



# PHYLOGENETIC DISORDER OF MALE REPRODUCTIVE SYSTEM



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# PHYLOGENY

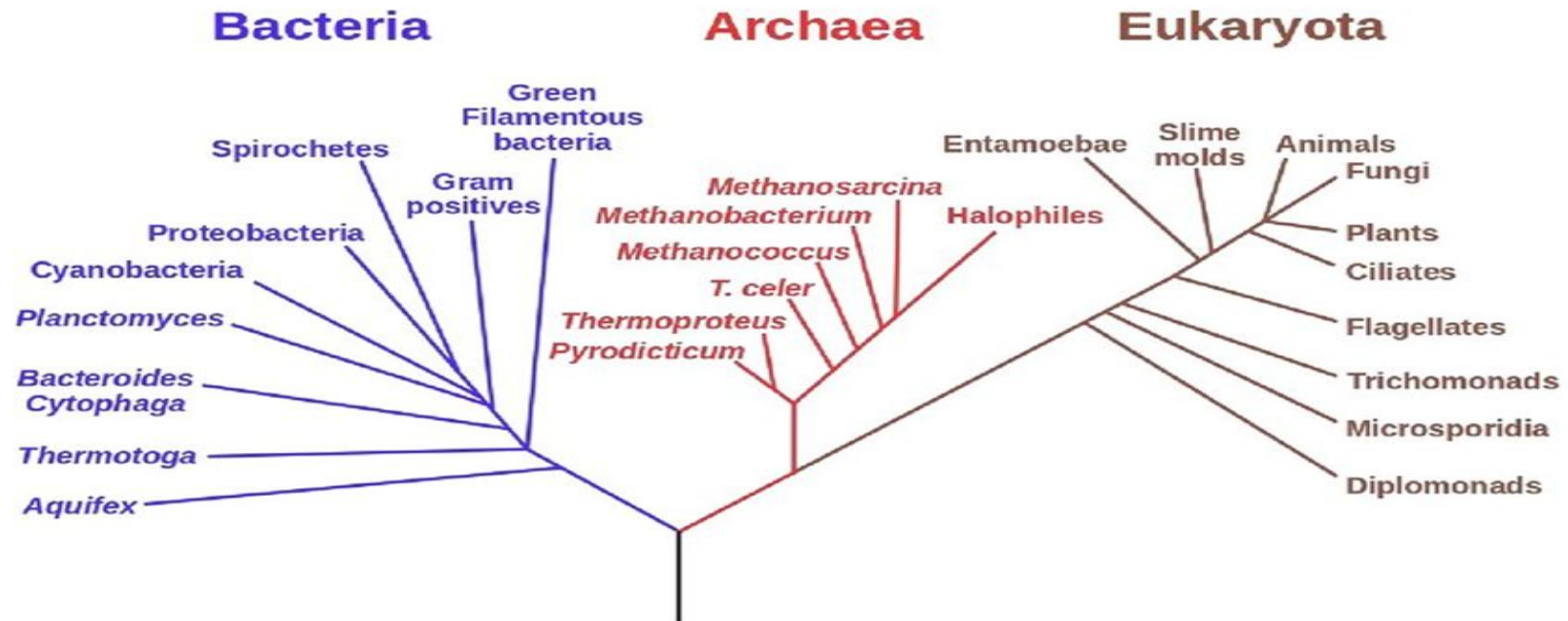
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- A phylogeny is a hypothetical relationship between groups of organisms being compared. A phylogeny is often depicted using a phylogenetic tree, such as the simple one below describing the evolutionary relationships between the great apes.
- A phylogeny is an expression of the evolutionary relationships of a group of animals.
- The purpose of a phylogeny is to illustrate which taxa (e.g., species, genera, etc).

# PHYLOGENY IN BIOLOGY

Applications of phylogeny. Phylogenies developed from molecular genetics have been particularly useful in conservation biology for identifying the evolutionary distinctiveness of endangered species, parentage in captive breeding programs, and levels of hybridization and inbreeding across species.

## Phylogenetic Tree of Life



# COMMON DISORDERS OF MALE REPRODUCTIVE SYSTEM

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- Prostate cancer
- Testicular cancer
- Enlarged prostate or BPH
- Prostatitis
- Erectile dysfunction
- Male infertility
- Testosterone deficiency
- Undescended testicle

# PHYLOGENETIC DISORDERS

Human genetic disorders give new insights into their etiology and evolution. Here, we have correlated a specific set of *Alu* repetitive DNA elements, known to be the basis of certain genetic defects, with their [phylogenetic](#) roots in primate evolution. From a differential distribution of *Alu* repeats among primate species, we identify the phylogenetic roots of three human genetic diseases involving the LPL, ApoB, and HPRT genes. The different phylogenetic age of these genetic disorders could explain the different susceptibility of various primate species to genetic diseases. Our results show that LPL deficiency is the oldest and should affect humans, apes, and monkeys. ApoB deficiency should affect humans and great apes, while a disorder in the HPRT gene (leading to the Lesch-Nyhan syndrome) is unique to human, chimpanzee, and gorilla. Similar results can be obtained for cancer. We submit that *de novo* transpositions of *Alu* elements, and saltatory appearances of *Alu*-mediated genetic disorders, represent singularities, places where behavior changes suddenly. *Alu*'s propensity to spread, not only increased the regulatory and developmental complexity of the primate genome, it also increased its instability and susceptibility to genetic defects and cancer. The dynamic spread not only provided markers of primate phylogeny, it must have actively shaped the course of that phylogeny.

# PHYLOGENETIC DISORDERS

Hypospadias

Epispadias

Cryptorchidism [undescended testicle]

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# CRYPTORCHIDISM

Cryptorchidism, also called cryptorchism, undescended testes, or undescended testicles, disorder in which one or both of the testes do not descend spontaneously to the usual position in the scrotum. (The testes normally descend around the time of the male infant's birth.)

## **CAUSES:**

Prune belly syndrome

Fanconi anaemia

BRESHECK syndrome

Sutherland-Haan syndrome

Opitz-Frias syndrome

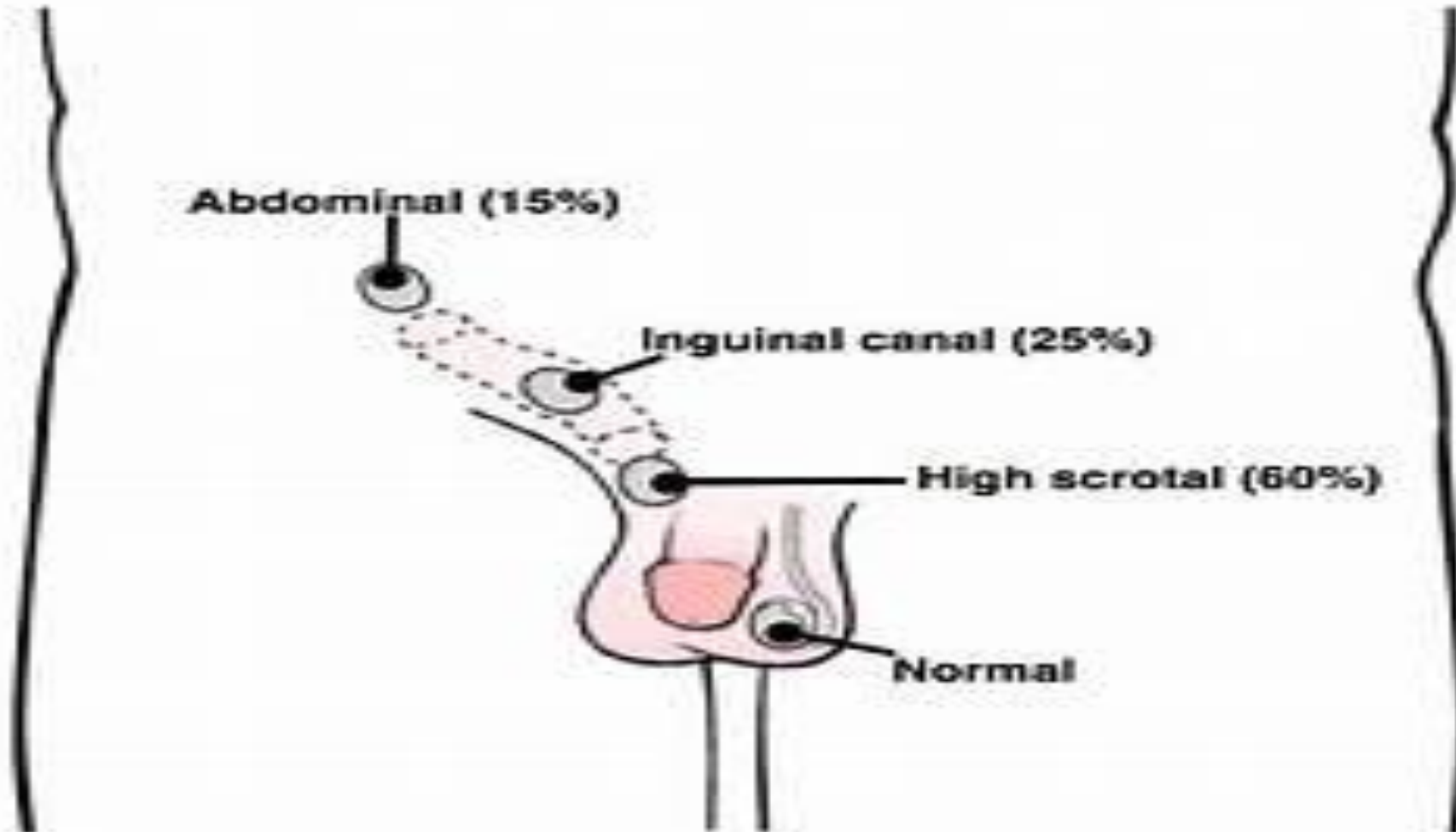
CHARGE syndrome

Fetal akinesia-hypokinesia sequence.

# Cryptorchidism

(Undescended testicle)

Most common congenital abnormality of the GU tract





# SOME SIGNS AND SYMPTOMS OF THE CRYPTORCHIDISM

- Misplaced testes hidden in the abdomen
- Missing testicle in male newborn
- Infertility
- Cancer risk
- Some psychological consequence etc..,

# TREATMENT

## **Time of treatment:**

Current guidelines recommend to plan treatment of cryptorchidism 6 month after birth. Treatment should be finished with the end of the first year of life.

## **Goals of Therapy:**

Preserving fertility

Reducing tumor risk

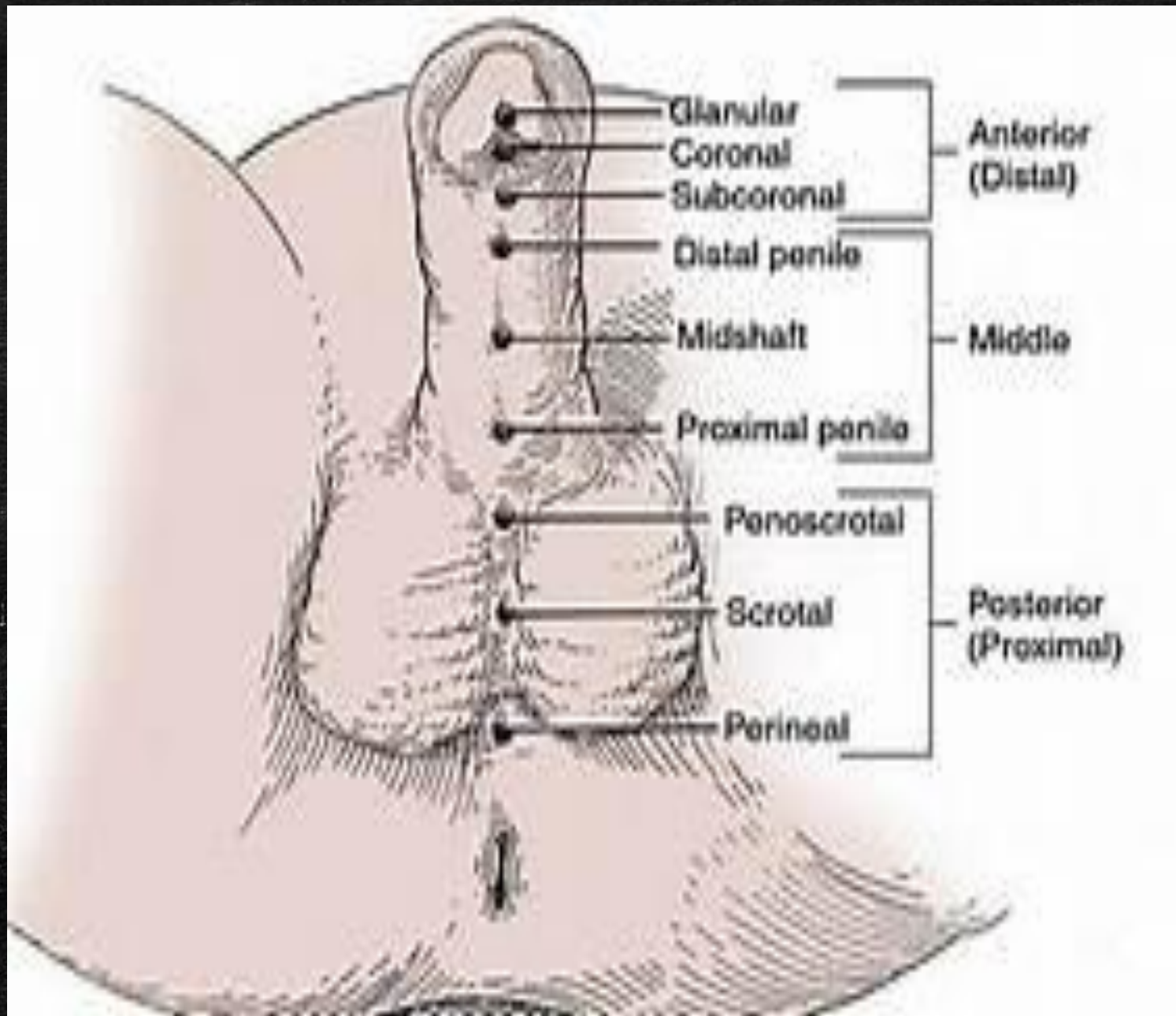
Improving early detection of testicular cancer

# HYPOSPADIAS

Hypospadias (hi-poe-SPAY-dee-us) is a birth defect (congenital condition) in which the opening of the urethra is on the underside of the penis instead of at the tip. The urethra is the tube through which urine drains from your bladder and exits your body. Hypospadias is common and doesn't cause difficulty in caring for your infant.

## **CAUSES:**

Hypospadias results when a malfunction occurs in the action of these hormones, causing the urethra to develop abnormally. In most cases, the exact cause of hypospadias is unknown. Sometimes, hypospadias is genetic, but environment also may play a role.



# SIGNS, SYMPTOMS AND TREATMENT

Signs and symptoms of hypospadias may include:

Opening of the urethra at a location other than the tip of the penis  
Downward curve of the penis (chordee)  
Hooded appearance of the penis because only the top half of the penis is covered by foreskin  
Abnormal spraying during urination

Treatment of hypospadias:

The treatment for hypospadias is surgical repair. Hypospadias is generally repaired for functional and cosmetic reasons. The more proximally ectopic the position of the urethral meatus, the more likely the urinary stream is to be deflected downward, which may necessitate urination in a seated position. Any element of ventral curvature (chordee) can further deflect the urinary stream. Fertility may be affected. The abnormal deflection of ejaculate may preclude effective insemination, and significant chordee can preclude vaginal insertion of the penis or can be associated with inherently painful erections.

# EPISPADIAS

Epispadias is an uncommon and partial form of a spectrum of failures of abdominal and pelvic fusion in the first months of embryogenesis known as the exstrophy - epispadias complex. While epispadias is inherent in all cases of exstrophy it can also, much less frequently, appear in isolation as the least severe form of the complex spectrum.

## CAUSES:

Family history. This condition is more common in infants with a family history of hypospadias.

Genetics.

Maternal age over 35 and weight. ...

Fertility treatments: Women who used assisted reproductive technology to help with pregnancy had a higher risk of having a baby with hypospadias 55).

Exposure to certain substances during pregnancy. ...



# SYMPTOMS AND TREATMENT

## SYMPTOMS:

Most cases of epispadias are diagnosed at birth during physical examination of the newborn. In mild cases of epispadias, the condition can go unnoticed until parents note urine leaks after potty training.

## TREATMENT:

The main goals of epispadias treatment for boys are to:

- Make sure the penis works

- Fix bends in the penis (dorsal bend and chordee)

- Make the penis a good length

- Make the penis look normal

If the bladder and bladder neck are affected, surgery will be needed to make sure the child can control his urine. Surgery might also be needed to preserve fertility.

There are two popular types of surgery. Both provide a normal-looking penis that works properly. The surgery is often done while the child is a baby. The type chosen depends on:

- The type of epispadias

- Surgeon experience

- Surgeon preference

Sometimes, boys with the exstrophy-epispadias complex are born with a very underdeveloped penis. In these cases, surgery requires more skill and experience.

You may ask your urologist for the name of a specialist.



