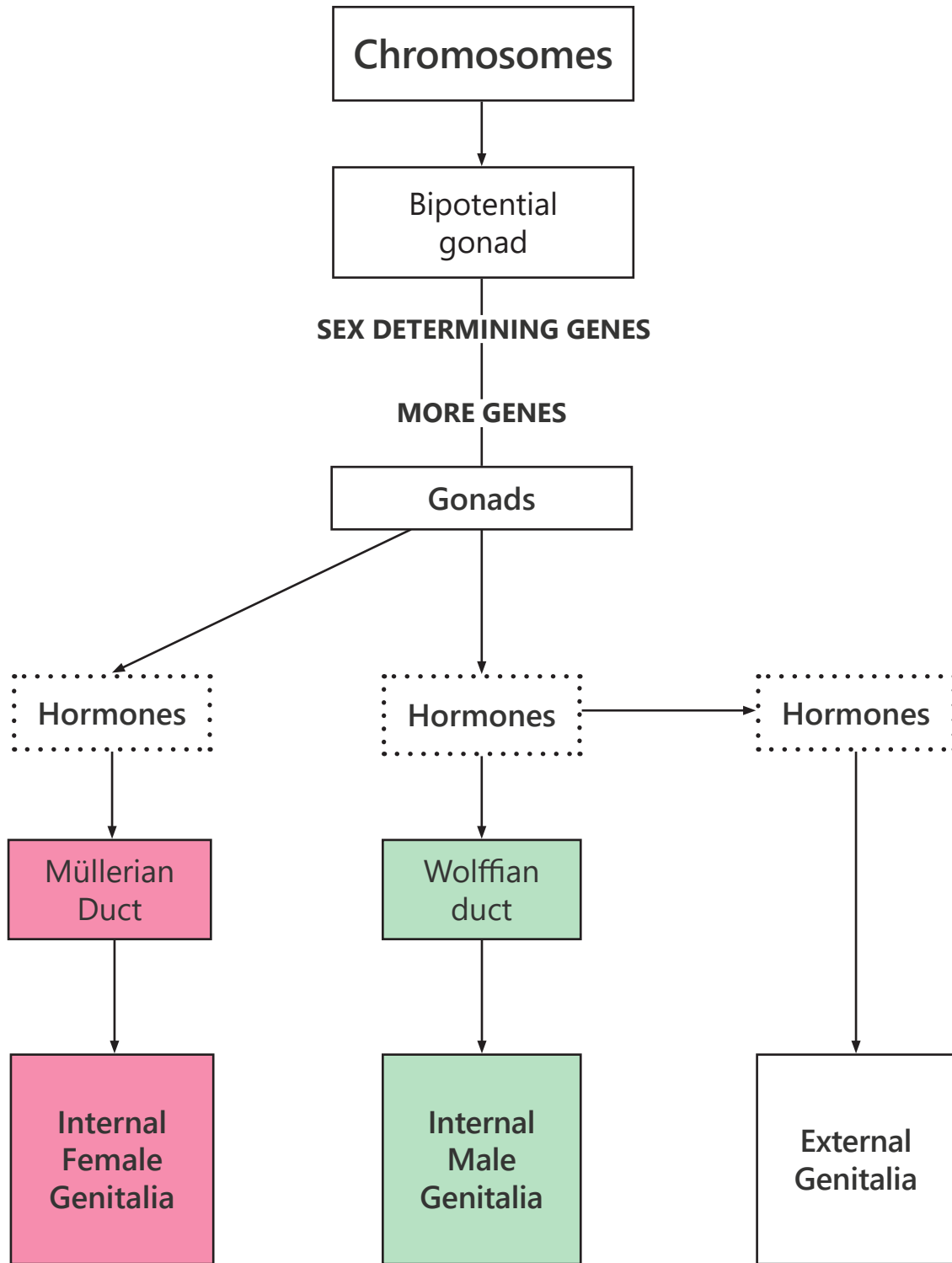


SEX DIFFERENTIATION



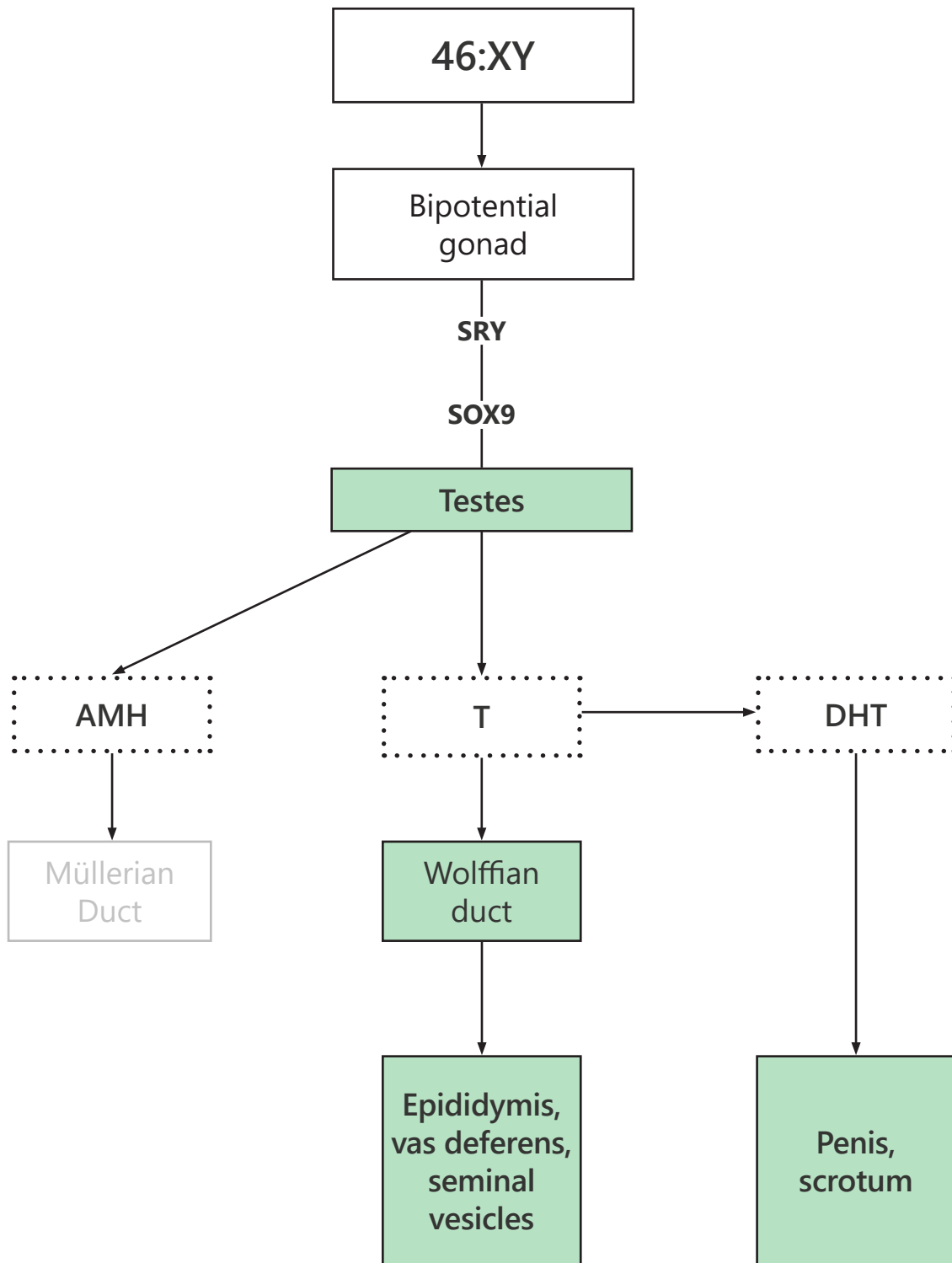
[1] Rey, R., Josso, N., Racine, C. (2020). Sexual differentiation. In: *Endotext*. South Dartmouth, MDText, Inc.

[2] Jones, R., Lopez, K. (2014). *Human Reproductive Biology*, 4th edition. Elsevier.

[3] Wilhelm, D., et al. (2007). Sex determination and gonadal development in mammals. *Physiol Rev*, 87(1-28).

[4] Ogilvy-Stuart, A., et al. (2004). Determination of sex: Early assessment of ambiguous genitalia. *Arch Dis Child*.

TYPICAL MALE



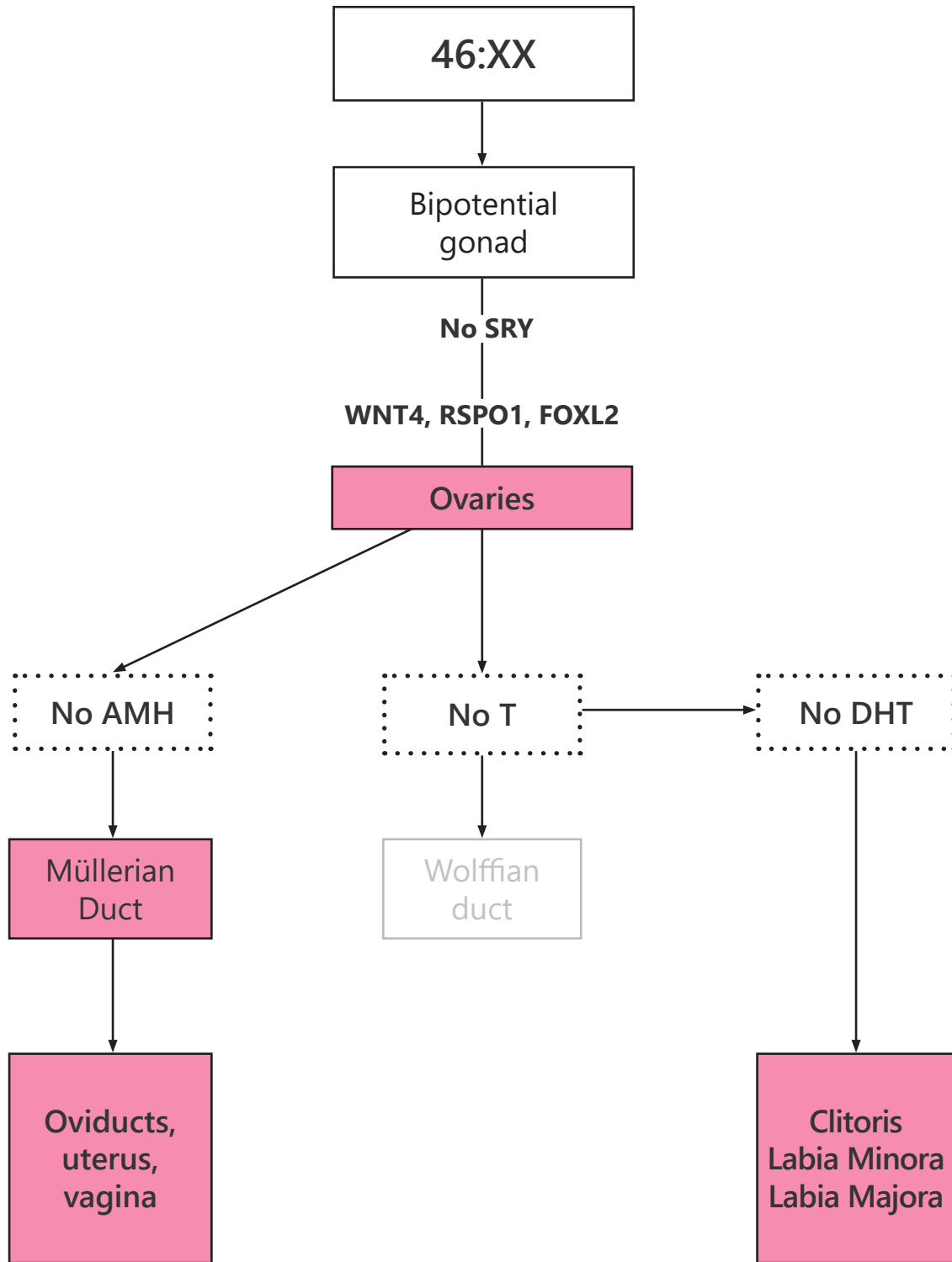
[1] Rey, R., Josso, N., Racine, C. (2020). Sexual differentiation. In: *Endotext*. South Dartmouth, MDText, Inc.

[2] Jones, R., Lopez, K. (2014). *Human Reproductive Biology*, 4th edition. Elsevier.

[3] Wilhelm, D., et al. (2007). Sex determination and gonadal development in mammals. *Physiol Rev*, 87(1-28).

[4] Ogilvy-Stuart, A., et al. (2004). Determination of sex: Early assessment of ambiguous genitalia. *Arch Dis Child*.

TYPICAL FEMALE



[1] Rey, R., Josso, N., Racine, C. (2020). Sexual differentiation. In: *Endotext*. South Dartmouth, MDText, Inc.

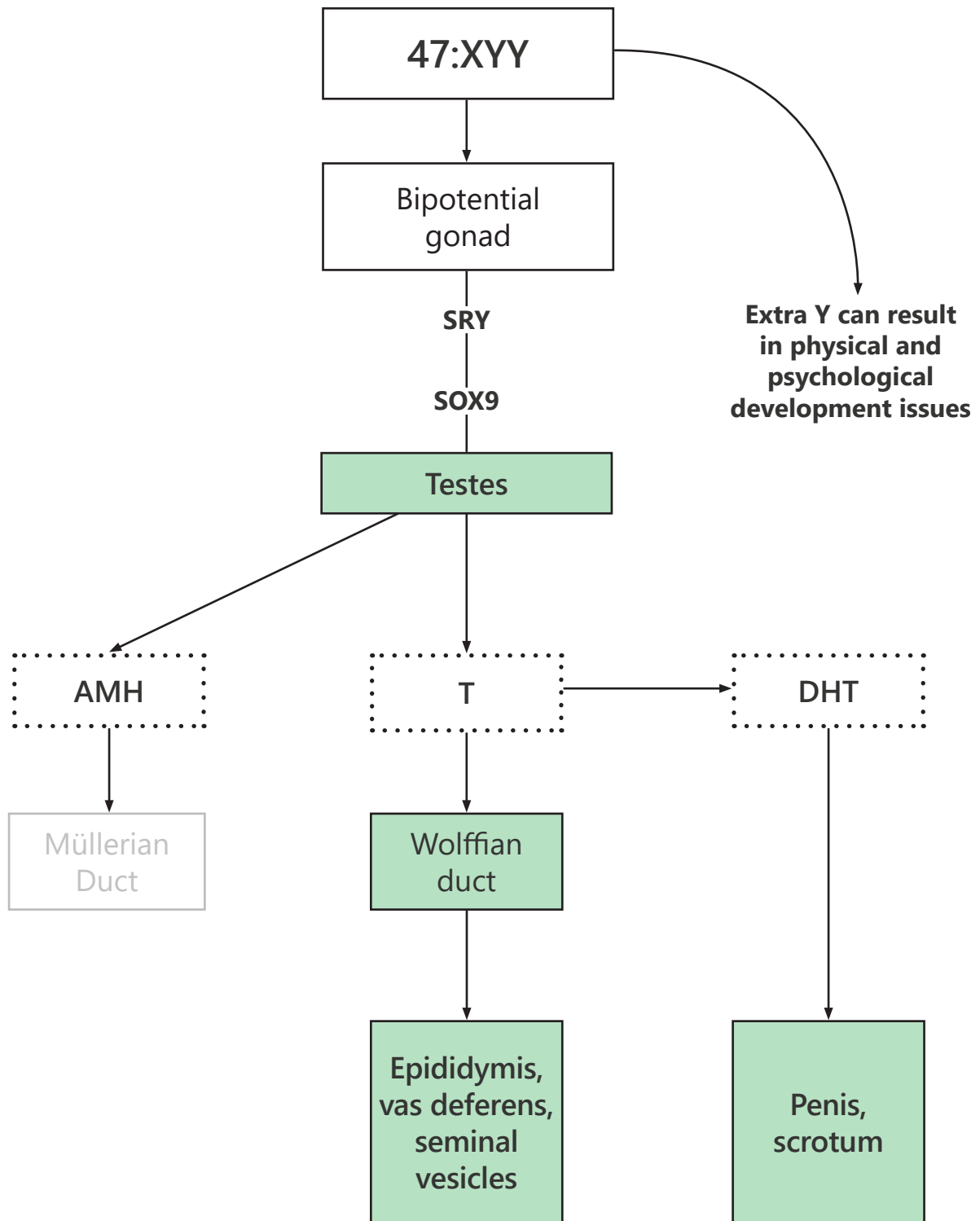
[2] Jones, R., Lopez, K. (2014). *Human Reproductive Biology*, 4th edition. Elsevier.

[3] Wilhelm, D., et al. (2007). Sex determination and gonadal development in mammals. *Physiol Rev*, 87(1-28).

[4] Ogilvy-Stuart, A., et al. (2004). Determination of sex: Early assessment of ambiguous genitalia. *Arch Dis Child*.

47:XYY (JACOB'S SYNDROME)

Male | 1 in 1,000 births | Usually fertile



[1] Rey, R., Josso, N., Racine, C. (2020). Sexual differentiation. In: *Endotext*. South Dartmouth, MDText, Inc.

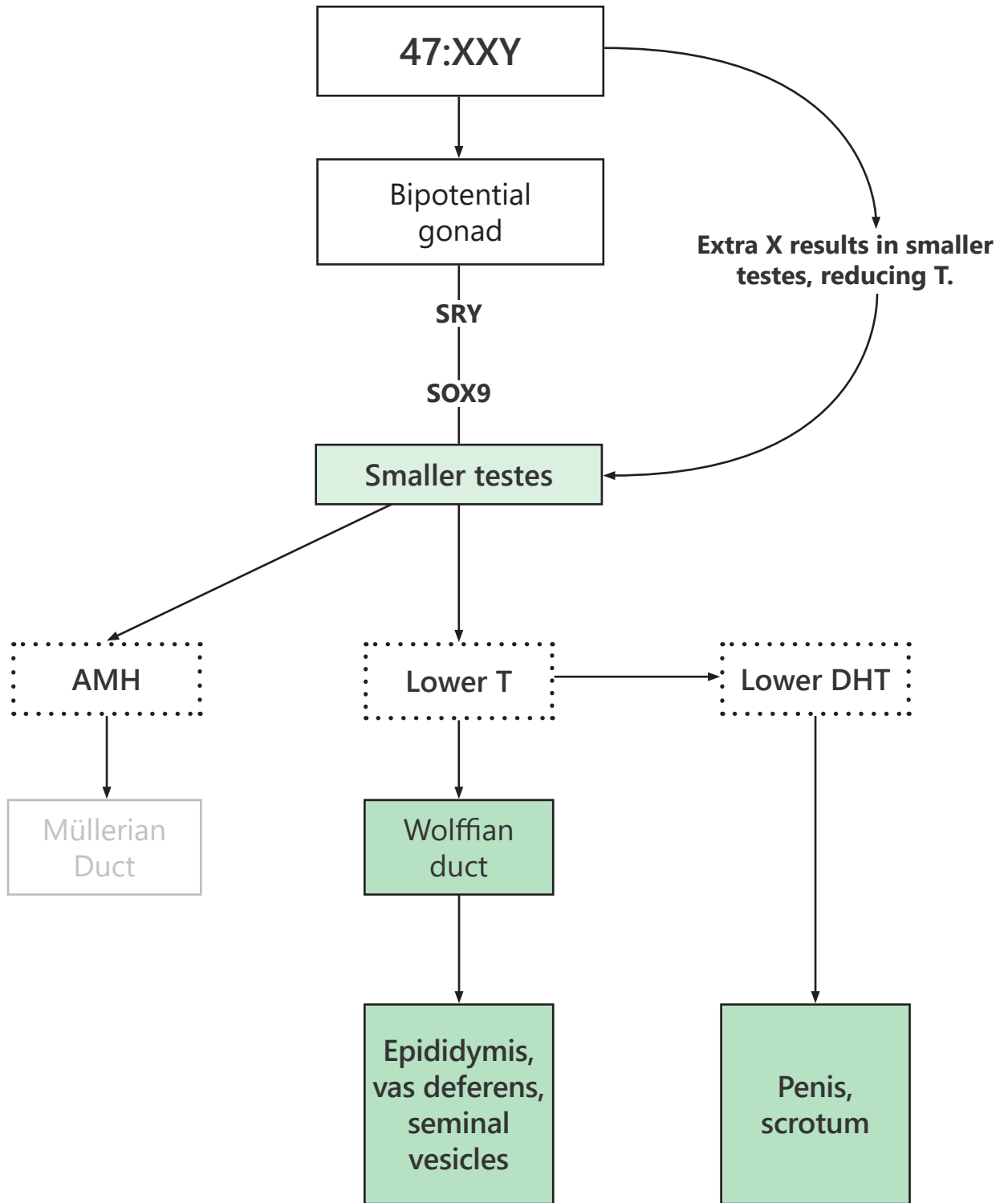
[2] NIH. (2020). 47:XYY syndrome. *Genetics Home Reference*, National Library of Medicine.

[3] Witchel, S. (2018). Disorders of sex development. *Best Practice and Research in Clinical Obstetrics and Gynecology*.

[4] Ogilvy-Stuart, A., et al. (2004). Determination of sex: Early assessment of ambiguous genitalia. *Arch Dis Child*.

47:XXY (KLINEFELTER SYNDROME)

Male | 1 in 650 births | Usually infertile



[1] Rey, R., Josso, N., Racine, C. (2020). Sexual differentiation. In: *Endotext*. South Dartmouth, MDText, Inc.

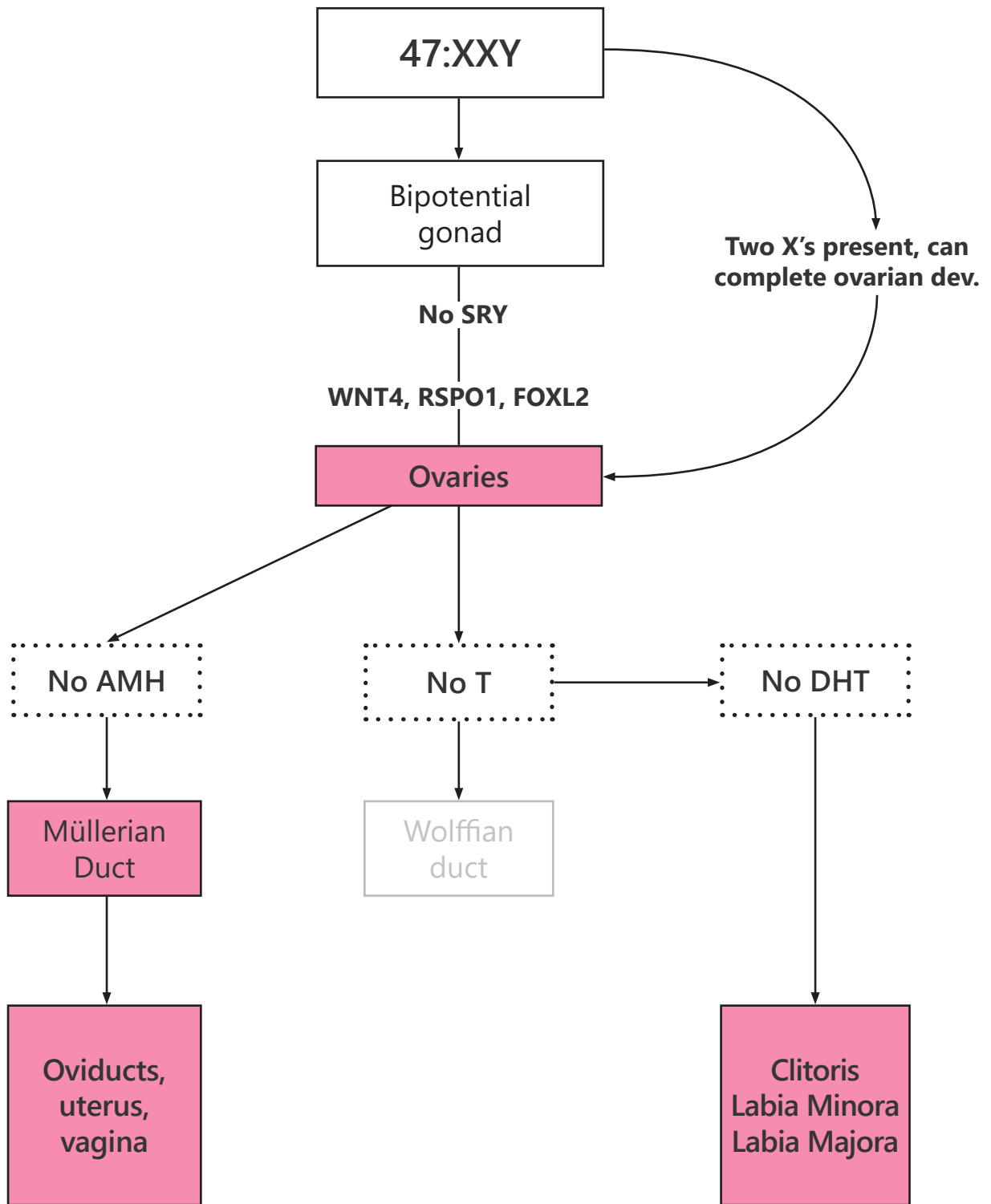
[2] NIH. (2020). Klinefelter syndrome. *Genetics Home Reference*, National Library of Medicine.

[3] Witchel, S. (2018). Disorders of sex development. *Best Practice and Research in Clinical Obstetrics and Gynecology*.

[4] Ogilvy-Stuart, A., et al. (2004). Determination of sex: Early assessment of ambiguous genitalia. *Arch Dis Child*.

47:XXY (SRY NEGATIVE FEMALE)

Female | Vanishingly rare | Unknown fertility rate



[1] Rey, R., Josso, N., Racine, C. (2020). Sexual differentiation. In: *Endotext*. South Dartmouth, MDText, Inc.

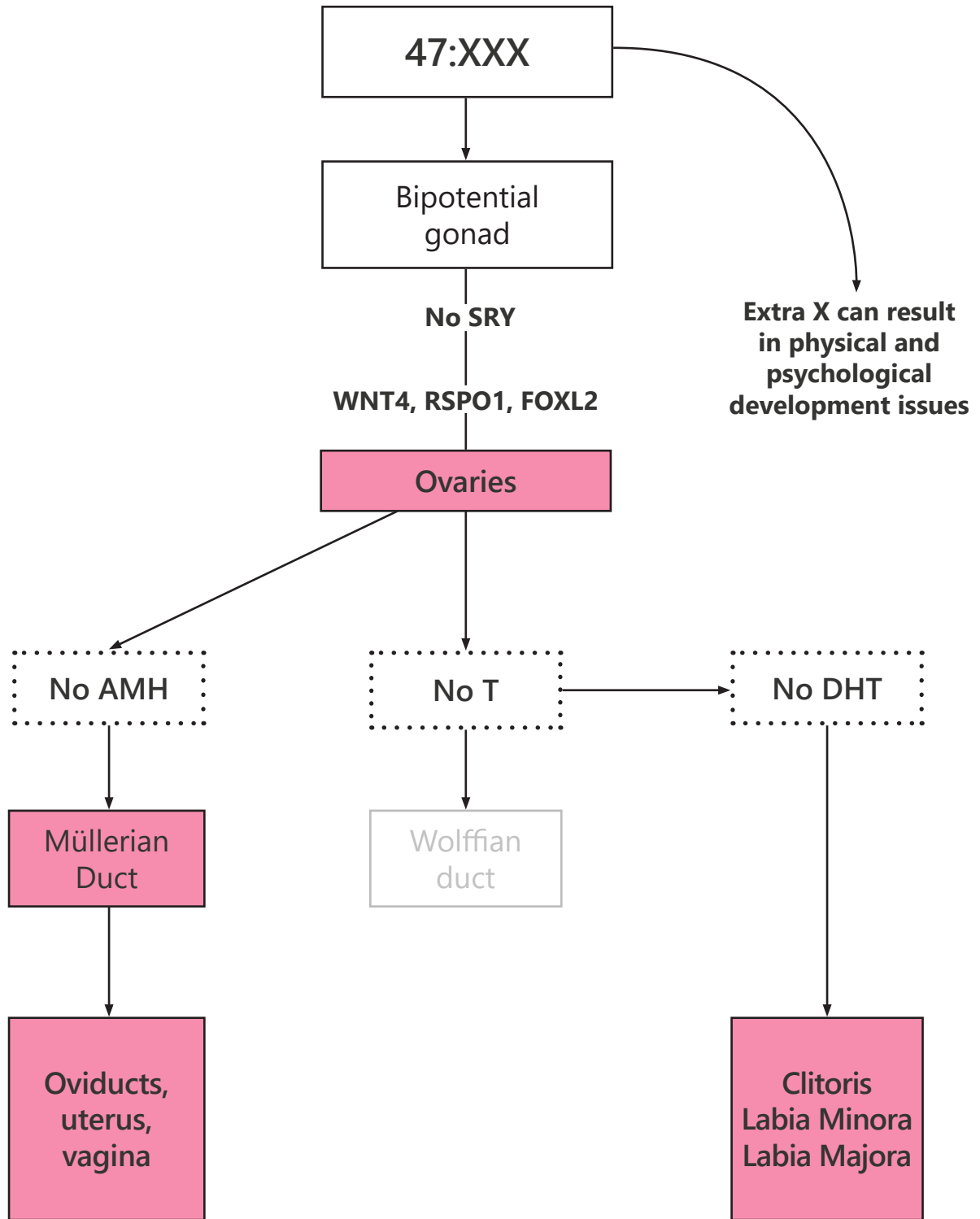
[2] Hu, L., et al. (2019). A 47:XXY pregnant woman without the SRY gene. *Sex Development*, 13(83-86).

[3] Witchel, S. (2018). Disorders of sex development. *Best Practice and Research in Clinical Obstetrics and Gynecology*.

[4] Ogilvy-Stuart, A., et al. (2004). Determination of sex: Early assessment of ambiguous genitalia. *Arch Dis Child*.

47:XXX (TRISOMY X)

Female | 1 in 1,000 births | Almost always fertile



[1] Rey, R., Josso, N., Racine, C. (2020). Sexual differentiation. In: *Endotext*. South Dartmouth, MDText, Inc.

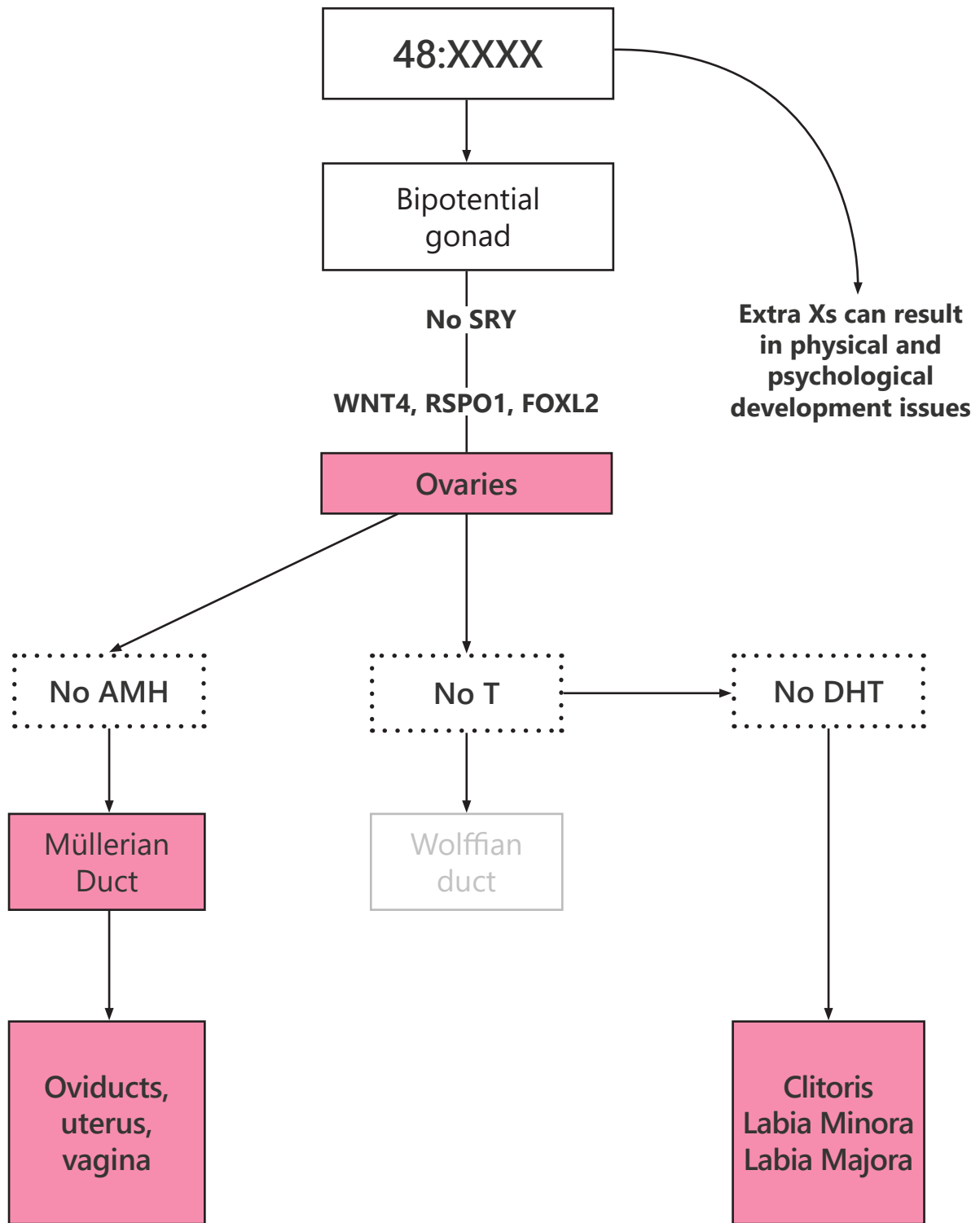
[2] NIH. (2020). Triple X syndrome. *Genetics Home Reference*, National Library of Medicine.

[3] Witchel, S. (2018). Disorders of sex development. *Best Practice and Research in Clinical Obstetrics and Gynecology*.

[4] Ogilvy-Stuart, A., et al. (2004). Determination of sex: Early assessment of ambiguous genitalia. *Arch Dis Child*.

48:XXXX (TETRASOMY X)

Female | 1 in 50,000 births | Usually fertile



[1] Rey, R., Josso, N., Racine, C. (2020). Sexual differentiation. In: *Endotext*. South Dartmouth, MDText, Inc.

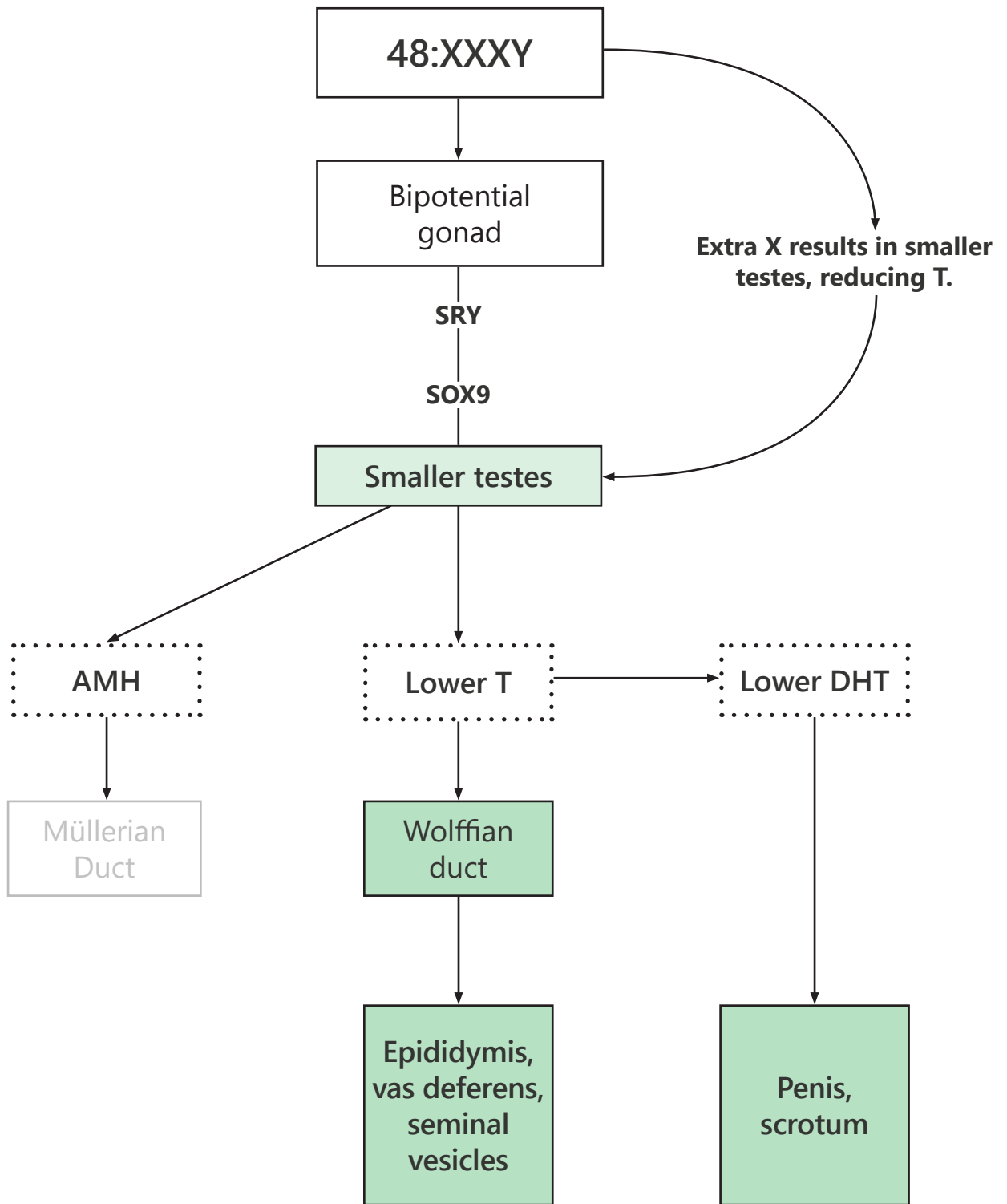
[2] NIH. (2020). Tetrasomy X. *Genetic and Rare Diseases Information Center*. National Library of Medicine.

[3] Witchel, S. (2018). Disorders of sex development. *Best Practice and Research in Clinical Obstetrics and Gynecology*.

[4] Ogilvy-Stuart, A., et al. (2004). Determination of sex: Early assessment of ambiguous genitalia. *Arch Dis Child*.

48:XXXY SYNDROME

Male | 1 in 17,000 to 1 in 50,000 births | Usually infertile



[1] Rey, R., Josso, N., Racine, C. (2020). Sexual differentiation. In: *Endotext*. South Dartmouth, MDText, Inc.

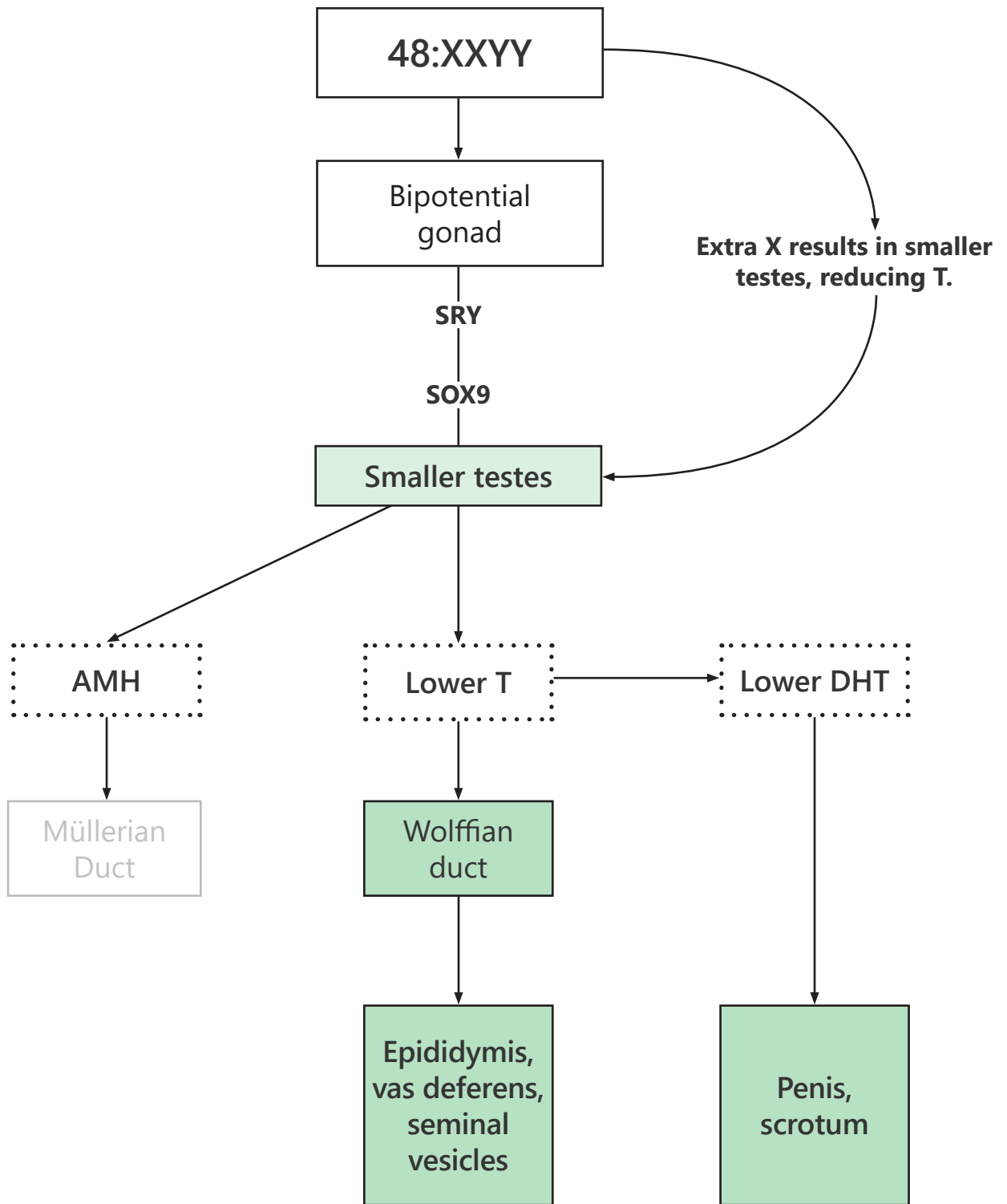
[2] NIH. (2020). 48:XXXY syndrome. *Genetics Home Reference*, National Library of Medicine.

[3] Witchel, S. (2018). Disorders of sex development. *Best Practice and Research in Clinical Obstetrics and Gynecology*.

[4] Ogilvy-Stuart, A., et al. (2004). Determination of sex: Early assessment of ambiguous genitalia. *Arch Dis Child*.

48:XXYY SYNDROME

Male | 1 in 18,000 to 1 in 40,000 births | Usually infertile



[1] Rey, R., Josso, N., Racine, C. (2020). Sexual differentiation. In: *Endotext*. South Dartmouth, MDText, Inc.

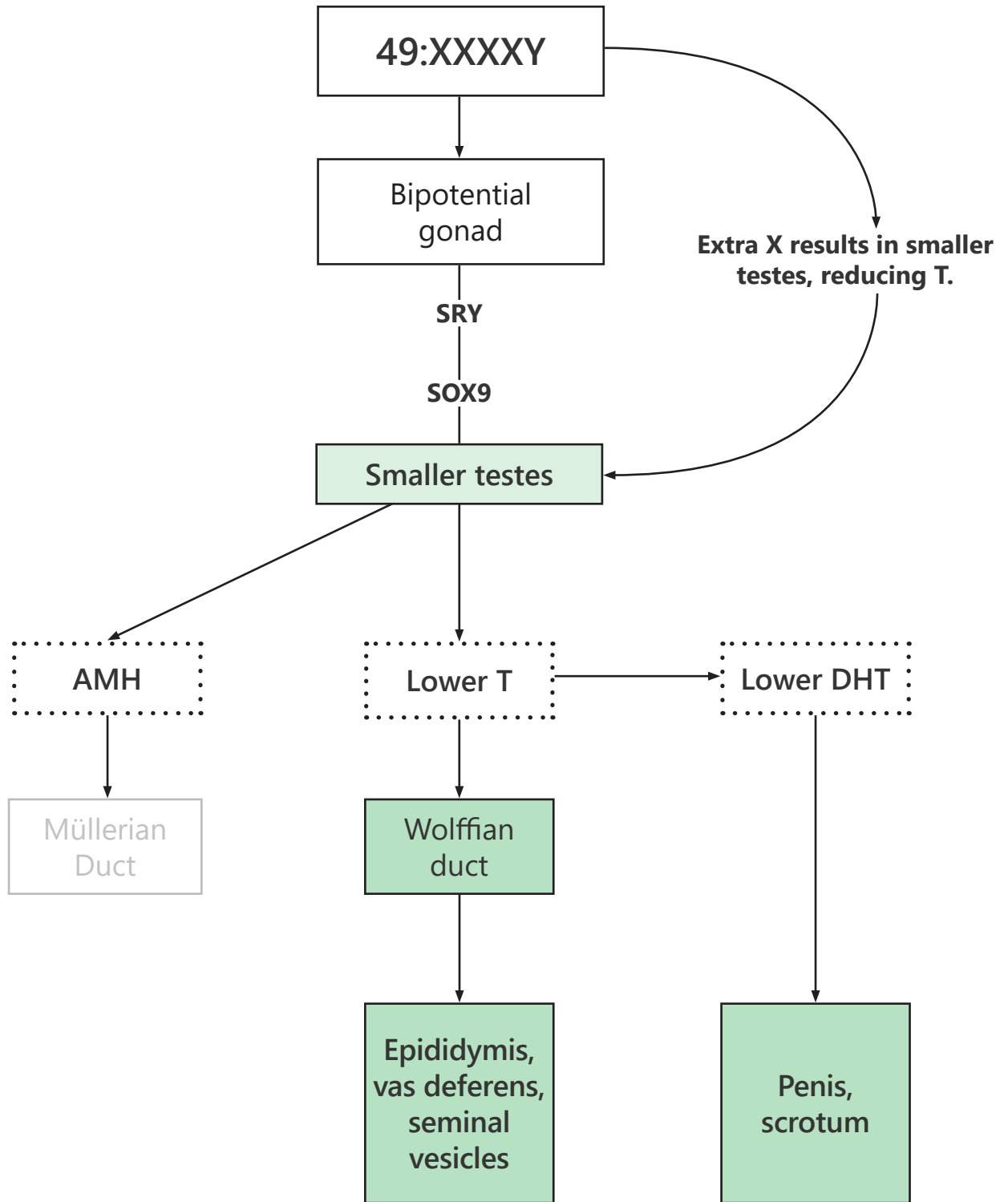
[2] NIH. (2020). 48:XXYY syndrome. *Genetics Home Reference*, National Library of Medicine.

[3] Witchel, S. (2018). Disorders of sex development. *Best Practice and Research in Clinical Obstetrics and Gynecology*.

[4] Ogilvy-Stuart, A., et al. (2004). Determination of sex: Early assessment of ambiguous genitalia. *Arch Dis Child*.

49:XXXXY SYNDROME

Male | 1 in 85,000 to 1 in 100,000 births | Usually infertile



[1] Rey, R., Josso, N., Racine, C. (2020). Sexual differentiation. In: *Endotext*. South Dartmouth, MDText, Inc.

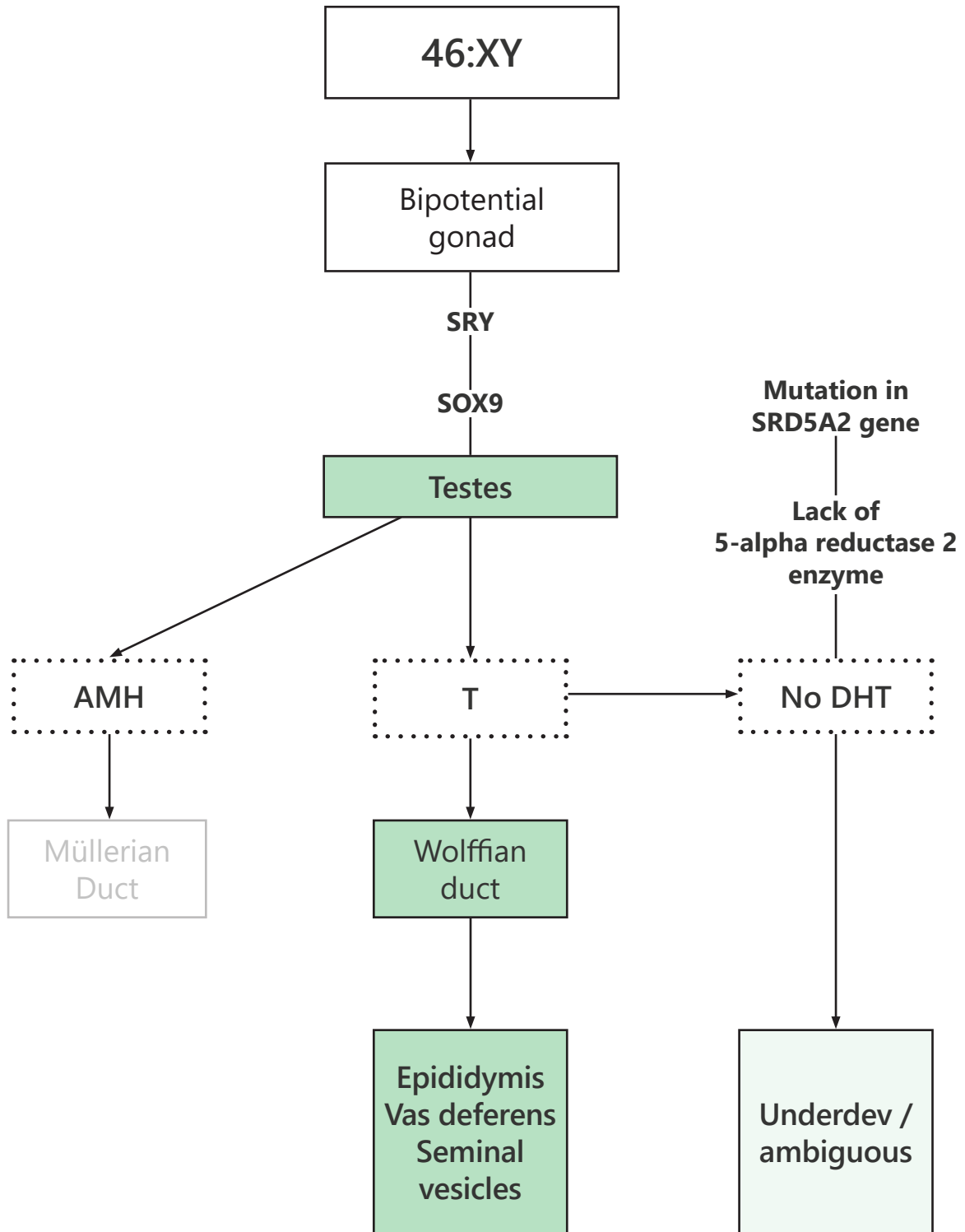
[2] NIH. (2020). 49:XXXXY syndrome. *Genetics Home Reference, National Library of Medicine*.

[3] Witchel, S. (2018). Disorders of sex development. *Best Practice and Research in Clinical Obstetrics and Gynecology*.

[4] Ogilvy-Stuart, A., et al. (2004). Determination of sex: Early assessment of ambiguous genitalia. *Arch Dis Child*.

5-ALPHA REDUCTASE DEFICIENCY

Male | Unknown birth rate | Often fertile



[1] Rey, R., Josso, N., Racine, C. (2020). Sexual differentiation. In: *Endotext*. South Dartmouth, MDText, Inc.

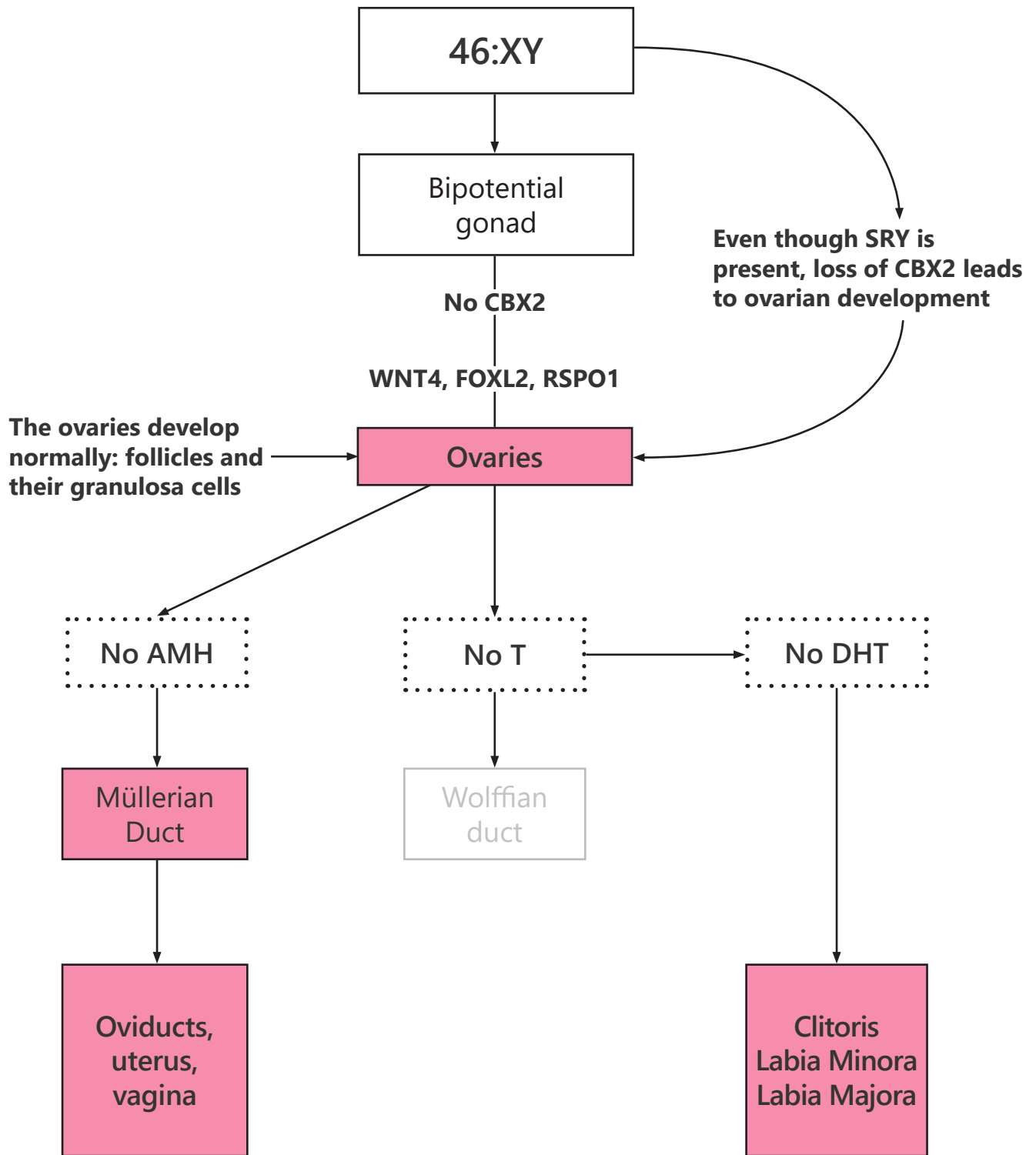
[2] NIH. (2020). 5-alpha reductase deficiency. *Genetics Home Reference*, National Library of Medicine.

[3] Witchel, S. (2018). Disorders of sex development. *Best Practice and Research in Clinical Obstetrics and Gynecology*.

[4] Ogilvy-Stuart, A., et al. (2004). Determination of sex: Early assessment of ambiguous genitalia. *Arch Dis Child*.

CBX2 NEGATIVE FEMALE

Female | Vanishingly rare | Unknown fertility rate

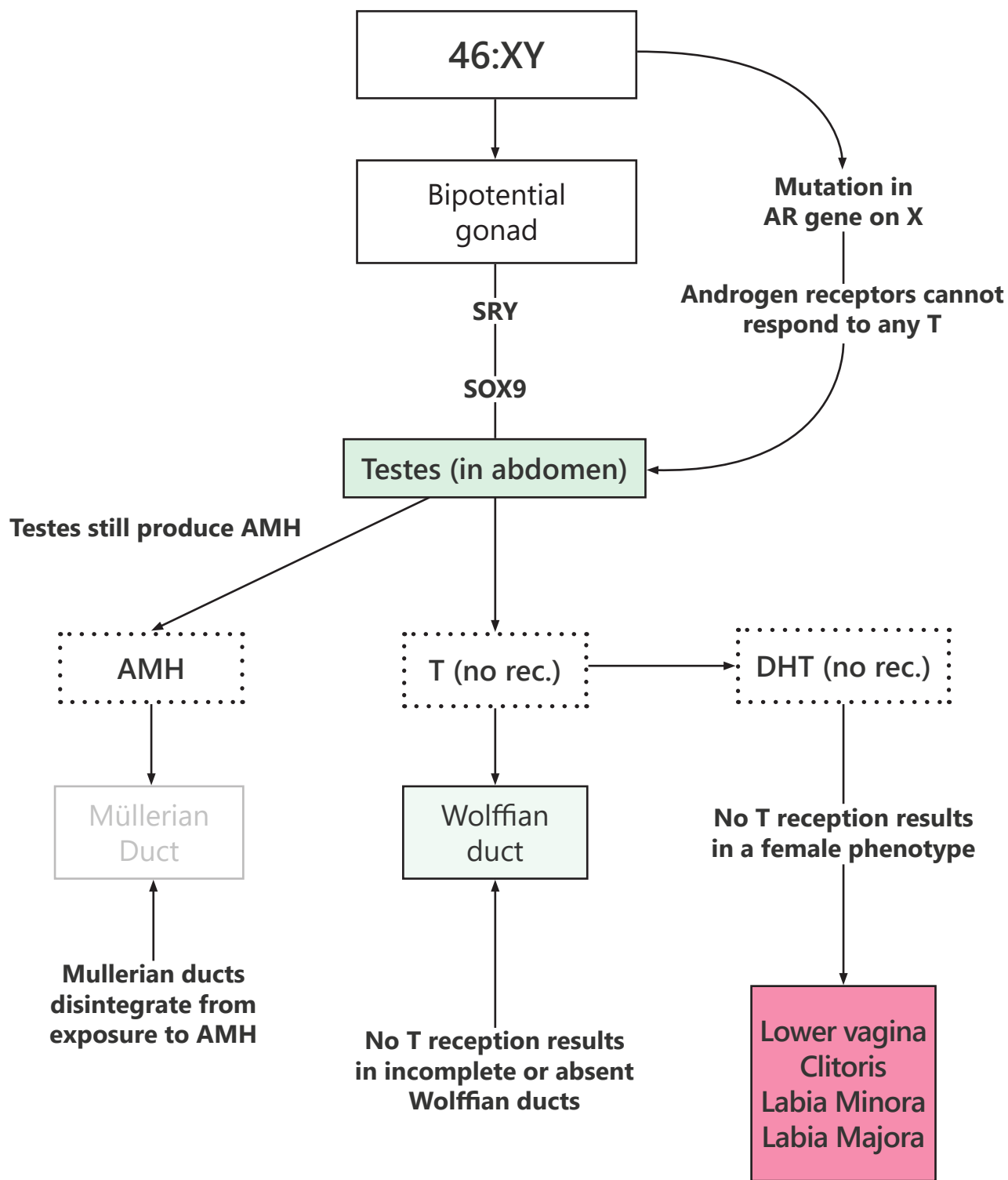


[1] Biason-Lauber, A., et al. (2009). Ovaries and female phenotype in a girl with 46,XY karyotype and mutations in the CBX2 gene. *Am J Human Gen*, 84(658-663).

[2] