



## Sri Lakshmi Narayana Institute of Medical Sciences

Date: 18-04-2020

From

Dr.G.SOMASUNDRAM  
Principal of Allied Health Sciences,  
Sri Lakshmi Narayana Institute of Medical Sciences  
Bharath Institute of Higher Education and Research,  
Chennai.

To

The Dean,  
Sri Lakshmi Narayana Institute of Medical College  
Bharath Institute of Higher Education and Research,  
Chennai.

### **Sub: Permission to conduct value-added course: Genetic disorders**

Dear Sir,

With reference to the subject mentioned above, the department proposes to conduct a value-added course titled: **genetic disorders** from May to June 2020. We solicit your kind permission for the same.

Kind Regards

Dr.G.Somasundram

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### **FOR THE USE OF DEANS OFFICE**

Names of Committee members for evaluating the course:

The Dean: **Dr. Balagurunathan. K**

The HOD: **Dr. Somasundram. G**

The Expert: **Dr. Kabilan**

The committee has discussed about the course and is approved.

Dean

(Sign & Seal)

Subject Expert

(Sign & Seal)

HOD

(Sign & Seal)

**Dr. G. JAYALAKSHMI, BSC., MBBS., DTCD., M.D.,**  
**DEAN**  
Sri Lakshmi Narayana Institute of Medical Sciences  
Osudu, Agaram, Kudapakkam Post,  
Villanur Commune, Puducherry - 605502.

**PRINCIPAL**  
Allied Health Sciences  
Sri Lakshmi Narayana Institute of Allied Health Sciences  
Osudu, Agaram Post, Puducherry - 605 502.



OFFICE OF THE DEAN

# Sri Lakshmi Narayana Institute of Medical Sciences

OSUDU, AGARAM VILLAGE, VILLIANUR COMMUNE, KUDAPAKKAM POST,  
PUDUCHERRY - 605 502.

[ Recognised by Medical Council of India, Ministry of Health letter No. U/12012/249/2005-ME ( P -II ) dt. 11/07/2011 ]  
[ Affiliated to Bharath University, Chennai - TN ]

## Circular

29.04.2020

**Sub: Organizing Value-added Course: “genetic disorders “.reg**

With reference to the above-mentioned subject, it is to bring to your notice that Sri Lakshmi Narayana Institute of Medical Sciences, **Bharath Institute of Higher Education and Research** is organizing “**genetic disorders**”. The course content and registration form is enclosed below.”

The application must reach the institution along with all the necessary documents as mentioned. The hard copy of the application should be sent to the institution by registered/ speed post only so as to reach on or before May to June 2020. Applications received after the mentioned date shall not be entertained under any circumstances.

DEAN

Encl: Copy of Course content

**Dr. G. JAYALAKSHMI, BSC., MBBS., DTCD., M.D.,**

**DEAN**

**Sri Lakshmi Narayana Institute of Medical Sciences**

**Osudu, Agaram, Kudapakkam Post,**

**Villianur Commune, Puducherry - 605502.**

## VALUE ADDED COURSE

### 1. Name of the programme & Code

“Genetic disorders” & VAC07/AHS/2020-18/05

### 2. Duration & Period

30 hrs. & May to June 2020

### 3. Information Brochure and Course Content of Value-Added Courses

*Enclosed as Annexure- I*

### 4. List of students enrolled

*Enclosed as Annexure- II*

### 5. Assessment procedures:

Assessment - *Enclosed as Annexure- III*

### 6. Certificate model

*Enclosed as Annexure- IV*

### 7. No. of times offered during the same year:

1 time May to June 2020

### 8. Year of discontinuation: 2021

### 9. Summary report of each program year-wise

Value Added Course- May to June 2020					
Sl. No	Course Code	Course Name	Resource Persons	Target Students	Strength & Year
1	VAC07/AHS/2020-18/05	Genetic disorders	DR . KABILAN	AHS	25 students May to June 2020

### 10. Course Feed Back

*Enclosed as Annexure- V*

RESOURCE PERSON



COORDINATOR

Dr.G.Somasundram

**PRINCIPAL**  
Allied Health Sciences  
Sri Lakshmi Narayana Institute of Allied Health Sciences  
Osudu, Agaram Post, Puducherry - 605 502.

## Course Proposal

Course Title: “Genetic disorder”

### **Course Objective:**

1. To enhance the performance skill in Genetic disorders.
2. To assess the objectives and protocols in Genetic disorder.
3. To assess the reaction of target allied Health students towards the Genetic disorder by getting their feedback.

**Course Outcome:** Improvement in the “Genetic disorder”

**Course Audience:** Students of AHS Batch 2020

**Course Coordinator:** Dr.G. Somasundaram

**Course Faculties with Qualification and Designation:**

**1.Course Curriculum/Topics with schedule (Min of 30 hours)**

SlNo	Date	Topic	Time	Hours
1.	18.05.2020	Introduction to Genetic disorders, Background, Objectives,	4-5p.m	1
2.	20.05.2020	Types of genetic disorders	2-3p.m	1
3.	21.05.2020	Single gene disorder	4-6p.m	2
4.	22.05.2020	Explanation about autosomal dominant and receive	4-6p.m	2
5.	25.05.2020	X linked dominant & receive	4-6p.m	2
6.	28.05.2020	Examples for the genetic disorders	4-5p.m	2
7.	30.05.2020	conventional didactic lecture and video	4-5P.M	1
8.	02.06.2020	Multifractional disorders	4-5p.m	1
9.	03.06.2020	Explanation about chromosomal disorders	4-6p.m	1
10.	04.06.2020	Common genetic disorders	4-6p.m	2
11.	05.06.2020	Causes of genetic disorders	4-6p.m	1
12.	08.06.2020	Symptoms of genetic disorders	4-6p.m	2
13.	09.06.2020	Pre course and Post Course evaluation, Feedback analysis from Likert scale	2-5p.m	3
		<b>Practical Class I</b>		
13.	10.06.2020	Steps model explanation and various performance assessment methods		1
14.	11.06.2020	Orientation of the students about the training program and assessment methodology by DOPS		1

15.	12.06.2017	Video demonstration of genetic disorders		2
16.	13.06.2017	Genetic disorders procedure by STEPS model		2
17.	15.06.2017	Assessment by DOPS procedure and giving feedback in weaker areas	2-6p.m	4
		<b>Total</b>		30 hrs

