>
<th>Last name</th>
<th>MRN</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>First name</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Address</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
</tr>
</tbody>
</table>

- Postcode

- Date of birth ……./……. /……. (dd/mm/yyyy)  
- Sex  
  - M  
  - F

### Genetic Counselling

- Has the individual been offered counselling consistent with *Specialised/DNA Testing for Genetic Disorders*?

<table>
<thead>
<tr>
<th>☐ Yes</th>
<th>☐ No</th>
<th>☐ Refused</th>
</tr>
</thead>
</table>

### Consent to Testing

- Has a *Consent Form for Specialised/DNA Testing* been completed?

<table>
<thead>
<tr>
<th>☐ Yes</th>
<th>☐ No</th>
</tr>
</thead>
</table>

### Payment of test cost

- ☐ Public patient - Payment to be made by Area Health Service by arrangement (see over)
- ☐ Privately referred non-inpatient - Payment to be made by Area Health Service by arrangement (see over)

<table>
<thead>
<tr>
<th>Name of Area Health Service</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
</tr>
</tbody>
</table>

- ☐ Private patient - Payment to be made by patient.
- Has the patient been advised about the cost of the test and agreed to meet the cost?

<table>
<thead>
<tr>
<th>☐ Yes</th>
<th>☐ No</th>
</tr>
</thead>
<tbody>
<tr>
<td>Purpose of test</td>
<td>Comments</td>
</tr>
<tr>
<td>----------------</td>
<td>----------</td>
</tr>
<tr>
<td>□ Confirm clinical diagnosis</td>
<td></td>
</tr>
<tr>
<td>□ Predictive/presymptomatic testing</td>
<td></td>
</tr>
<tr>
<td>□ Carrier Status</td>
<td></td>
</tr>
<tr>
<td>□ Prenatal Diagnosis - complete box below</td>
<td></td>
</tr>
<tr>
<td>□ Determine feasibility of prenatal Dx</td>
<td></td>
</tr>
<tr>
<td>□ Family study (no report for this individual)</td>
<td></td>
</tr>
<tr>
<td>□ For research (no report for this individual)</td>
<td></td>
</tr>
<tr>
<td>□ Bank DNA until further notice</td>
<td></td>
</tr>
<tr>
<td>□ Other……………………………………………</td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Pregnancy Information (if applicable)</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Is this individual or the partner of this individual currently pregnant</td>
<td></td>
</tr>
<tr>
<td>L.M.P. (dd/mm/yyyy)</td>
<td></td>
</tr>
<tr>
<td>...</td>
<td></td>
</tr>
<tr>
<td>Amnio (dd/mm/yyyy)</td>
<td></td>
</tr>
<tr>
<td>...</td>
<td></td>
</tr>
<tr>
<td>CVS   (dd/mm/yyyy)</td>
<td></td>
</tr>
<tr>
<td>...</td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Family Information</th>
<th>Copy of report to:</th>
</tr>
</thead>
<tbody>
<tr>
<td>Have samples from this family been sent to a DNA lab before?</td>
<td>Name</td>
</tr>
<tr>
<td>□ Yes</td>
<td></td>
</tr>
<tr>
<td>□ No</td>
<td></td>
</tr>
<tr>
<td>If Yes, specify</td>
<td></td>
</tr>
<tr>
<td>..................................................</td>
<td></td>
</tr>
<tr>
<td>Date of birth or age</td>
<td></td>
</tr>
<tr>
<td>..................................................</td>
<td></td>
</tr>
<tr>
<td>Ethnic background</td>
<td></td>
</tr>
<tr>
<td>..................................................</td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Test requested by:</th>
<th>Address</th>
</tr>
</thead>
<tbody>
<tr>
<td>Name .................</td>
<td>Initials</td>
</tr>
<tr>
<td>.................</td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Address</th>
</tr>
</thead>
<tbody>
<tr>
<td>..................................................</td>
</tr>
<tr>
<td>.................</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Telephone No</th>
</tr>
</thead>
<tbody>
<tr>
<td>..................................................</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Signature</th>
<th>Date</th>
</tr>
</thead>
<tbody>
<tr>
<td>.............</td>
<td>............</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Specialty</th>
<th>Appointment</th>
</tr>
</thead>
<tbody>
<tr>
<td>.............</td>
<td>.............</td>
</tr>
</tbody>
</table>
Appendix 4

Consent form for predictive and diagnostic DNA testing for serious adult onset neurogenetic disorders with predictive implications for other family members

This form has been designed to ensure that your consent is on an informed basis. Please read and consider each section.

<table>
<thead>
<tr>
<th>Title</th>
<th>Family Names</th>
<th>MRN</th>
</tr>
</thead>
<tbody>
<tr>
<td>Given Name</td>
<td>VMO</td>
<td></td>
</tr>
<tr>
<td>Address</td>
<td>Street</td>
<td>DOB Sex H1S</td>
</tr>
<tr>
<td>Suburb</td>
<td>Postcode</td>
<td>Admission Date</td>
</tr>
</tbody>
</table>
PROVISION OF INFORMATION TO PATIENT

To be completed by
Health Professional

I, ______________________________________ have informed this patient as detailed below
Insert name of Health Professional and designation
the nature, likely results, and risks associated with gene testing for ________________________

name of disorder

Interpreter present Yes/No

_____________________  _________________________  __________
Signature of Interpreter    Signature of Health Professional
Date

PATIENT CONSENT

To be completed by
Patient/Guardian

______________________________________ and I have discussed predictive testing
Insert name of Health Professional
testing for the analysis of the gene fault (mutation) for ________________________

name of disorder

He/she has told me that:

• The collection of blood will be used to examine my DNA and tested for the gene involved in

_________________________________________

name of disorder

• A positive test result indicates that I have inherited a faulty gene (mutation). This means that I am
at high risk of developing _________________________ and my children and siblings have a ______
%chance of inheriting the mutation

name of disorder

percentage

• A positive test result cannot accurately predict the age of onset of the disorder.

• A negative test result means that I have not inherited the faulty gene (mutation). I will not develop

__________________________ and cannot pass it on to my children

name of disorder

• An intermediate result means that I may or may not develop

_________________________

name of disorder

• In some instances this may have implications for my siblings and children and their descendents

• Test results of one individual can change the estimation of risk for other family members and I have
been advised to inform other adult family members who may be at risk.

• The test result may affect the ability to obtain some types of insurance or employment.
• Testing may reveal non-paternity or non-maternity of a presumed natural parent
• Genetic counselling will be available for myself and other family members during the testing process and after the test result has been given.

I have been told about storage of the test results and the DNA sample. I understand the following:
• The test result will be held by this centre and will only be known by those involved in the testing process.
• My own test result, the fact that I have had a test, and my DNA sample will not be revealed or made available to any other person or organisation outside of the testing process, except with my written consent (as detailed below), or in situations where disclosure is required by law.
• The test results will be given to me first.
• The DNA sample will remain the property of the laboratory. It will be stored in good faith, but its suitability for future use cannot be guaranteed. It will be disposed of at a time determined by standard laboratory practices or regulatory requirements.
• My identified DNA sample will not be used for any other purpose except in accordance with my written consent (as detailed below).

I request and consent to the test described above.

I understand the potential benefits of testing and storing this sample and I accept the risks involved. I have had the chance to ask questions and am satisfied with the explanations and the answers to my questions.

I understand that I may withdraw my consent for this test to be processed.

I consent to my test results being revealed at any time to the following people:

☐ Any family member
☐ Only to the following individuals (specify) ________________________________
☐ My doctor(s) (specify) ________________________________
☐ No other individual
☐ In the event of my death test results may be made known to: ________________________________

After testing has been completed:

☐ I consent to my de-identified DNA sample being used for future Institutional Ethics Committee approved research

OR

☐ My DNA sample may not be used for research without my written consent

___________________
Signature of Patient/Guardian    Print name of Patient    Date

Explanation of terms used in this consent form
• A gene test involves analysis of one or more of those genes to determine whether a mutation is present
• Mutation: Change in the normal DNA code which may cause disease
• DNA (Deoxyribonucleic acid): The chemical compound of which the genes are made