

# Variability in neonatal genitalia

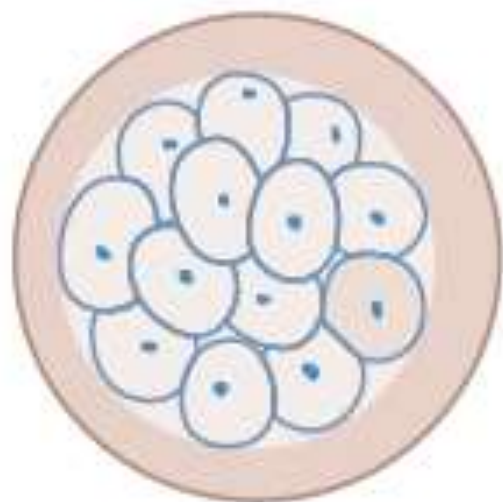




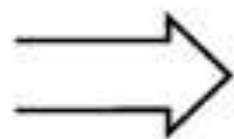




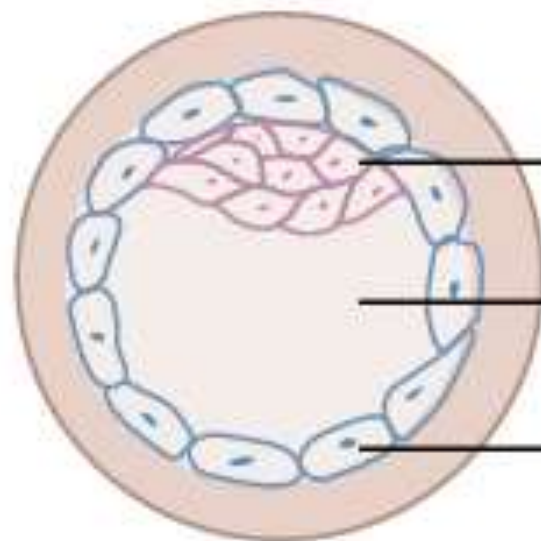
**Morula**



16 or more cells



**Blastocyst**



Embryoblast

Blastocoele

Trophoblast

Day 6

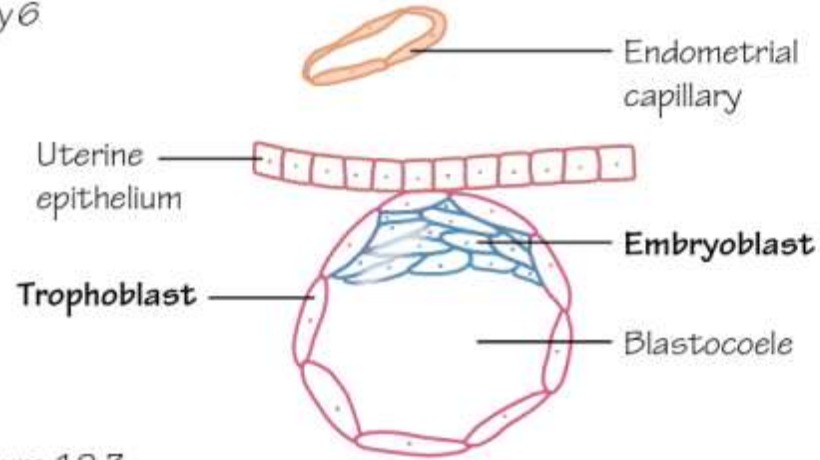
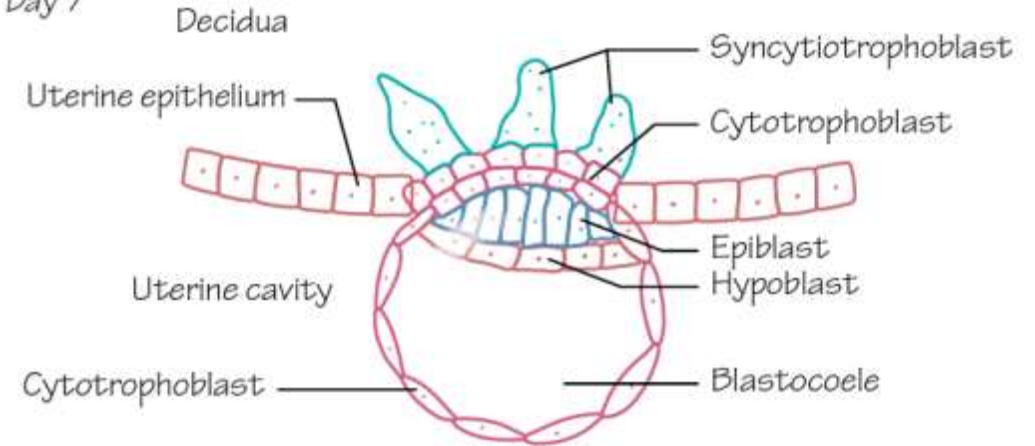


Figure 12.3

Implantation of the blastocyst into the uterine wall begins

Day 7



Day 9

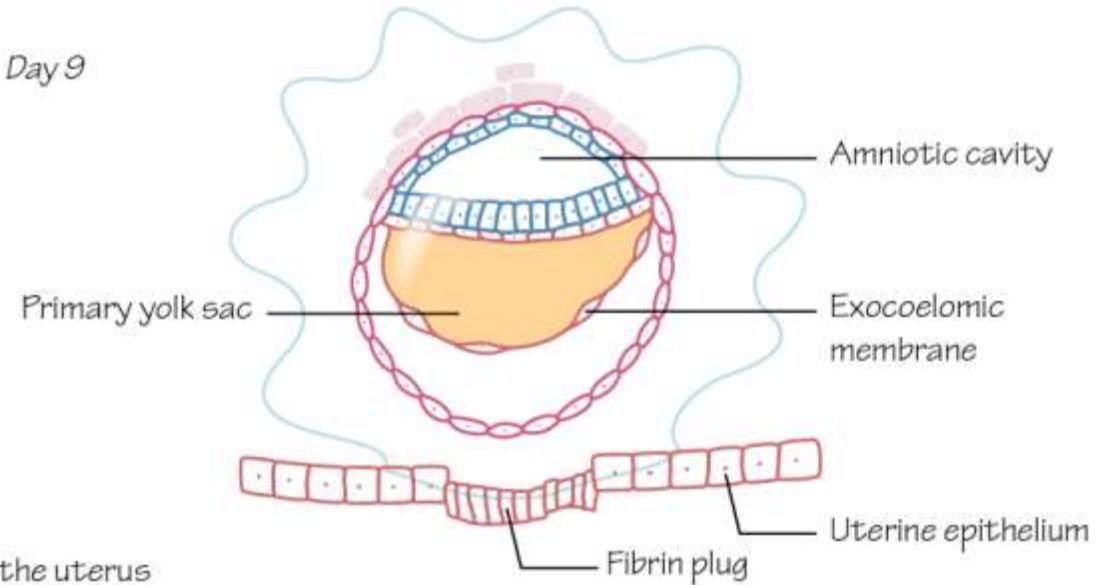


Figure 12.4

Implantation continues and the blastocyst becomes embedded within the wall of the uterus

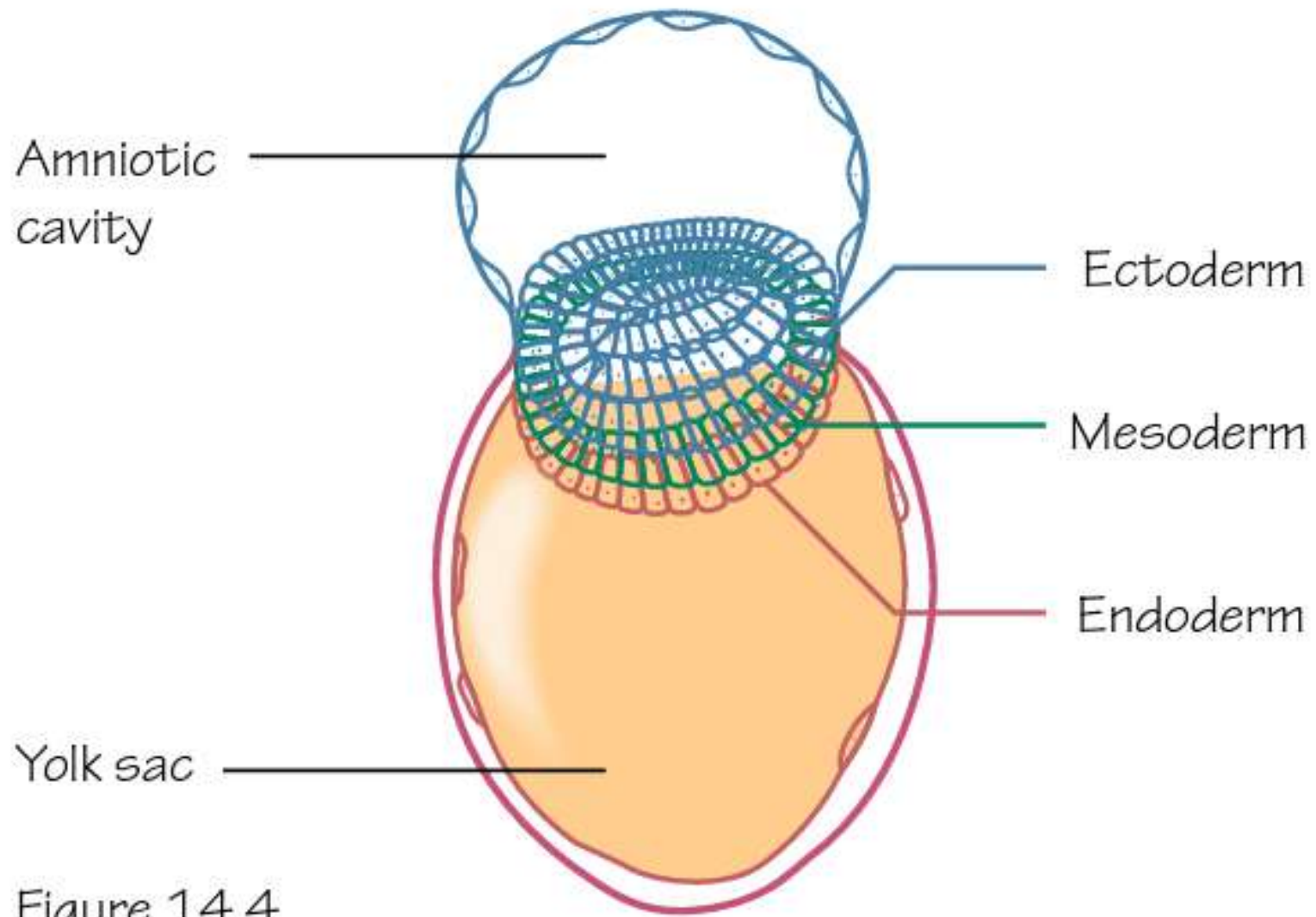
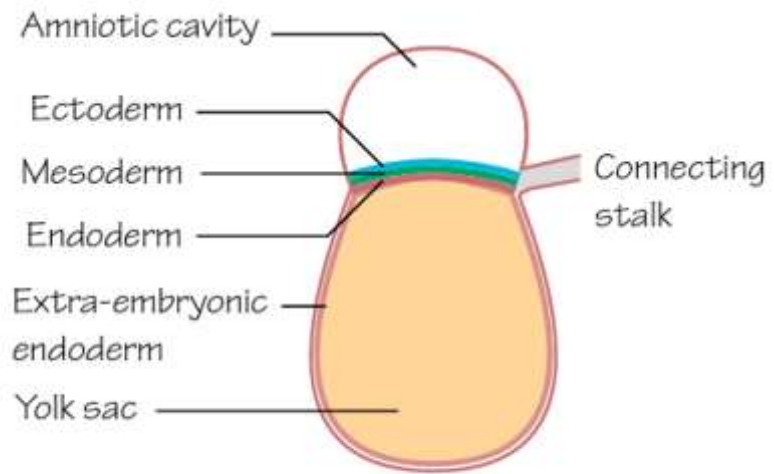
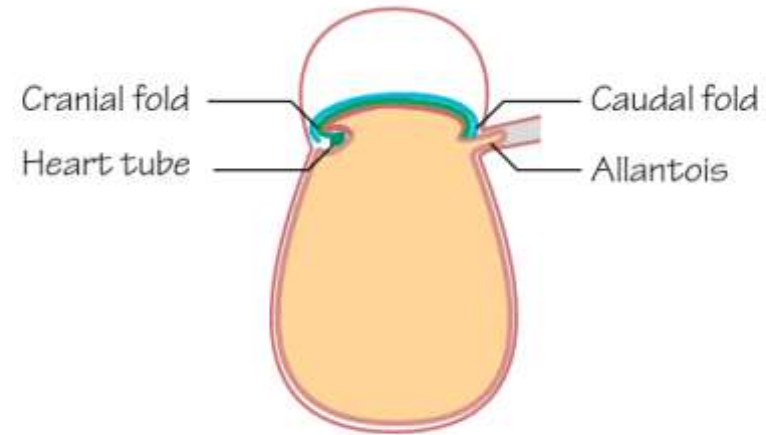


Figure 14.4

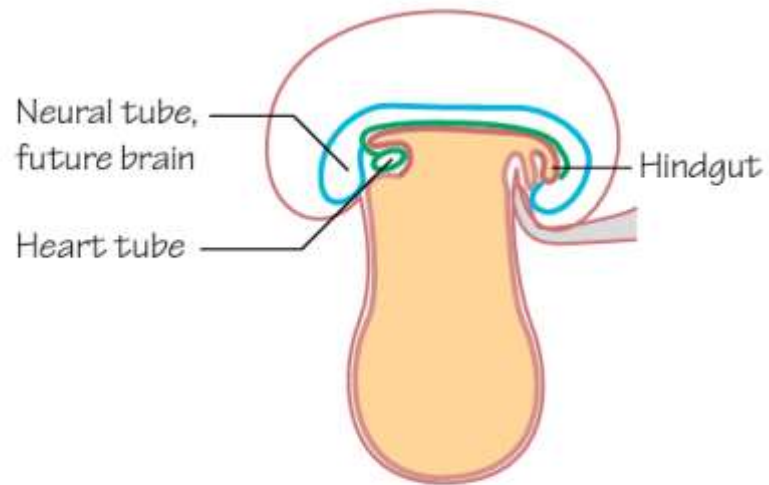
The three layers of the trilaminar germ disc