**REFERENCE BOOKS:**

1. GIRISH C & YATHISH K.R, Genetics Made Easy.
2. Dr. Ropali Forta, An Easy Approach to Human Genetics
3. Pranab Kr Banerjee , Problems on Genetics , Molecular Genetics and Evolutionary Genetics

# GENETIC DISORDERS

- A genetic disorder is a health problem caused by one or more abnormalities in the [genome](#).
- It can be caused by a [mutation](#) in a single [gene](#) (monogenic) or multiple genes (polygenic) or by a [chromosomal abnormality](#).
- Although polygenic disorders are the most common, the term is mostly used when discussing disorders with a single genetic cause, either in a gene or [chromosome](#).
- The mutation responsible can occur spontaneously before [embryonic development](#) (a *de novo* mutation), or it can be [inherited](#) from two parents who are carriers of a faulty gene ([autosomal recessive](#) inheritance) or from a parent with the disorder ([autosomal dominant](#) inheritance).
- When the genetic disorder is inherited from one or both parents, it is also classified as a **hereditary disease**.
- Some disorders are caused by a mutation on the [X chromosome](#) and have [X-linked](#) inheritance.
- Very few disorders are inherited on the [Y chromosome](#) or [mitochondrial DNA](#).
- There are well over 6,000 known genetic disorders, and new genetic disorders are constantly being described in medical literature.
- More than 600 genetic disorders are treatable.
- Around 1 in 50 people are affected by a known single-gene disorder, while around 1 in 263 are affected by a [chromosomal disorder](#).
- Around 65% of people have some kind of health problem as a result of congenital genetic mutations.
- Due to the significantly large number of genetic disorders, approximately 1 in 21 people are affected by a genetic disorder classified as "[rare](#)" (usually defined as affecting less than 1 in 2,000 people). Most genetic disorders are rare in themselves

- Genetic disorders are present before birth, and some genetic disorders produce [birth defects](#), but birth defects can also be [developmental](#) rather than [hereditary](#).
- The opposite of a hereditary disease is an [acquired disease](#).
- Most [cancers](#), although they involve genetic mutations to a small proportion of cells in the body, are acquired diseases. Some [cancer syndromes](#), however, such as [BRCA mutations](#), are hereditary genetic disorders



## Single-gene

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- A **single-gene disorder** (or **monogenic disorder**) is the result of a single [mutated](#) gene.
- Single-gene disorders can be passed on to subsequent generations in several ways. [Genomic](#)

[imprinting](#) and [uniparental disomy](#), however, may affect inheritance patterns

- . The divisions between [recessive and dominant](#) types are not "hard and fast", although the divisions between [autosomal](#) and [X-linked](#) types are (since the latter types are distinguished purely based on the chromosomal location of the gene).
- For example, the common form of [dwarfism](#), [achondroplasia](#), is typically considered a dominant disorder, but children with two genes for achondroplasia have a severe and usually lethal skeletal disorder, one that achondroplasics could be considered carriers for. [Sickle-cell anemia](#) is also considered a recessive condition, but [heterozygous](#) carriers have increased resistance to [malaria](#) in early childhood, which could be described as a related dominant condition. When a couple where one partner or both are sufferers or carriers of a single-gene disorder wish to have a child, they can do so through *in vitro* fertilization, which enables preimplantation genetic diagnosis to occur to check whether the embryo has the genetic disorder.
- Most congenital [metabolic](#) disorders known as [inborn errors of metabolism](#) result from single-gene defects.
- Many such single-gene defects can decrease the fitness of affected people and are therefore present in the population in lower frequencies compared to what would be expected based on simple probabilistic calculations

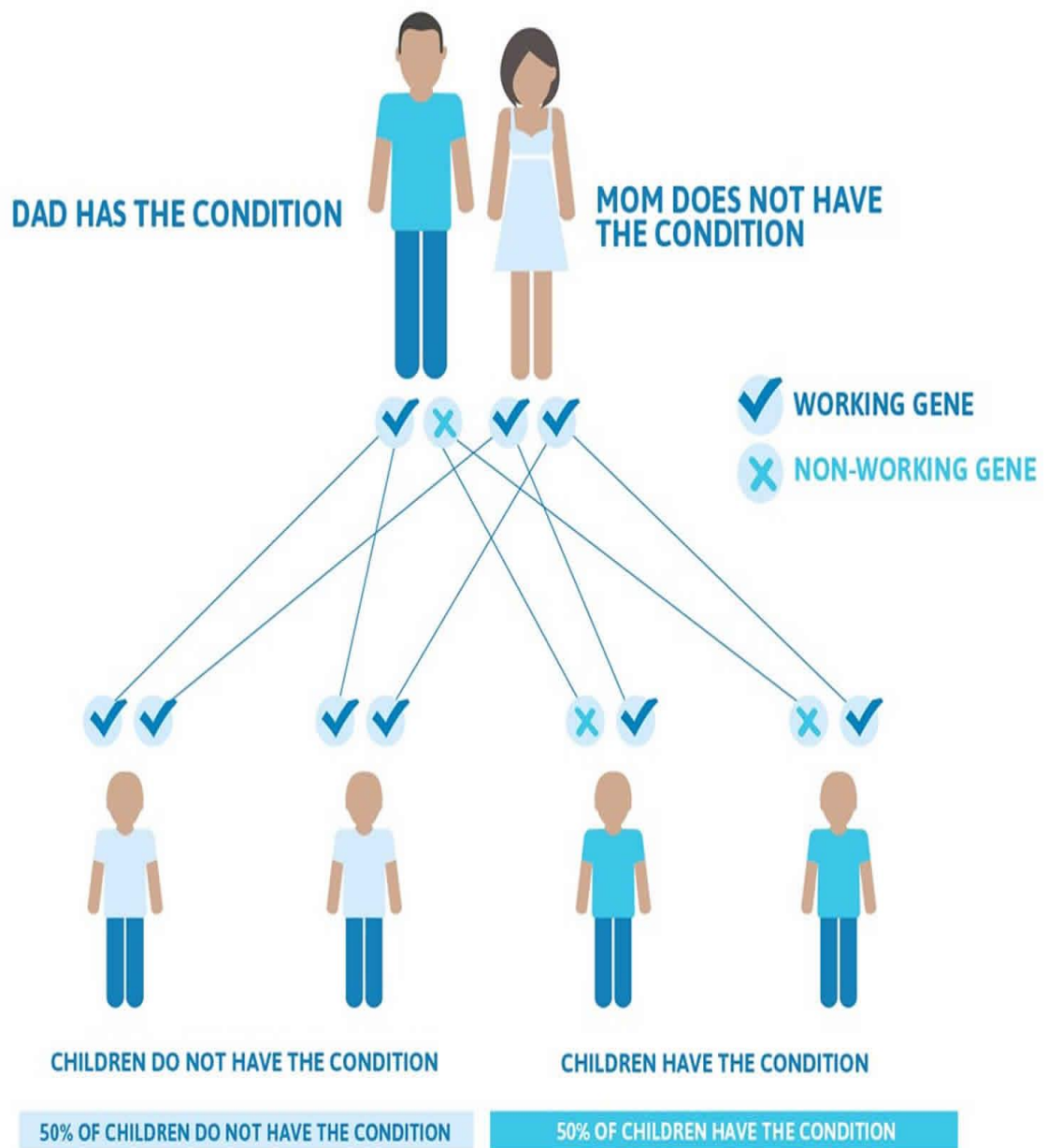




## Autosomal dominant

- Only one mutated copy of the gene will be necessary for a person to be affected by an autosomal dominant disorder. Each affected person usually has one affected parent.
- The chance a child will inherit the mutated gene is 50%. Autosomal dominant conditions sometimes have reduced [penetrance](#), which means although only one mutated copy is needed, not all individuals who inherit that mutation go on to develop the disease.
- Examples of this type of disorder are [Huntington's disease](#), [neurofibromatosis type 1](#), [neurofibromatosis type 2](#), [Marfan syndrome](#), [hereditary nonpolyposis colorectal cancer](#), [hereditary multiple exostoses](#) (a highly penetrant autosomal dominant disorder), [tuberous sclerosis](#), [Von Willebrand disease](#), and [acute intermittent porphyria](#). Birth defects are also called congenital anomalies

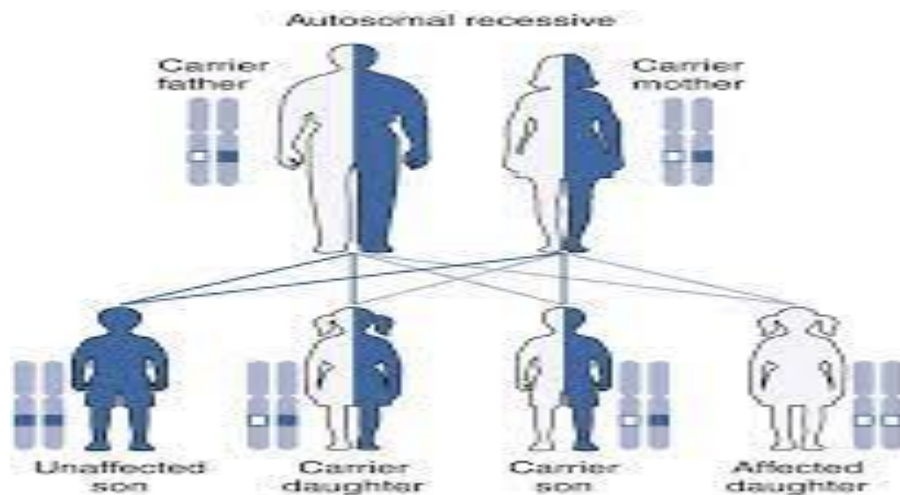
# Autosomal Dominant Inheritance Pattern



## Autosomal recessive

- Two copies of the gene must be mutated for a person to be affected by an autosomal recessive disorder.
- An affected person usually has unaffected parents who each carry a single copy of the mutated gene and are referred to as genetic carriers

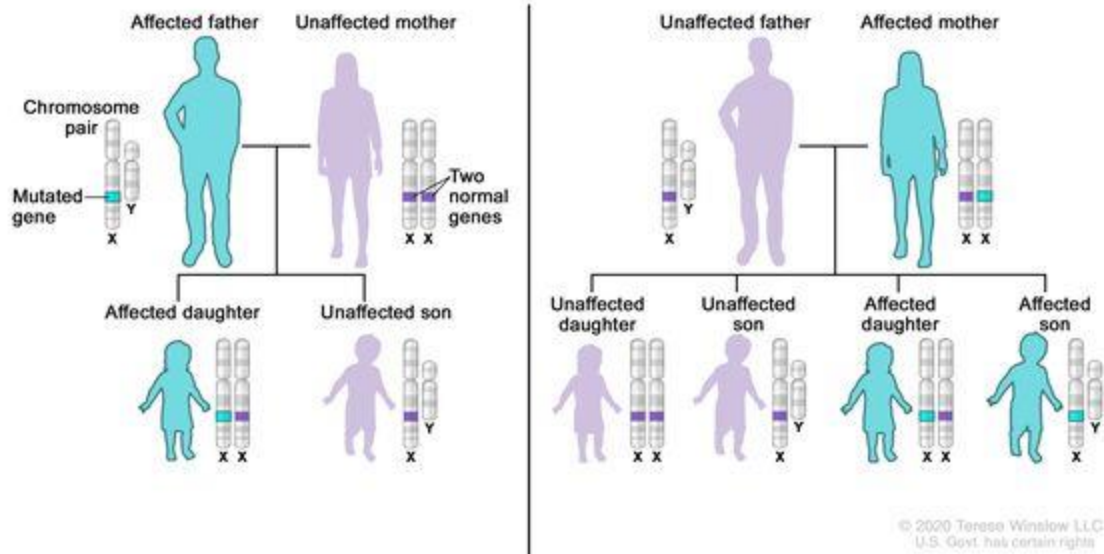
- Each parent with a defective gene normally do not have symptoms.
- Two unaffected people who each carry one copy of the mutated gene have a 25% risk with each pregnancy of having a child affected by the disorder.
- Examples of this type of disorder are [albinism](#), [medium-chain acyl-CoA dehydrogenase deficiency](#), [cystic fibrosis](#), [sickle cell disease](#), [Tay–Sachs disease](#), [Niemann–Pick disease](#), [spinal muscular atrophy](#), and [Roberts syndrome](#).
- Certain other phenotypes, such as wet versus dry [earwax](#), are also determined in an autosomal recessive fashion.
- Some autosomal recessive disorders are common because, in the past, carrying one of the faulty genes led to a [slight protection](#) against an infectious disease or [toxin](#) such as [tuberculosis](#) or [malaria](#). Such disorders include [cystic fibrosis](#),<sup>1</sup> [sickle cell disease](#), [phenylketonuria](#) and [thalassaemia](#).



## X-linked dominant

- x-linked dominant disorders are caused by mutations in genes on the [X chromosome](#). Only a few disorders have this inheritance pattern, with a prime example being [X-linked hypophosphatemic rickets](#).
- Males and females are both affected in these disorders, with males typically being more severely affected than females.
- Some X-linked dominant conditions, such as [Rett syndrome](#), [incontinentia pigmenti](#) type 2, and [Aicardi syndrome](#), are usually fatal in males either *in utero* or shortly after birth, and are therefore predominantly seen in females.
- Exceptions to this finding are extremely rare cases in which boys with [Klinefelter syndrome](#) (44+xy) also inherit an X-linked dominant condition and exhibit symptoms more similar to those of a female in terms of disease severity
- The chance of passing on an X-linked dominant disorder differs between men and women.
- The sons of a man with an X-linked dominant disorder will all be unaffected (since they receive their father's Y chromosome), but his daughters will all inherit the condition
- A woman with an X-linked dominant disorder has a 50% chance of having an affected fetus with each pregnancy, although in cases such as incontinentia pigmenti, only female offspring are generally viable.

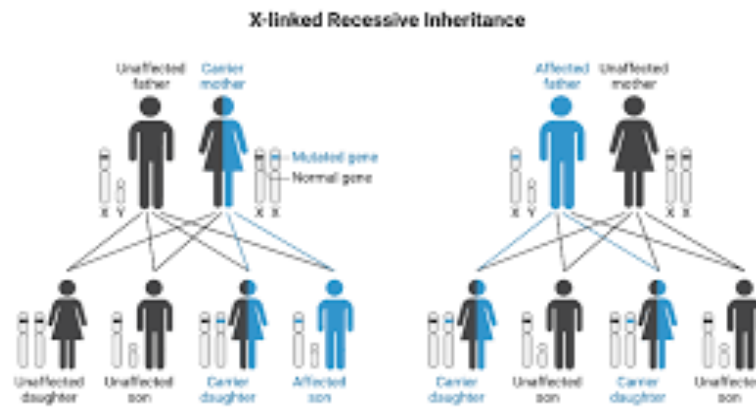
## X-Linked Dominant Inheritance



## X-linked recessive

- X-linked recessive conditions are also caused by mutations in genes on the X chromosome. Males are much more frequently affected than females, because they only have the one X chromosome necessary for the condition to present.
- The chance of passing on the disorder differs between men and women.
- The sons of a man with an X-linked recessive disorder will not be affected (since they receive their father's Y chromosome), but his daughters will be carriers of one copy of the mutated gene.
- A woman who is a carrier of an X-linked recessive disorder ( $X^R X^r$ ) has a 50% chance of having sons who are affected and a 50% chance of having daughters who are carriers of one copy of the mutated gene.

- X-linked recessive conditions include the serious diseases [hemophilia A](#), [Duchenne muscular dystrophy](#), and [Lesch–Nyhan syndrome](#), as well as common and less serious conditions such as [male pattern baldness](#) and red–green [color blindness](#).
- X-linked recessive conditions can sometimes manifest in females due to [skewed X-inactivation](#) or monosomy X ([Turner syndrome](#)).

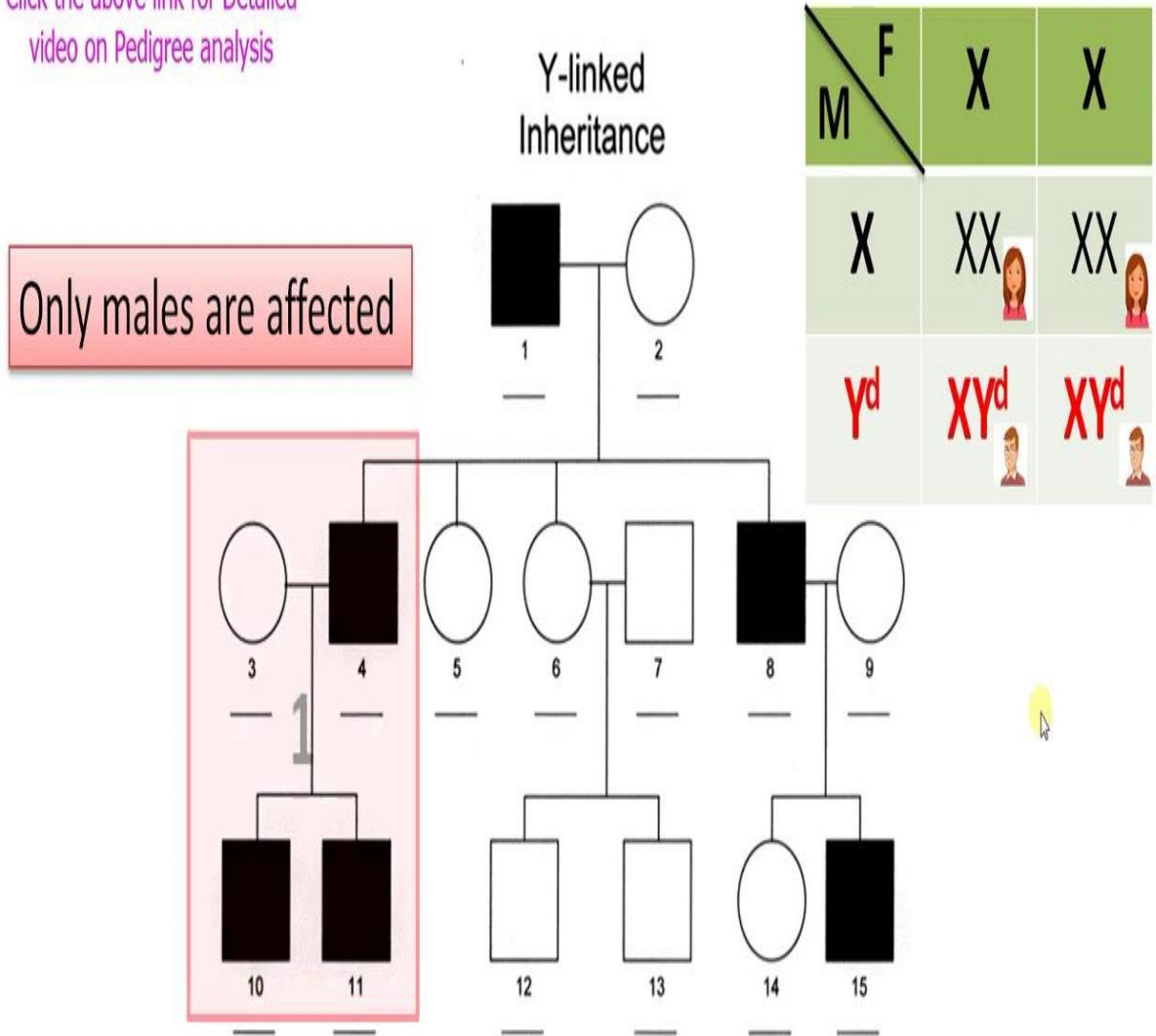


## Y-linked

- Y-linked disorders are caused by mutations on the Y chromosome. These conditions may only be transmitted from the heterogametic sex (e.g. male humans) to offspring of the same sex.
- More simply, this means that Y-linked disorders in humans can only be passed from men to their sons; females can never be affected because they do not possess Y-allosomes.
- Y-linked disorders are exceedingly rare but the most well-known examples typically cause infertility.

- Reproduction in such conditions is only possible through the circumvention of infertility by medical intervention.

Click the above link for Detailed video on Pedigree analysis



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## Mitochondrial

- This type of inheritance, also known as maternal inheritance, is the rarest and applies to the 13 genes encoded by [mitochondrial DNA](#).
- Because only egg cells contribute mitochondria to the developing embryo, only mothers (who are affected) can pass on mitochondrial DNA conditions to their children. An example of this type of disorder is [Leber's hereditary optic neuropathy](#).
- It is important to stress that the vast majority of [mitochondrial diseases](#) (particularly when symptoms develop in early life) are actually caused by a [nuclear gene](#) defect, as the mitochondria are mostly developed by non-mitochondrial DNA. These diseases most often follow autosomal recessive inheritance

## Multifactorial disorder

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- Genetic disorders may also be complex, multifactorial, or polygenic, meaning they are likely associated with the effects of multiple genes in combination with lifestyles and environmental factors.
- Multifactorial disorders include [heart disease](#) and [diabetes](#). Although complex disorders often cluster in families, they do not have a clear-cut pattern of inheritance.
- This makes it difficult to determine a person's risk of inheriting or passing on these disorders.
- Complex disorders are also difficult to study and treat because the specific factors that cause most of these disorders have not yet been identified. Studies that aim



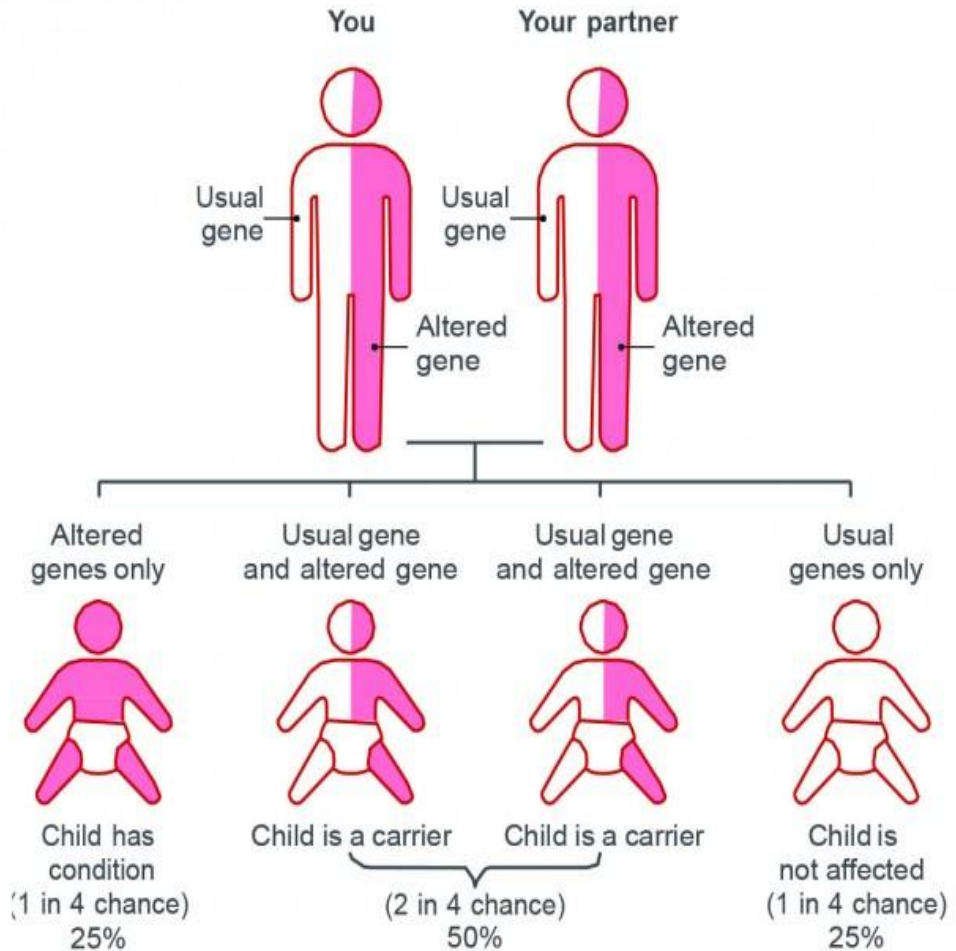
to identify the cause of complex disorders can use several methodological approaches to determine [genotype–phenotype](#) associations.

- One method, the [genotype-first approach](#), starts by identifying genetic variants within patients and then determining the associated clinical manifestations.
- This is opposed to the more traditional phenotype-first approach, and may identify causal factors that have previously been obscured by clinical [heterogeneity](#), [penetrance](#), and expressivity

## Chromosomal disorder

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- A chromosomal disorder is a missing, extra, or irregular portion of chromosomal DNA.
- It can be from an atypical number of chromosomes or a structural abnormality in one or more chromosomes. An example of these disorders is trisomy 21 ([Down syndrome](#)), in which there is an extra copy of chromosome 21.



## What are common genetic disorders?

- There are many types. They include:
- **Chromosomal disorders**
- [Down syndrome](#) (Trisomy 21).
- [FragileX syndrome](#).
- [Klinefelter syndrome](#).
- [Triple-X syndrome](#).
- [Turner syndrome](#).
- Trisomy 18.

- Trisomy 13.

## Multifactorial disorders

- Late-onset [Alzheimer's disease](#).
- [Arthritis](#).
- [Autism spectrum disorder](#), in most cases.
- [Cancer](#), in most cases.
- [Coronary artery disease](#).
- [Diabetes](#).
- [Migraine headaches](#).
- [Spina bifida](#).

## Monogenic disorders

- [Cystic fibrosis](#).
- Deafness that's present at birth (congenital).
- Duchenne [muscular dystrophy](#).
- Familial hypercholesterolemia, a type of [high cholesterol disease](#).
- [Hemochromatosis \(iron overload\)](#).
- [Neurofibromatosis type 1 \(NF1\)](#).
- [Sickle cell disease](#).
- [Tay-Sachs disease](#).

## Are there other types of genetic disorders?

Genetic disorders may also cause rare diseases. This group of conditions affects fewer than 200,000 people in the U.S. According to experts, there may be as many as 7,000 of these diseases.

Rare genetic disorders include:

- [AA amyloidosis](#).
- [Adrenoleukodystrophy \(ALD\)](#).
- [Ehlers-Danlos syndrome](#).

- [Mitochondrial diseases.](#)
- [Usher syndrome.](#)

## What are the causes of genetic disorders?

- To understand genetic disorder causes, it's helpful to learn more about how your genes and DNA work.
- Most of the DNA in your genes instructs the body to make proteins. These proteins start complex cell interactions that help you stay healthy.
- When a mutation occurs, it affects the genes' protein-making instructions. There could be missing proteins.
- Or the ones you have do not function properly. Environmental factors (also called mutagens) that could lead to a genetic mutation include:
  - Chemical exposure.
  - Radiation exposure.
  - Smoking.
  - UV exposure from the sun

## What are the symptoms of genetic disorders?

Symptoms vary depending on the type of disorder, organs affected and how severe it is.

- Behavioral changes or disturbances.
- Breathing problems.
- Cognitive deficits, when the brain can't process information as it should.

- [Developmental delays](#) that include challenges with speech or social skills.
- Eating and digestive issues, such as [difficulty swallowing](#) or an inability to process nutrients.
- Limb or facial anomalies, which include missing fingers or a [cleft lip and palate](#).
- Movement disorders due to muscle stiffness or weakness.
- Neurological issues such as seizures or [stroke](#).
- Poor growth or [short stature](#).
- Vision or [hearing loss](#).

## How are genetic disorders identified?

If you have a family history of a genetic disorder, you may wish to consider genetic counseling to see if genetic testing is appropriate for you.

Lab tests can typically show whether you have gene mutations responsible for that condition. In many cases, carrying the mutation does not always mean you'll end up with it.

Genetic counselors can explain your risk and if there are steps you can take to protect your health.

If there's a family history, DNA testing for genetic disorders can be an important part of starting a family. Options include:

- **Carrier testing:** This blood test shows whether you or your partner carry a mutation linked to genetic disorders. This is recommended for everyone considering pregnancy, even if there is no family history.
- **Prenatal screening:** This testing usually involves blood testing from a pregnant woman that tells a person how likely it is that an unborn child could have a common chromosome condition.

- **Prenatal diagnostic testing:** You can find out whether your unborn child faces a higher risk for certain genetic disorders. Prenatal testing uses a sample of fluid from the womb ([amniocentesis](#)).
- **Newborn screening:** This test uses a sample of your newborn baby's blood and is performed on all babies born in Ohio. Detecting genetic disorders early in life can help your child receive timely care if needed.

## What is treatment for genetic disorders like?

Most genetic disorders do not have a cure. Some have treatments that may slow disease progression or lessen their impact on your life. The type of treatment that's right for you depends on the type and severity of the disease. With others, we may not have treatment but we can provide medical surveillance to try to catch complications early.

You may need:

- Medications to manage symptoms or [chemotherapy](#) to slow abnormal cell growth.
- Nutrition counseling or dietary supplements to help you get the nutrients your body needs.
- Physical, occupational or speech therapy to maximize your abilities.
- [Blood transfusion](#) to restore levels of healthy blood cells.
- Surgery to repair abnormal structures or treat complications.
- Specialized treatments, such as [radiation therapy](#) for cancer.
- [Organ transplant](#), which is a procedure to replace a nonfunctioning organ with one from a healthy donor.





# SRI LAKSHMI NARAYANA INSTITUTE OF HIGHER EDUCATION AND RESEARCH

## Assessment Form

*Sri Lakshmi Narayana Institute of Medical Sciences, Puducherry*

### Multiple choice questions

1. Genetic disorder caused by abnormalities in
  - a) Chromosome
  - b) genome
  - c) cells
  - d) none of above
2. The mutation responsible can occur spontaneously before
  - a) Embryonic development
  - b) foetal
  - c) growth
  - d) birth
3. The opposite of a hereditary disease is an
  - a) Acquired disease
  - b) mutation
  - c) inherited
  - d) by birth
4. A single-gene disorder is the result of
  - a) single muted gene
  - b) double muted
  - c) triple gene
  - d) four gene
5. single gene disorder also called as
  - a) monogenic disorder
  - b) myogenic disorder
  - c) cytogenic disorder
  - d) minogenic disorder



6. Example for single genome disorder
  - a) dwarfism
  - b) marfan syndrome
  - c) Willebrand disease
  - d) TB
7. Example for autosomal dominant disorder
  - a) Huntington's disease
  - b) dwarfism
  - c) typhoid
  - d) sickle cell anemia
8. An effected person usually has unaffected parents who each carry a single copy of the mutated gene and are referred to as
  - a) Genetic carriers
  - b) mutation carriers
  - c) chromosome carriers
  - d) cells carriers
9. Example for Autosomal recessive
  - a) Albinism
  - b) Rett syndrome
  - c) porphyria
  - d) dwarfism
10. X-linked dominant disorders are caused by mutations in genes on the
  - a) x chromosome
  - b) y chromosome
  - c) mutation
  - d) genome
11. Example for x linked dominant
  - a. Klinefelter syndrome
  - b. Roberts syndrome
  - c. colorectal cancer
  - d. malaria
12. Y-linked disorders are caused by mutations on the
  - a. Y chromosome
  - b. gene
  - c. X chromosome

d. mutation

13. mitochondrial inheritance also called as

- a) maternal inheritance
- b) eternal
- c) paternal
- d) genome

14. Example for mitochondrial inheritance

- a) lebers hereditary optic neuropathy
- b) hemophilia A
- c) color blindness
- d) turner syndrome

15. chromosomal disorder is missing of

- a) chromosome
- b) gene
- c) mutation
- d) genome

16. Example for trisomy 21

- a) down syndrome
- b) hemophilia
- c) arthritis
- d) Alzheimer disease

17. identify any multifractional disease

- a) spina difidia
- b) fragile x syndrome
- c) triple x syndrome
- d) cystic fibrosis

18. identify the non-environmental factor that leads to genetic disorder

- a) chemical exposure

b) radiation exposure

c) genetics

d) smoking

19. Test used to diagnosis genetic disorder

a) Prenatal screening

b) Newborn screening

c) both a & b

d) none of the above

20. which of the following not a rare genetic disorder

a) AA amyloidosis

b) Adrenoleukodystrophy (ALD)

c) Ehlers-Danlos syndrome

d) AIDS



SRI LAKSHMI NARAYANA INSTITUTE OF HIGHER  
EDUCATION AND RESEARCH

Assessment Form

ANNEXURE - III

*Sri Lakshmi Narayana Institute of Medical Sciences, Puducherry*

COURSE CODE :- VAC07/AHS/2020-18/05

**Multiple choice questions**

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  - a) Chromosome
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- b) marfan syndrome
- c) Willebrand disease
- d) TB

7. Example for autosomal dominant disorder

- a) Huntington's disease
- b) dwarfism
- c) typhoid
- d) sickle cell anemia

8. An affected person usually has unaffected parents who each carry

a single copy of the mutated gene and are referred to as

- a) Genetic carriers
- b) mutation carriers
- c) chromosome carriers
- d) cells carriers

9. Example for Autosomal recessive

- a) Albinism
- b) Rett syndrome
- c) porphyria
- d) dwarfism

10. X-linked dominant disorders are caused by mutations in genes on the

- a) x chromosome
- b) y chromosome
- c) mutation
- d) genome

11. Example for x linked dominant

- a. Klinefelter syndrome
- b. Roberts syndrome
- c. colorectal cancer
- d. malaria

12. Y-linked disorders are caused by mutations on the

- a. Y chromosome
- b. gene
- c. X chromosome

d. mutation

13. mitochondrial inheritance also called as

- a) maternal inheritance
- b) eternal
- c) paternal
- d) genome

14. Example for mitochondrial inheritance

- a) lebers hereditary optic neuropathy
- b) hemophilia A
- c) color blindness
- d) turner syndrome

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- b) gene
- c) mutation
- d) genome

16. Example for trisomy 21

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**SRI LAKSHMI NARAYANA INSTITUTE OF HIGHER  
EDUCATION AND RESEARCH**

**Assessment Form**

ANNEXURE - III

*Sri Lakshmi Narayana Institute of Medical Sciences, Puducherry*

COURSE CODE - VAC07/AHS/2020-18/05

### Multiple choice questions

1. Genetic disorder caused by abnormalities in
  - a) Chromosome
  - b) genome
  - c) cells
  - d) none of above
2. The mutation responsible can occur spontaneously before
  - a) Embryonic development
  - b) foetal
  - c) growth
  - d) birth
3. The opposite of a hereditary disease is an
  - a) Acquired disease
  - b) mutation
  - c) inherited
  - d) by birth
4. A single-gene disorder is the result of
  - a) single muted gene
  - b) double muted
  - c) triple gene
  - d) four gene
5. single gene disorder also called as
  - a) monogenic disorder
  - b) myogenic disorder
  - c) cytogenic disorder
  - d) minogenic disorder

6. Example for single genome disorder

- a) dwarfism
- b) marfan syndrome
- c) Willebrand disease
- d) TB

7. Example for autosomal dominant disorder

- a) Huntington's disease
- b) dwarfism
- c) typhoid
- d) sickle cell anemia

8. An effected person usually has unaffected parents who each carry

a single copy of the mutated gene and are referred to as

- a) Genetic carriers
- b) mutation carriers
- c) chromosome carriers
- d) cells carriers

9. Example for Autosomal recessive

- a) Albinism
- b) Rett syndrome
- c) porphyria
- d) dwarfism

10. X-linked dominant disorders are caused by mutations in genes on the

- a) x chromosome
- b) y chromosome
- c) mutation
- d) genome

11. Example for x linked dominant

- a. Klinefelter syndrome
- b. Roberts syndrome
- c. colorectal cancer
- d. malaria

12. Y-linked disorders are caused by mutations on the

- a. Y chromosome
- b. gene
- c. X chromosome

d. mutation

13. mitochondrial inheritance also called as

- a) maternal inheritance
- b) eternal
- c) paternal
- d) genome

14. Example for mitochondrial inheritance

- a) lebers hereditary optic neuropathy
- b) hemophilia A
- c) color blindness
- d) turner syndrome

15. chromosomal disorder is missing of

- a) chromosome
- b) gene
- c) mutation
- d) genome

16. Example for trisomy 21

- a) down syndrome
- b) hemophilia
- c) arthritis
- d) Alzheimer disease

17. identify any multifractional disease\*

- a) spina difidia
- b) fragile x syndrome
- c) triple x syndrome
- d) cystic fibrosis

18. identify the non-environmental factor that leads to genetic disorder

- a) chemical exposure

b) radiation exposure

c) genetics

d) smoking

19. Test used to diagnosis genetic disorder

a) Prenatal screening

b) Newborn screening

c) both a & b

d) none of the above

20. which of the following not a rare genetic disorder

a) AA amyloidosis

b) Adrenoleukodystrophy (ALD)

c) Ehlers-Danlos syndrome

d) AIDS







# Sri Lakshmi Narayana Institute of Medical Sciences

Affiliated to Bharath Institute of Higher Education & Research  
(Deemed to be University under section 3 of the UGC Act 1956)



## CERTIFICATE OF MERIT

This is to certify that GOKUL A UAH1803184 has actively participated in the Value Added Course on Genetic disorder VAC07/AHS/2020-18/05 held during May to June 2020 Organized by Sri Lakshmi Narayana Institute of Medical Sciences, Pondicherry- 605 502, India.

RESOURCE PERSON

Dr. G.Somasundram

COORDINATOR

**Student Feedback Form****Course Name : GENITIC DISORDER**Subject Code: VAC07/AHS/2020-18/05

Name of Student: \_\_\_\_\_ Roll No.:

We are constantly looking to improve our classes and deliver the best training to you. Your evaluations, comments and suggestions will help us to improve our performance

**Feedback Form**

	Strongly agree	Agree	Neutral	Disagree	Strongly disagree
1. The course met my expectations.	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
2. I will be able to apply the knowledge learned.	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
3. The course objectives for each topic were identified and followed.	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
4. The content was organised and easy to follow.	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
5. The quality of instruction was good.	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
6. Class participation and interaction were encouraged.	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
7. Adequate time was provided for questions and discussion.	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

8. How do you rate the course overall?

- Excellent
- Good
- Average
- Poor
- Very poor

9. The aspects of the course could be improved?

10. Other comments?

**Signature of the student:****Date:**



**Student Feedback Form**

Course Name : GENITIC DISORDER

Subject Code: VAC07/AHS/2020-18/05

Name of Student: DONALD MORISON Roll No.: UAH1805199

We are constantly looking to improve our classes and deliver the best training to you. Your evaluations, comments and suggestions will help us to improve our performance

**Feedback Form**

	Strongly agree	Agree	Neutral	Disagree	Strongly disagree
1. The course met my expectations.	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
2. I will be able to apply the knowledge learned.	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
3. The course objectives for each topic were identified and followed.	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
4. The content was organised and easy to follow.	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
5. The quality of instruction was good.	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
6. Class participation and interaction were encouraged.	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
7. Adequate time was provided for questions and discussion.	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

8. How do you rate the course overall?

- Excellent
- Good
- Average
- Poor
- Very poor

9. The aspects of the course could be improved?

*practical classes should be improved*

10. Other comments?

*→ our staff very good at teaching*

Signature of the student:

Date: 15/06/17

*Donald Morison*

**Student Feedback Form**

Course Name : GENITIC DISORDER

Subject Code: VAC07/AHS/2020-18/05Name of Student: ASHIK R Roll No.: UA11803182

We are constantly looking to improve our classes and deliver the best training to you. Your evaluations, comments and suggestions will help us to improve our performance

**Feedback Form**

	Strongly agree	Agree	Neutral	Disagree	Strongly disagree
1. The course met my expectations.	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
2. I will be able to apply the knowledge learned.	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
3. The course objectives for each topic were identified and followed.	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
4. The content was organised and easy to follow.	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
5. The quality of instruction was good.	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
6. Class participation and interaction were encouraged.	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
7. Adequate time was provided for questions and discussion.	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

8. How do you rate the course overall?

- Excellent
- Good
- Average
- Poor
- Very poor

9. The aspects of the course could be improved? *Improved practical classes only.*10. Other comments? *Very good at teaching.*Signature of the student: *Asht*Date: *15/06/17*

Date: 14.06.2020

From

Dr.G.Somasundram  
Department of Pharmacology,  
Sri Lakshmi Narayana Institute of Medical Sciences  
Bharath Institute of Higher Education and Research,  
Chennai.

Through Proper Channel

To

The Dean,  
Sri Lakshmi Narayana Institute of Medical Sciences  
Bharath Institute of Higher Education and Research,  
Chennai.

**Sub: Completion of value-added course: GENITIC DISORDER for AHS**

Dear Sir,

With reference to the subject mentioned above, the department has conducted the value-added course titled: “ **GENITIC DISORDER**” May to June 2020 for 25 AHS Students . We solicit your kind action to send certificates for the participants that is attached with this letter. Also, I am attaching the photographs captured during the conduct of the course.

Kind Regards,

Dr.G.Somasundram

**Encl: Certificates :**

**Photographs:**






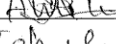
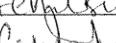



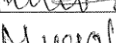
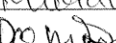
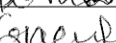
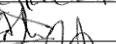

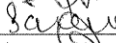



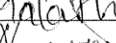
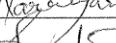
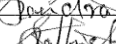
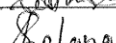
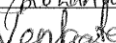


**SRI LAKSHMI NARAYANA INSTITUTE OF ALLIED HEALTH SCIENCE  
MEDICAL EDUCATIONAL PROJECT**

GENITIC DISORDER - VAC07/AHS/2020-18/05

<b>S.No.</b>	<b>Name of the Students</b>	<b>University Register Number</b>	<b>Signature</b>
1	AKSHAY SURESH	UAH1803178	
2	ANAGHA SUKUMARAN	UAH1803179	
3	APARNA REMESHAN	UAH1803180	
4	ASHIK.R	UAH1803182	
5	FEBA SUSAN ABRAHAM	UAH1803183	
6	GOKUL A	UAH1803184	
7	JIJI ELZA JOSE	UAH1803185	
8	MINNU MATHACHAN	UAH1803186	
9	MUHAMMED IRFAN.I	UAH1803187	
10	MURALIKRISHNAN.K	UAH1803188	
11	DONALD MORISON.S	UAH1805199	
12	GADDALA PRAVEEN	UAH1805200	
13	PRAVEEN.G	UAH1805201	
14	RAMANAN.V	UAH1805202	
15	SARATH P S	UAH1805203	
16	S.K.DHARSHINI	UAH1804193	
17	GAYATHRI.V	UAH1804194	
18	JAYABHAVANI.J	UAH1804195	
19	MALATHI.S	UAH1804196	
20	NARAYANADASS.M	UAH1804197	
21	SANDRA.S	UAH1803190	
22	SATHISHKUMAR.R	UAH1803191	
23	SHEHANAYI.M	UAH1803192	
24	VENKATESH.S	UAH1805204	
25	YAZHINI.P	UAH1805205	

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6	GOKUL A	UAH1803184	
7	JIJI ELZA JOSE	UAH1803185	
8	MINNU MATHACHAN	UAH1803186	
9	MUHAMMED IRFAN.I	UAH1803187	
10	MURALIKRISHNAN.K	UAH1803188	
11	DONALD MORISON.S	UAH1805199	
12	GADDALA PRAVEEN	UAH1805200	
13	PRAVEEN.G	UAH1805201	
14	RAMANAN.V	UAH1805202	
15	SARATH P S	UAH1805203	
16	S.K.DHARSHINI	UAH1804193	
17	GAYATHRI.V	UAH1804194	
18	JAYABHAVANI.J	UAH1804195	
19	MALATHI.S	UAH1804196	
20	NARAYANADASS.M	UAH1804197	
21	SANDRA.S	UAH1803190	
22	SATHISHKUMAR.R	UAH1803191	
23	SHEHANAYI.M	UAH1803192	
24	VENKATESH.S	UAH1805204	
25	YAZHINI.P	UAH1805205	