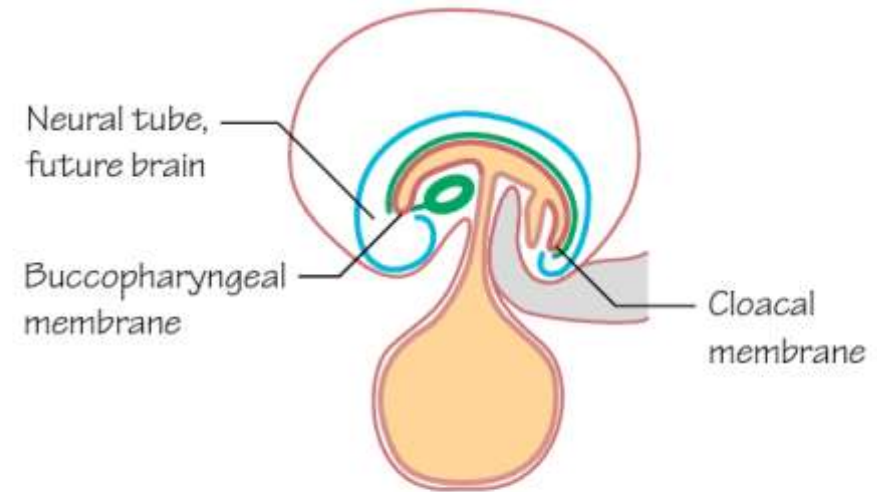
Day 18



Day 21



Day 24



Day 28

Figure 20.1

The cranial (head) and caudal (tail) folding of the embryonic disk





6 weeks

Allantois

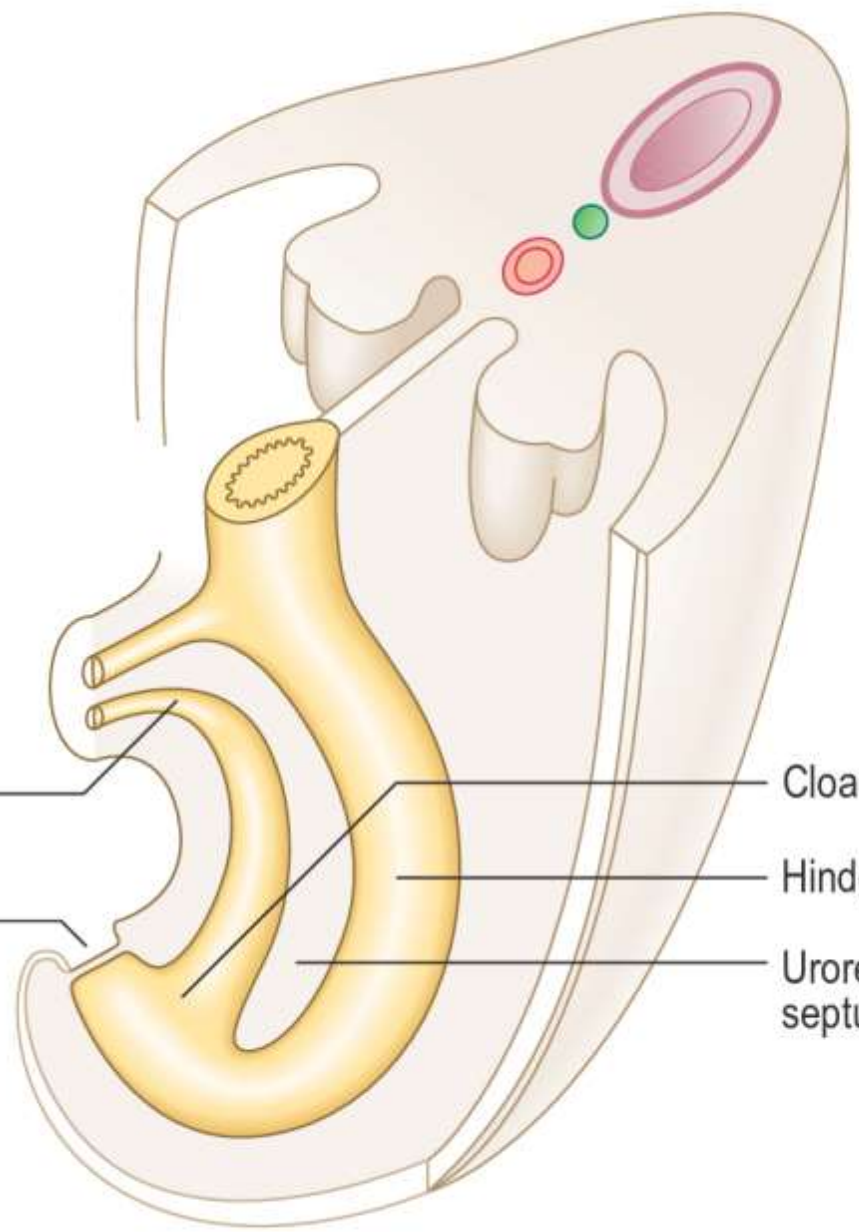
Cloaca

Hindgut

Cloacal membrane

Urorectal septum

A





7 weeks

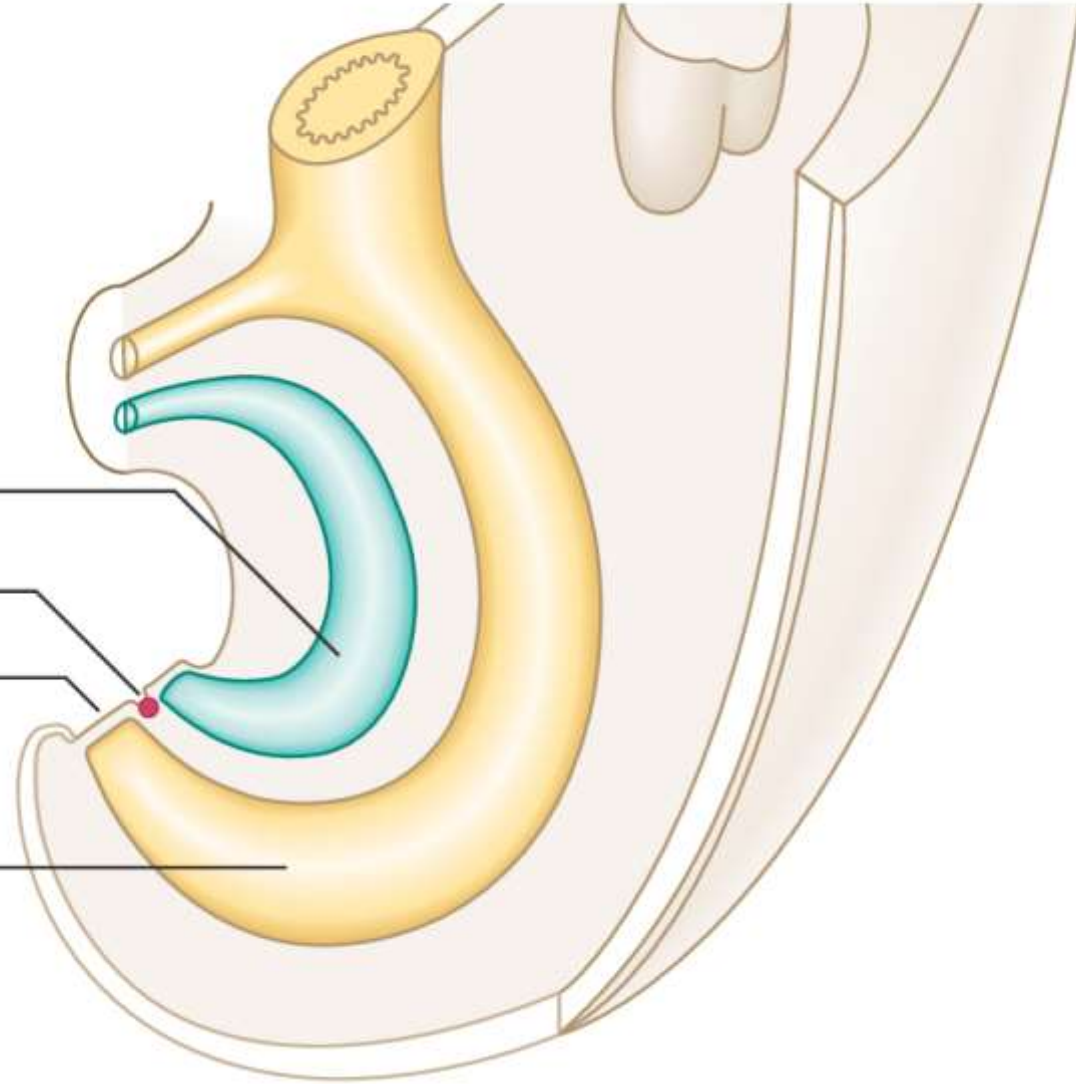
Urogenital sinus

Perineal body

Anal membrane

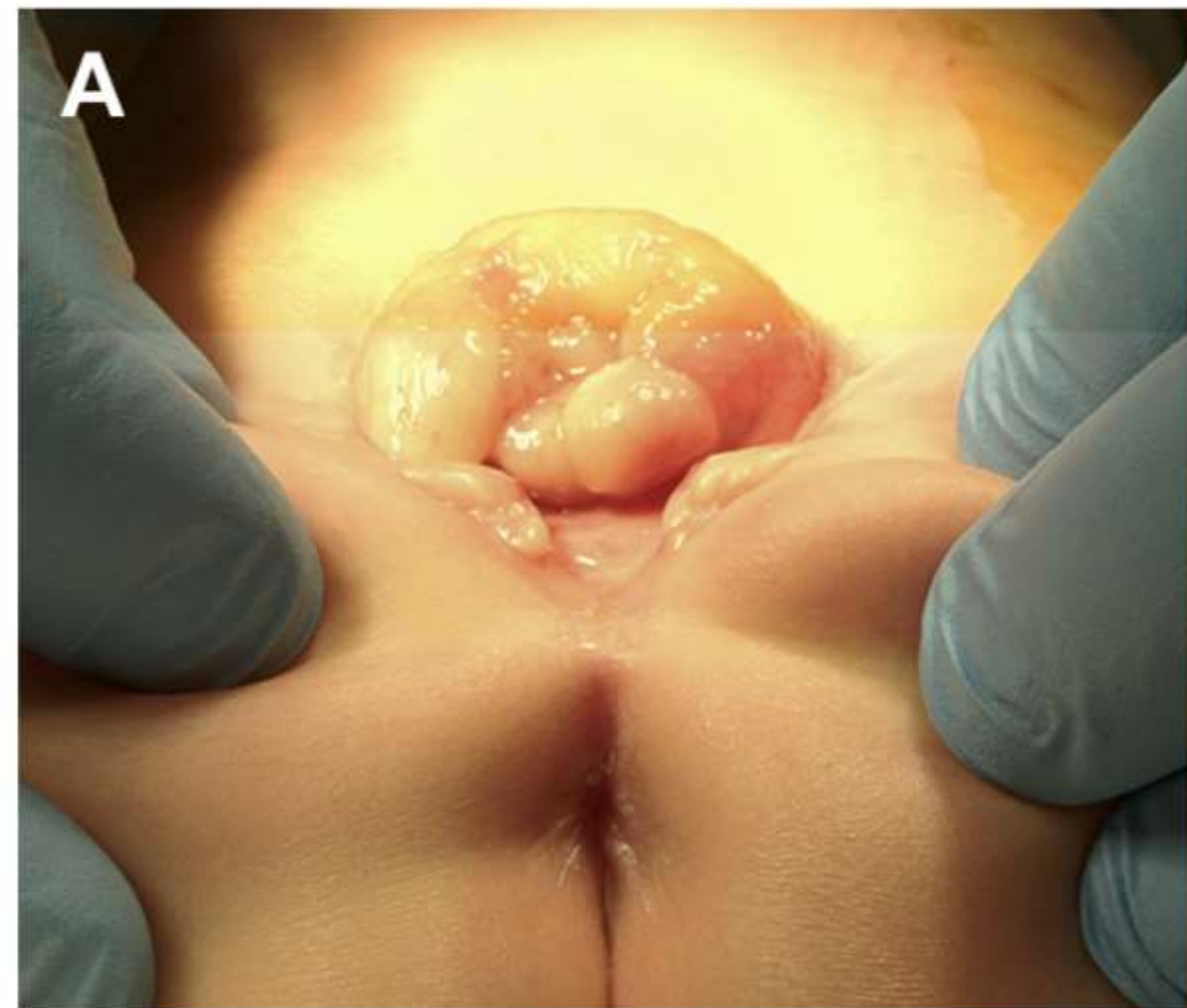
Rectum

**B**

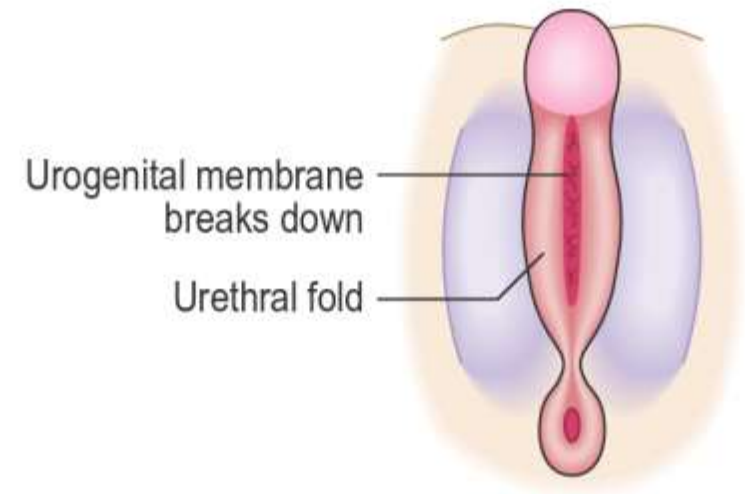
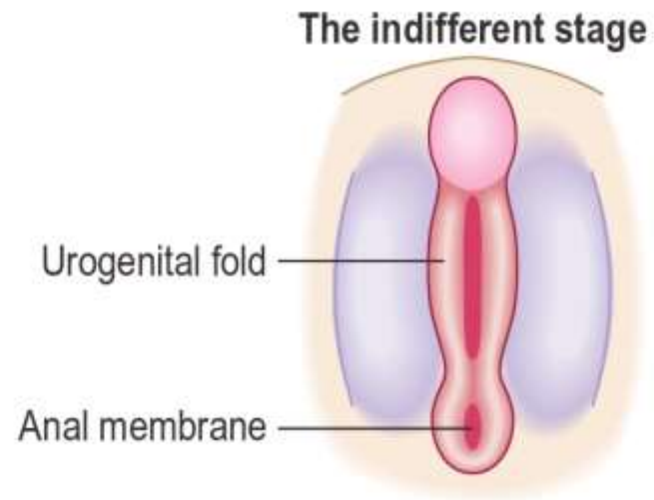
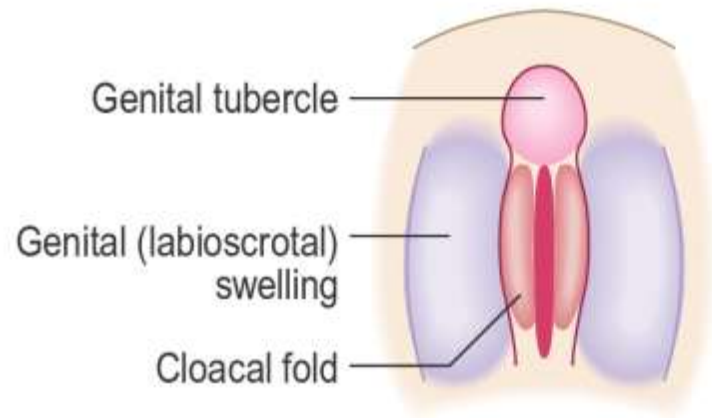


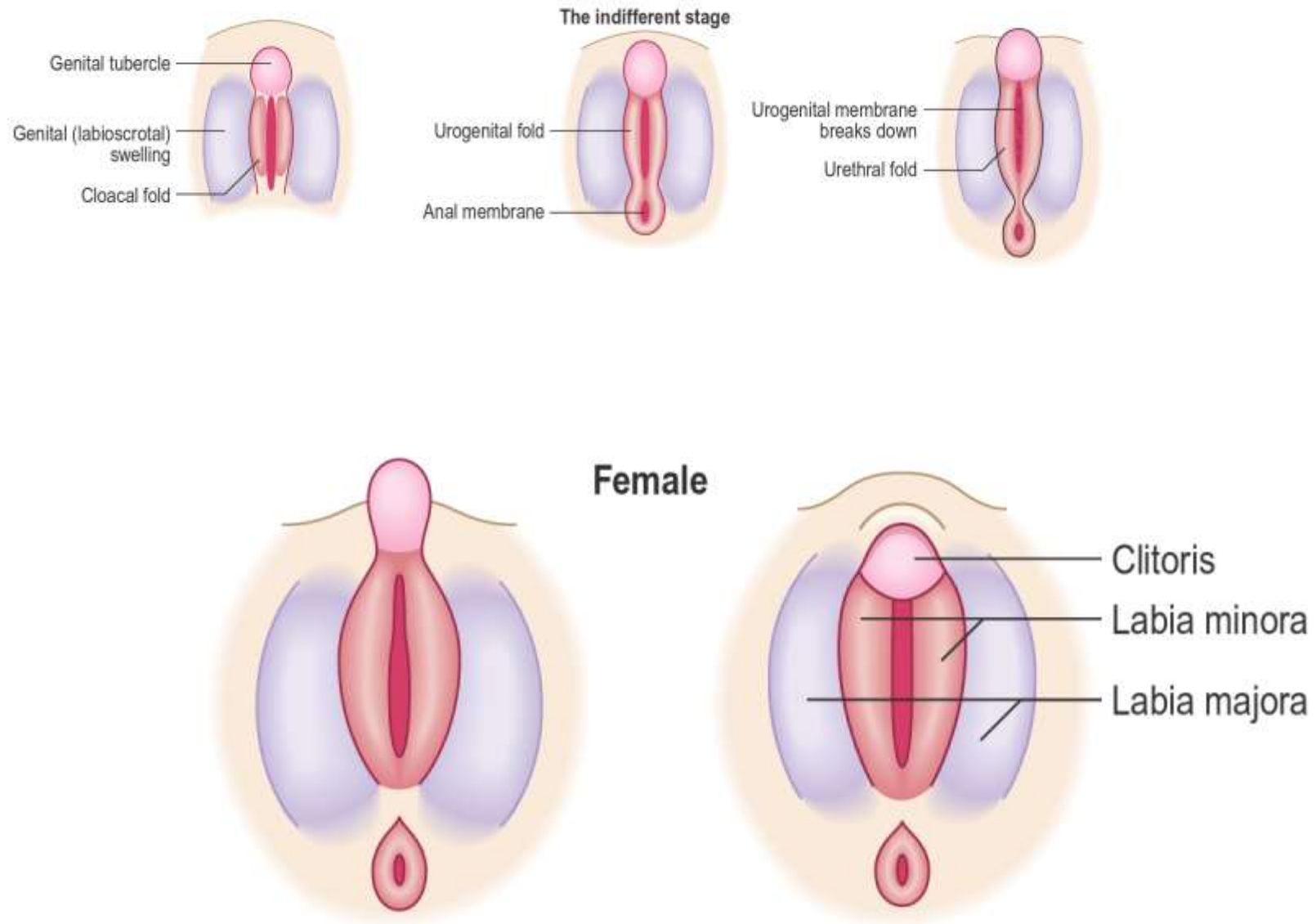


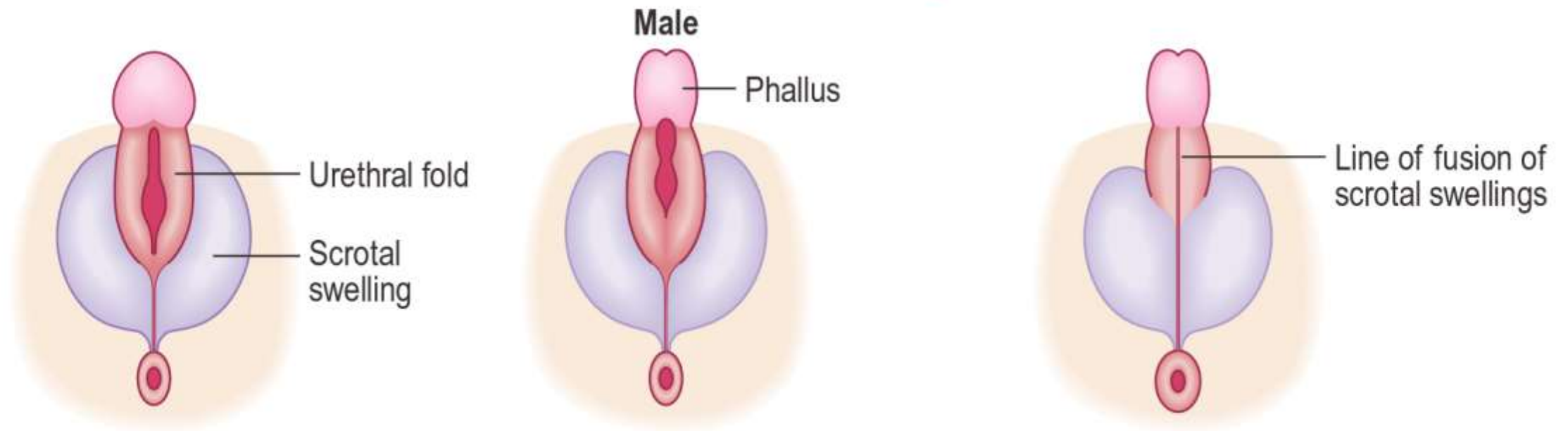
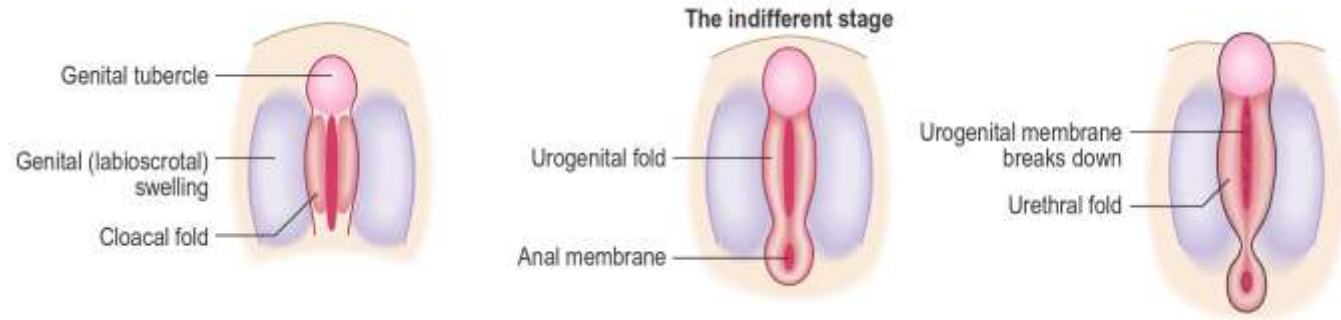


**A****B**















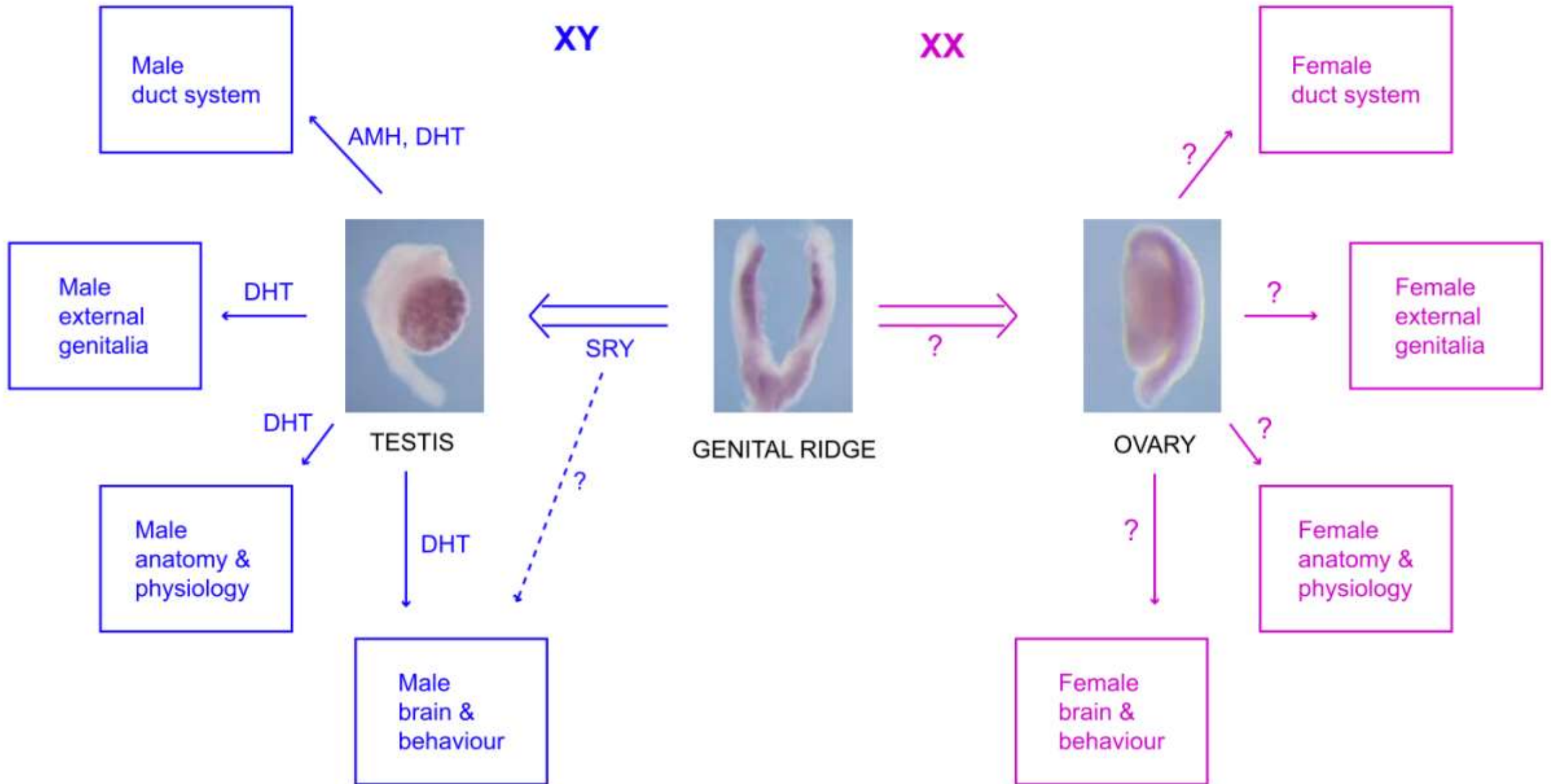


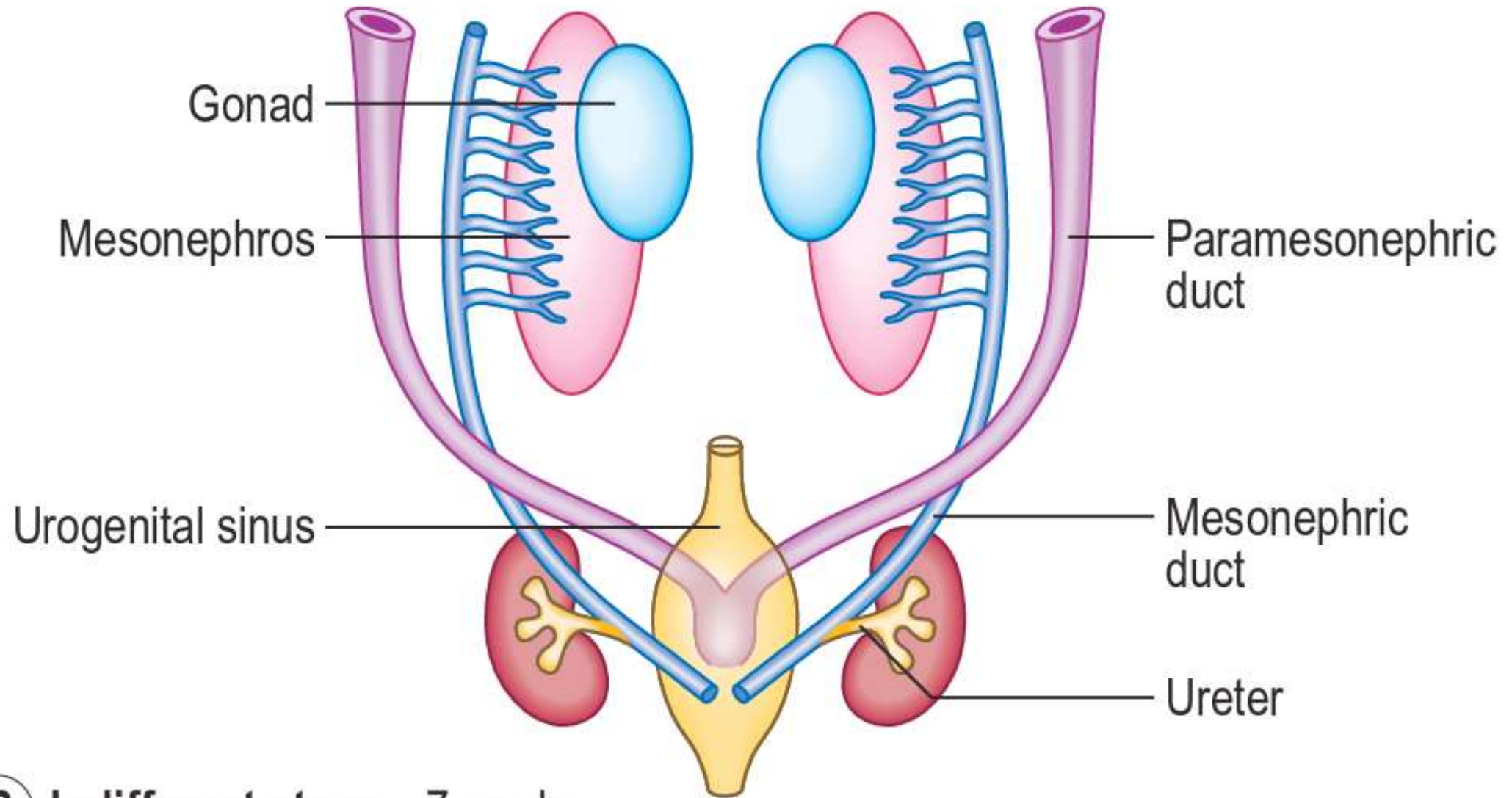




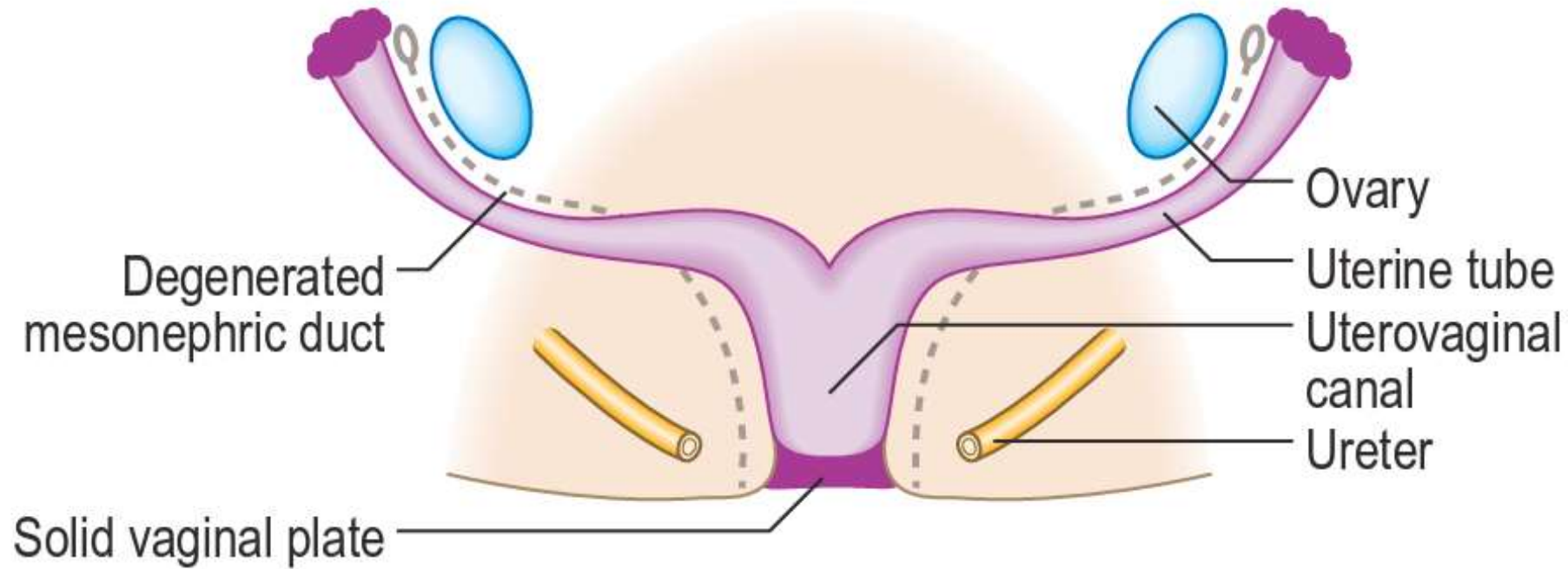
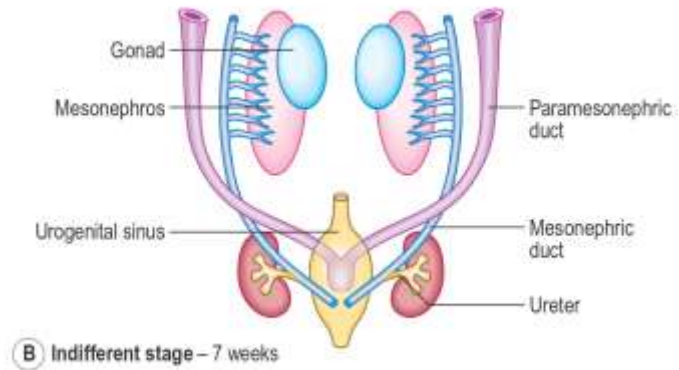
# Gonadal development



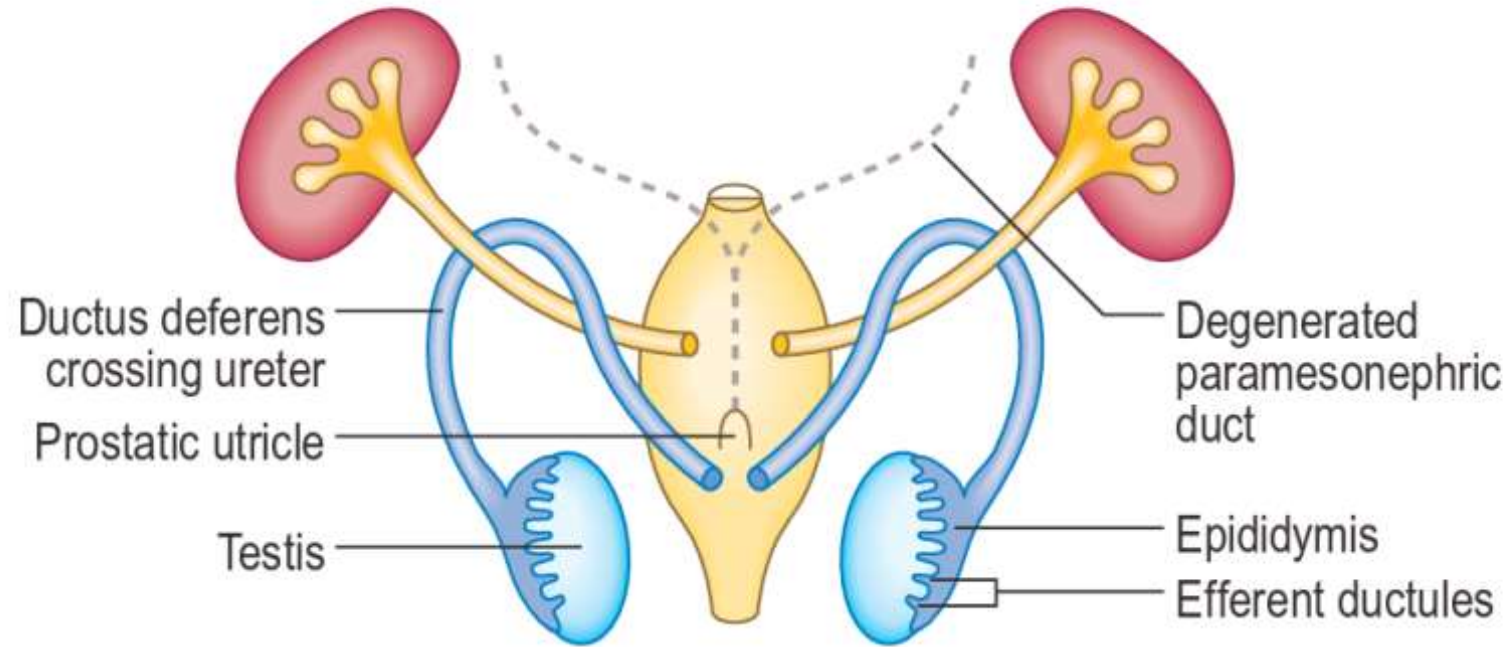
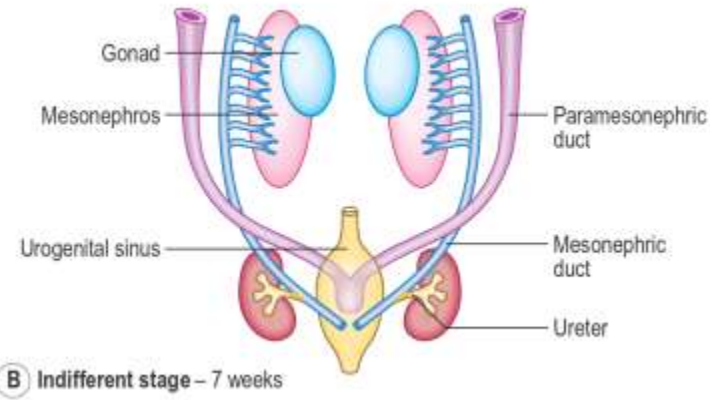




**B** Indifferent stage – 7 weeks



**D Female genital system – newborn**



**C Male genital system – newborn**



This is dependent on having  
normal gonads

# Examples of conditions causing gonadal dysfunction

- Turner's syndrome
- 45 X

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- Turner's syndrome
- 45 X
- Klinefelter's syndrome
- 47 XXY

# Examples of conditions causing gonadal dysfunction

- Turner's syndrome
  - Klinefelter's syndrome
  - Complete gonadal dysgenesis
- 45 X
  - 47 XXY
  - 46 XX
  - 46 XY (Swyer syndrome)



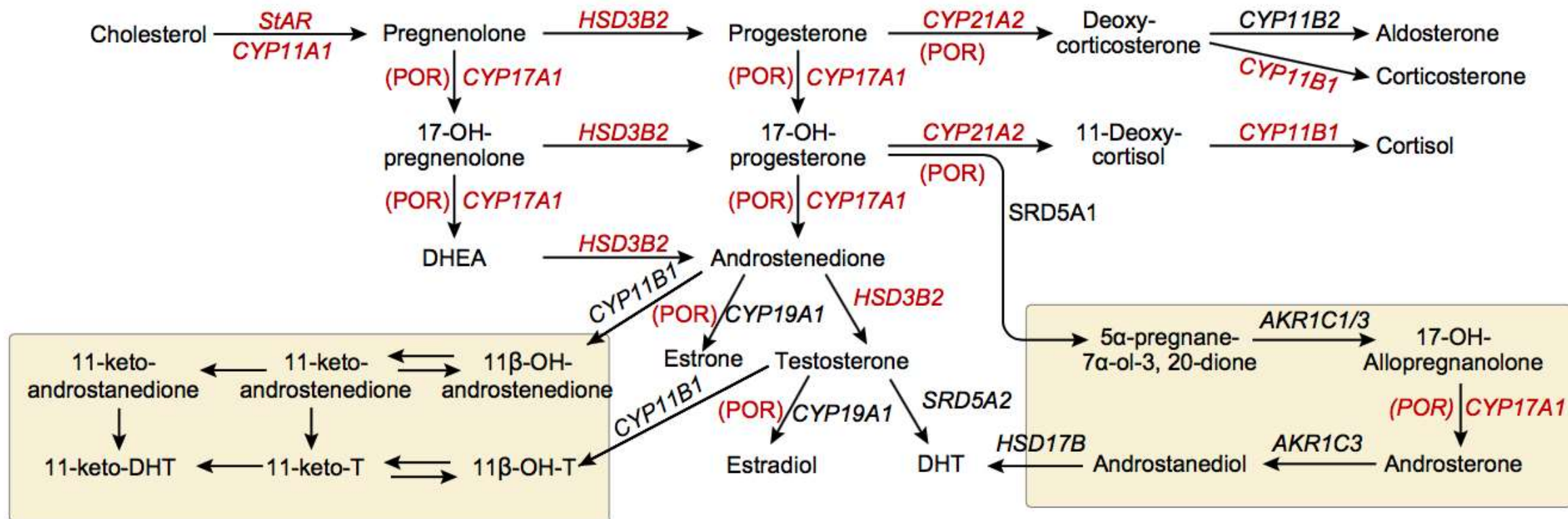
# Examples of conditions causing gonadal dysfunction

- Turner's syndrome
  - Klinefelter's syndrome
  - Complete gonadal dysgenesis
  - Partial gonadal dysgenesis
- 45 X
  - 47 XXY
  - 46 XX
  - 46 XY (Swyer syndrome)
  - 45 X / 46 XY
  - 46 XY

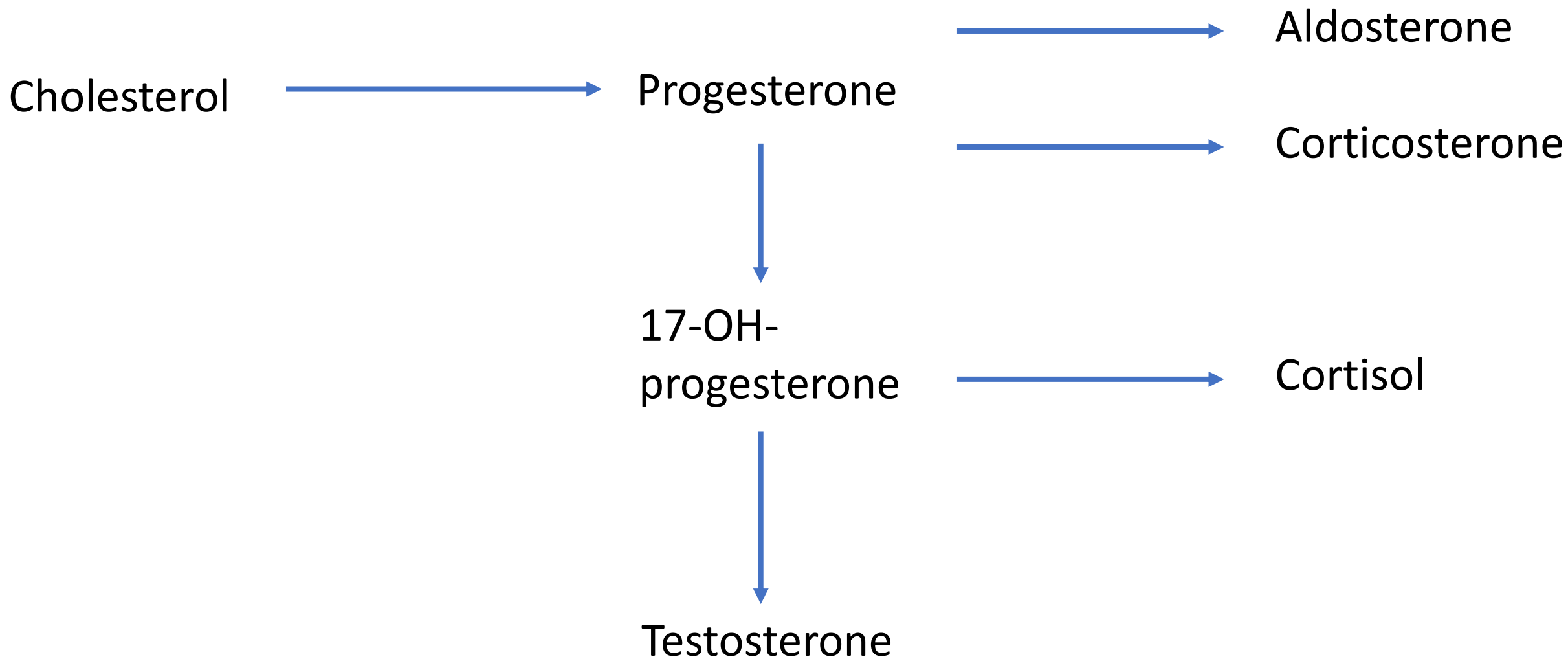
But even if the gonads are  
normal....



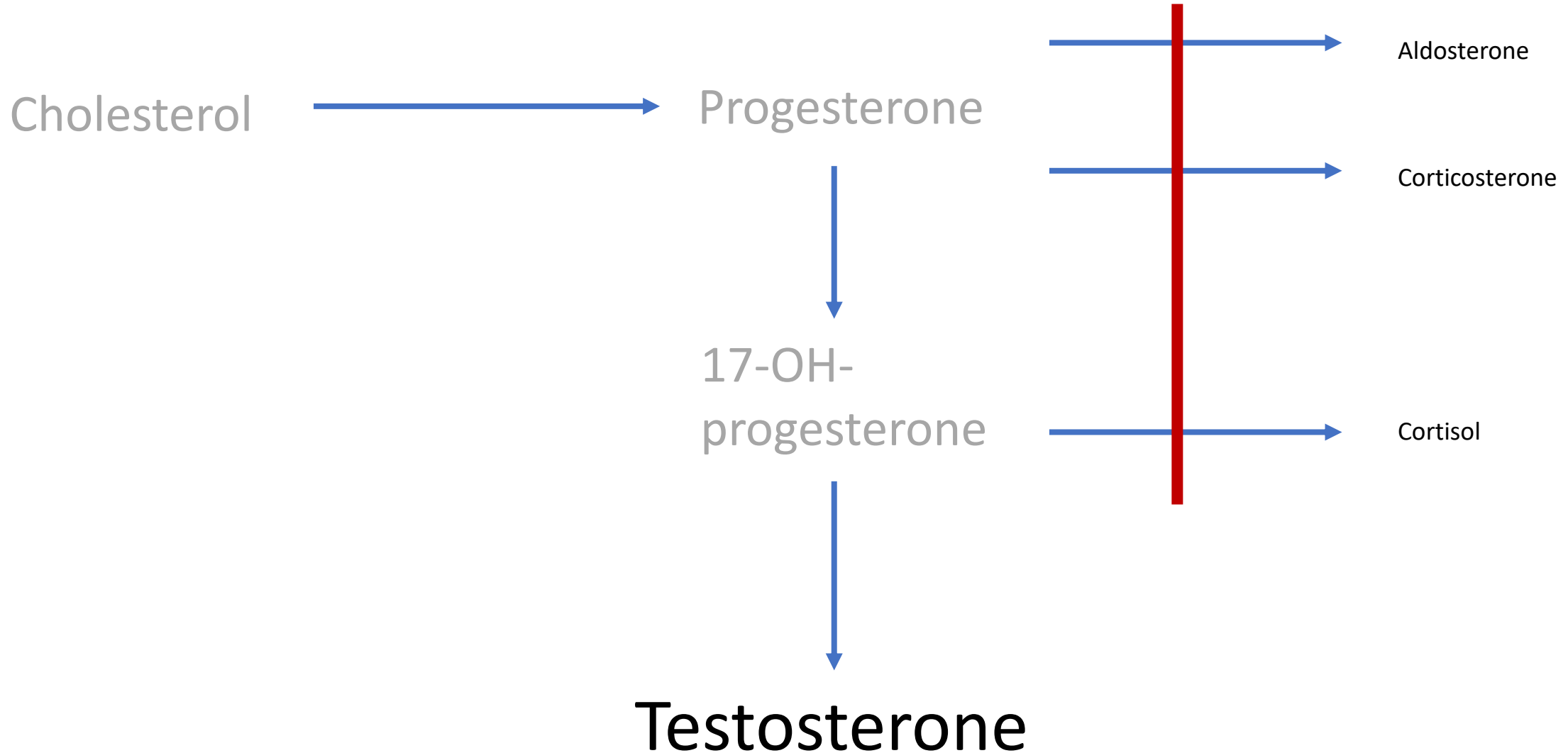
- Luigi De Crecchio 1865
- Described at autopsy
  - External male
  - Internal female organs
  - Enlarged adrenals







## Complete or partial 21-hydroxylase deficiency





0 – 4%

Mortality from salt wasting  
in congenital adrenal  
hyperplasia



# Evaluation of the Dutch neonatal screening for congenital adrenal hyperplasia

Annelieke A A van der Linde,<sup>1,2</sup> Yvonne Schönbeck,<sup>3</sup> Hetty J van der Kamp,<sup>4</sup> Erica L T van den Akker,<sup>5</sup> Mirjam E van Albada,<sup>6</sup> Anita Boelen,<sup>7</sup> Martijn J J Finken,<sup>8</sup> Sabine E Hannema,<sup>5,9</sup> Gera Hoorweg-Nijman,<sup>10</sup> Roelof J Odink,<sup>11</sup> Peter C J I Schielen,<sup>12</sup> Saartje Straetemans,<sup>13</sup> Paul S van Trotsenburg,<sup>14</sup> Hedi L Claahsen-van der Grinten,<sup>1</sup> Paul H Verkerk<sup>3</sup>

Of those detected clinically.....

- Almost all females identified by ambiguous genitalia

Of those detected clinically.....

- None of the males identified by ambiguous genitalia
- One (10%) had a pigmented scrotum

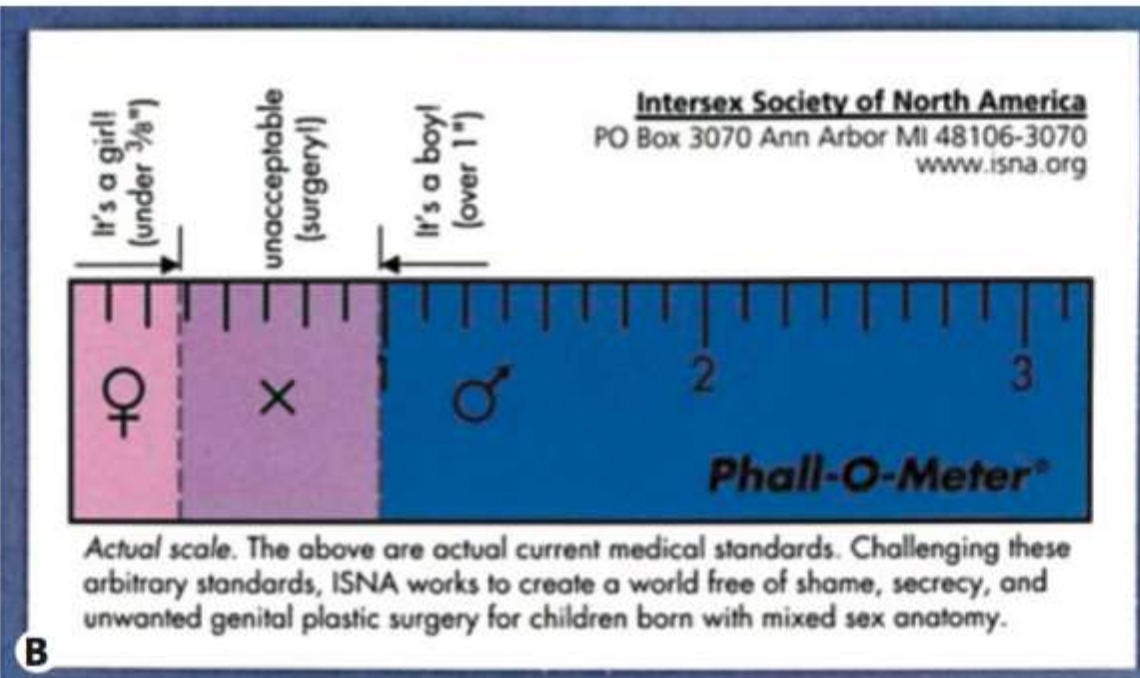
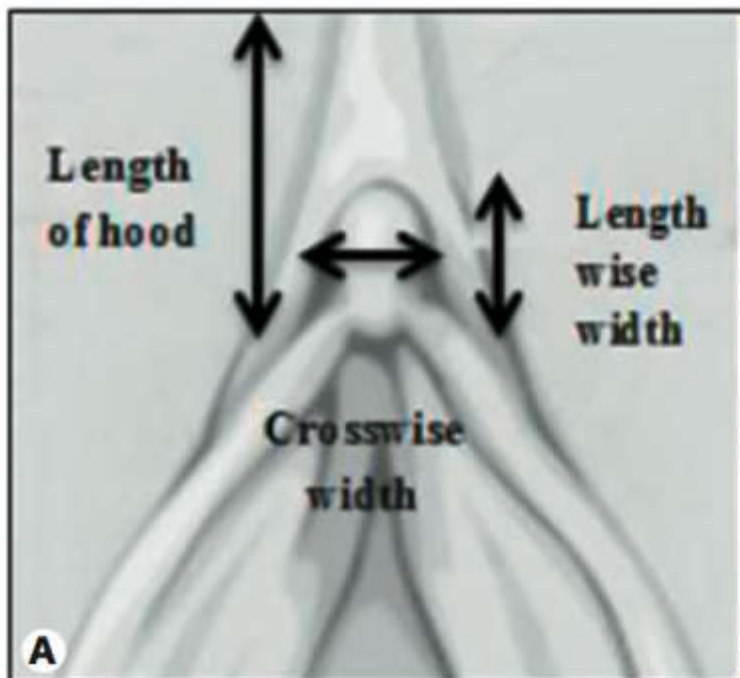
	Male	Female
Detected by screening	57	14
Detected clinically	10	23

When are subtle differences  
significant?



# Females

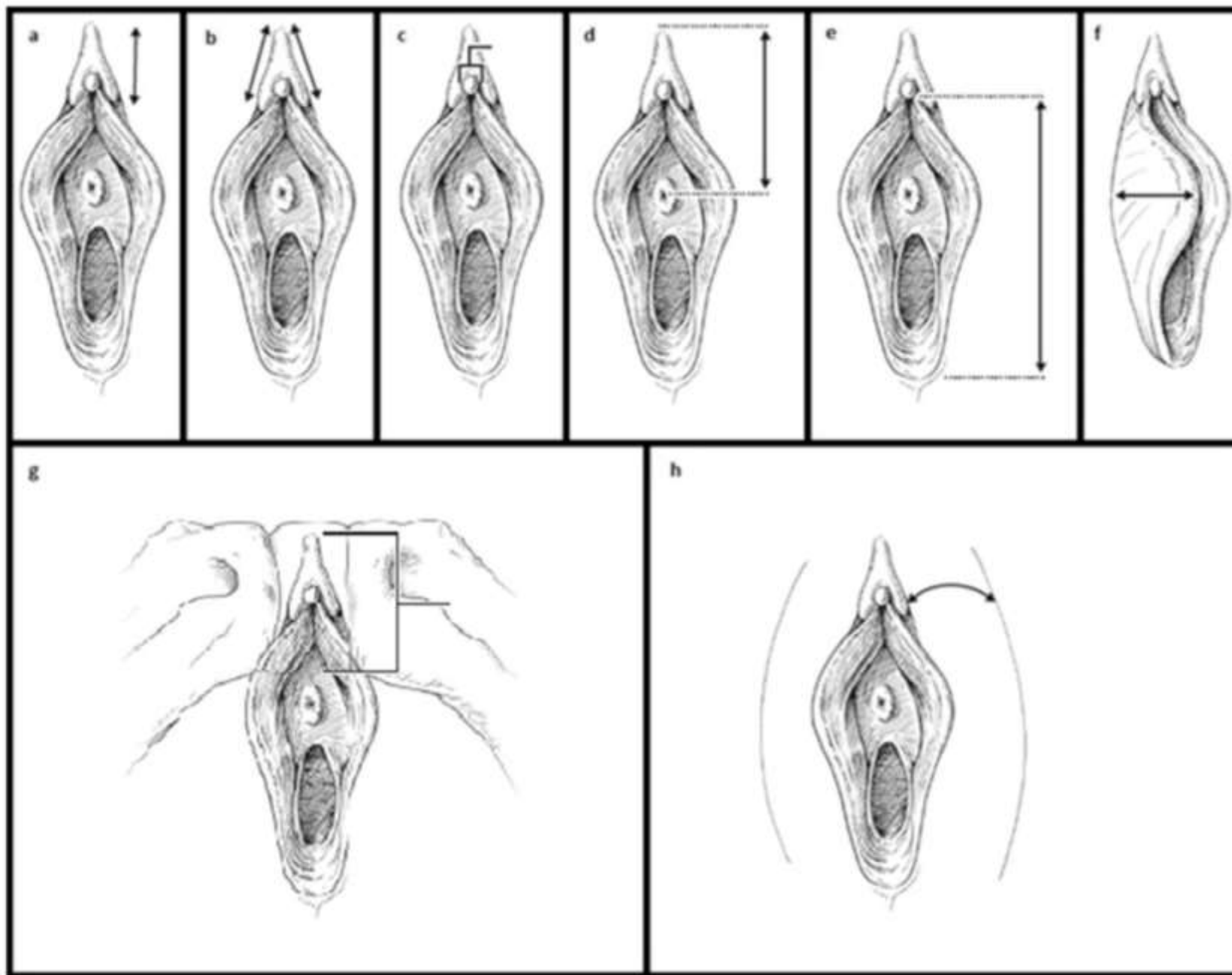
- Inguinal hernia
- Gonad palpable in the labia
- Labial rugosity or pigmentation
- Labial fusion
- Cliteromegaly



# Study of clitoral hood anatomy in the pediatric population



K.E. Brodie <sup>a</sup>, E.C. Grantham <sup>a</sup>, P.S. Huguelet <sup>b</sup>, B.T. Caldwell <sup>a</sup>,  
N.J. Westfall <sup>a</sup>, D.T. Wilcox <sup>a</sup>



Mean measurement in mm (SD)	Age category
	0–3 years ( $n = 18$ )
Length of clitoral hood	8.7 (3.9) Range: 3–17
Length of sides of clitoral hood (total)	21.6 (4.9) Range: 12–34
Clitoral diameter	2.5 (0.8) Range: 2–5
Clitoral hood to pubic symphysis	14.7 (6.1) Range: 4–23
Clitoral hood to labia majora	2.3 (1.6) Range: 0–5
Clitoral hood to urethral orifice	12.4 (5.2) Range: 4–22
Length of labia minora	9.4 (3.7) Range: 3–18
Depth of labia minora	3.5 (1.1) Range: 2–5

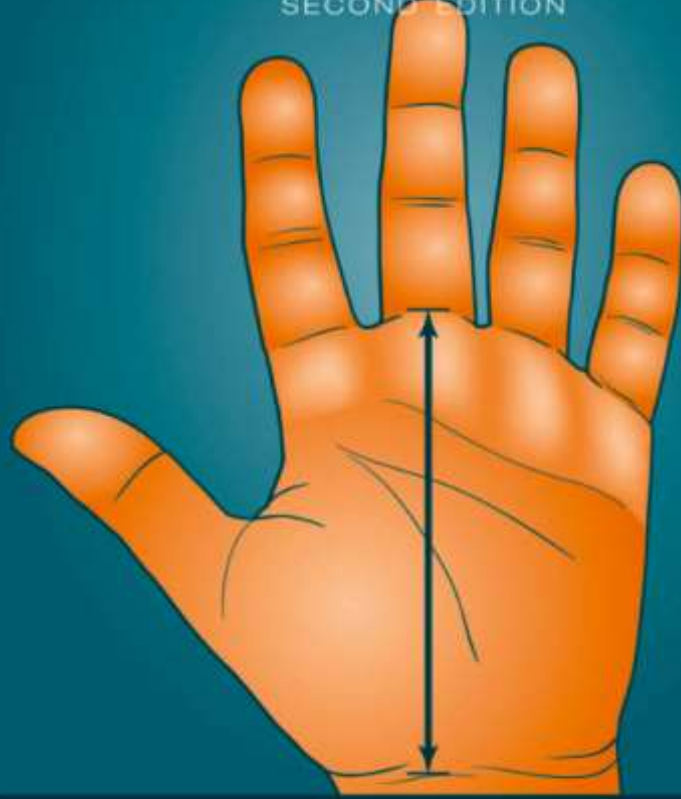


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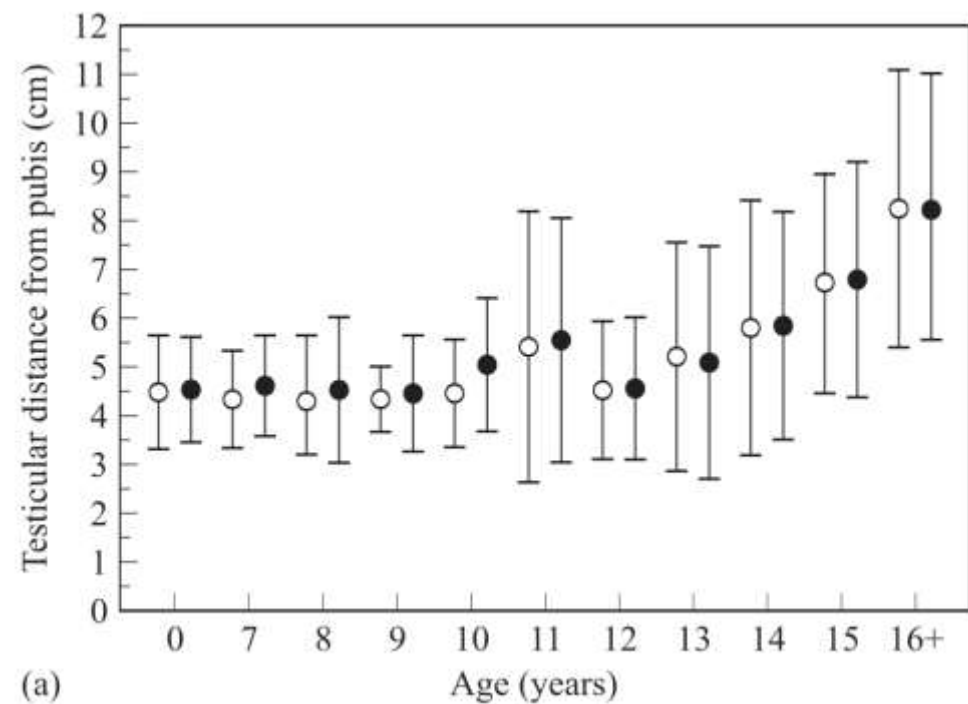
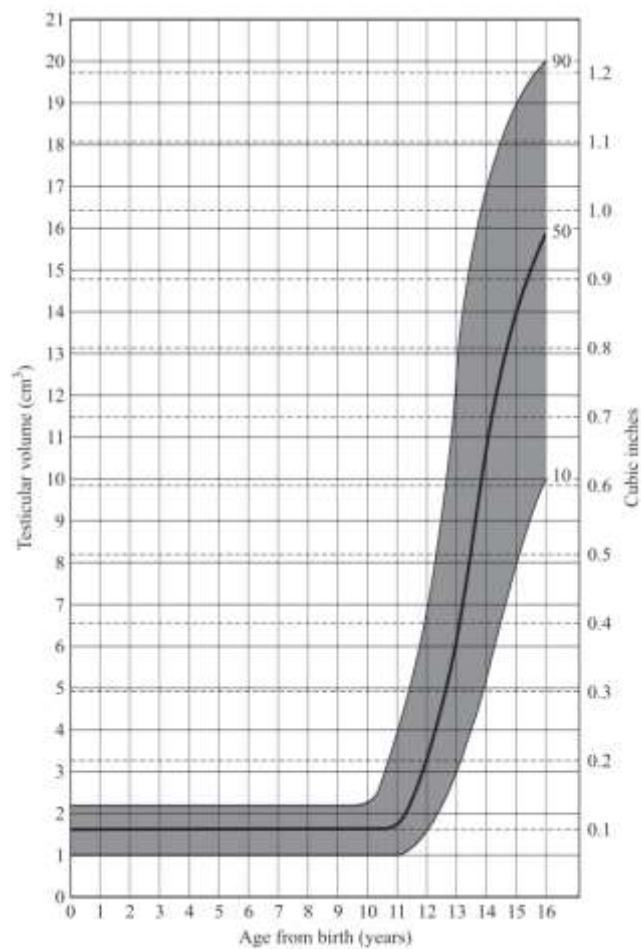
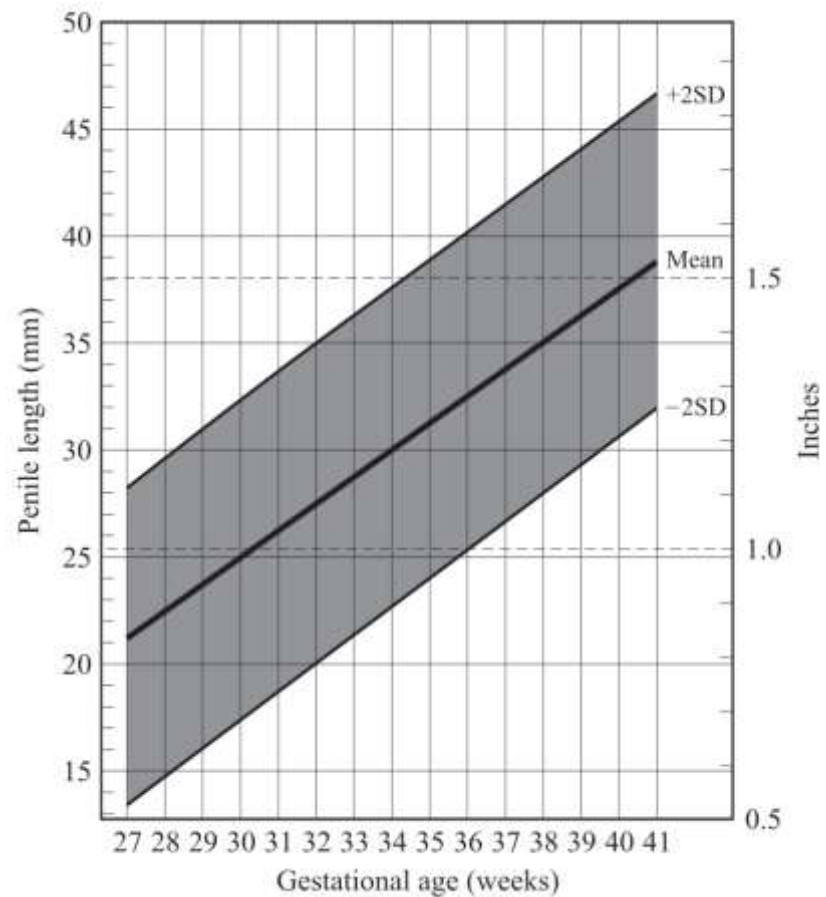
Males?

# HANDBOOK OF PHYSICAL MEASUREMENTS

SECOND EDITION



JUDITH G. HALL • JUDITH E. ALLANSON  
KAREN W. GRIPP • ANNE M. SLAVOTINEK



# CHROMOSOMAL ANOMALIES IN CRYPTORCHIDISM AND HYPOSPADIAS

MARTA MORENO-GARCÍA AND EMILIA BARREIRO MIRANDA

The Journal of Urology (2002): 168: 2170-2172



1.84%

Incidence of intersexuality in patients  
with isolated cryptorchidism

# **Increased Occurrence of Disorders of Sex Development, Prematurity and Intrauterine Growth Restriction in Children with Proximal Hypospadias Associated with Undescended Testes**

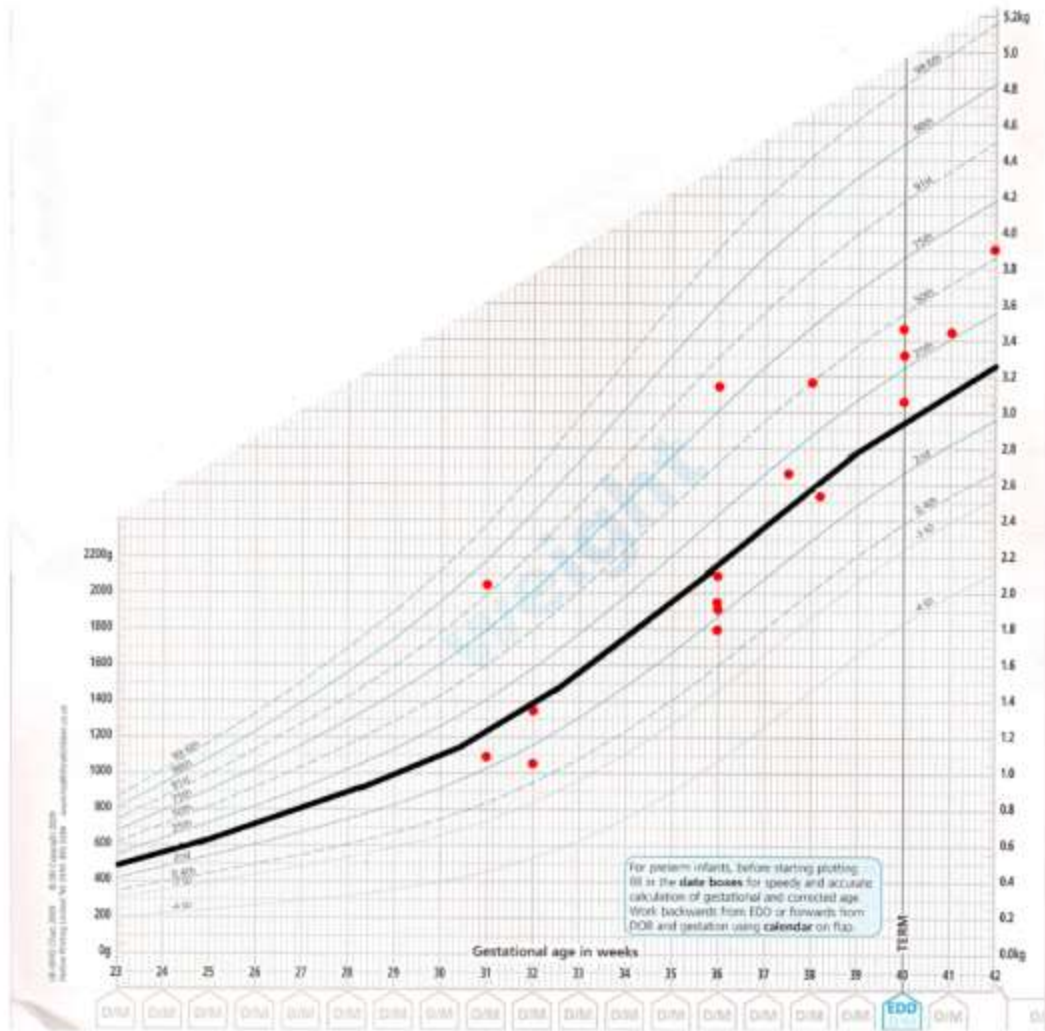
Prabhu Sekaran, Stuart O'Toole, Martyn Flett and Salvatore Cascio\*

The Journal of Urology (2013): 189: 1892 - 1896

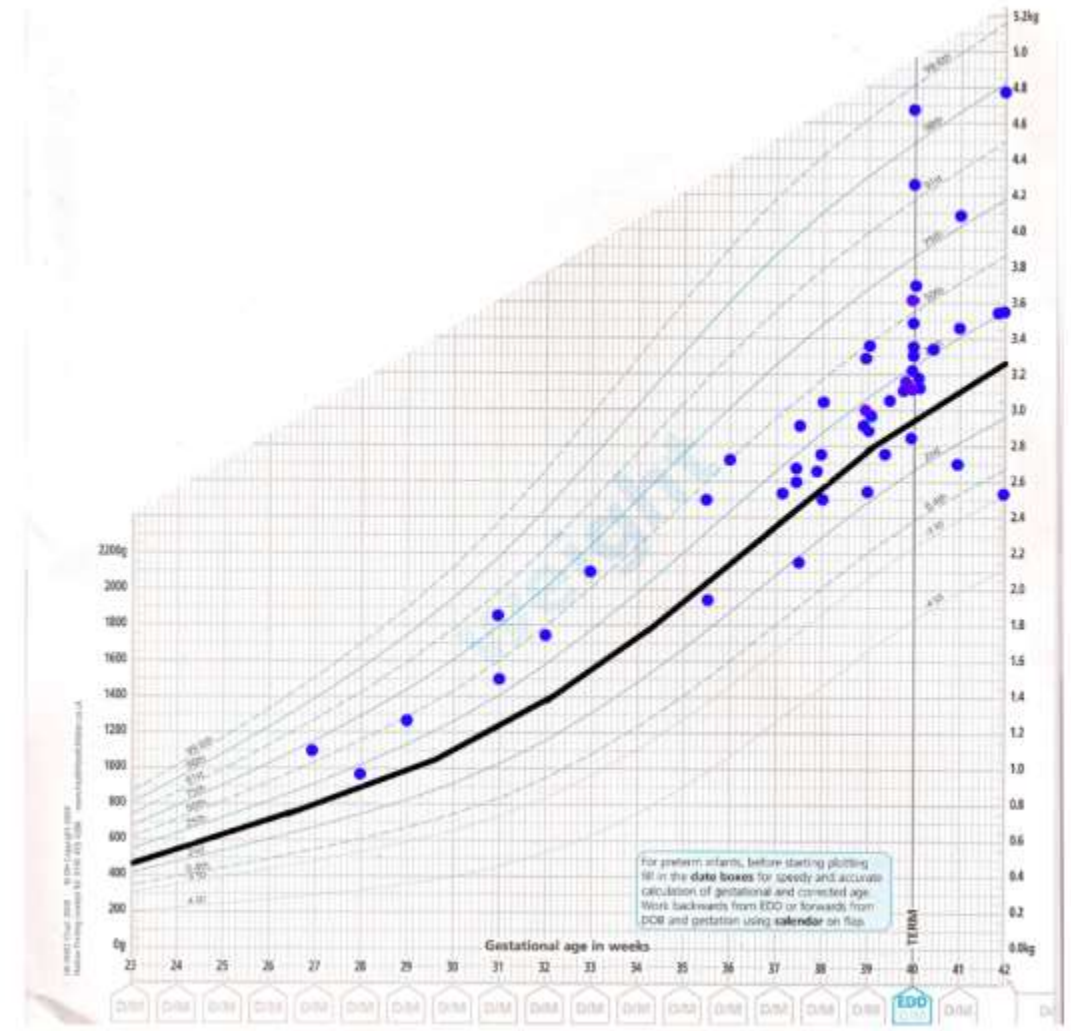
27%

Incidence of intersexuality in patients  
with isolated proximal hypospadias

## Proximal hypospadias and cryptorchidism



## Proximal hypospadias alone



# THE INCIDENCE OF INTERSEXUALITY IN CHILDREN WITH CRYPTORCHIDISM AND HYPOSPADIAS: STRATIFICATION BASED ON GONADAL PALPABILITY AND MEATAL POSITION

MARTIN KAEFER,\* DAVID DIAMOND, W. HARDY HENDREN, SREENU VEMULAPALLI,  
STUART B. BAUER, CRAIG A. PETERS, ANTHONY ATALA AND ALAN B. RETIK

The Journal of Urology (1999): 162: 1003 - 1007



# 47%

Incidence of intersexuality in patients with hypospadias  
and  
one or two impalpable testes

How to manage a patient

# Review the history

Review the history

Examine the patient

Review the history

Examine the patient

Talk to the parents

- Penile webbing
- Chordee
- Distal hypospadias
- Unilateral impalpable testes



- Penile webbing
- Chordee
- Distal hypospadias
- Unilateral impalpable testes





Thankyou

<https://tinyurl.com/v4qfvkf>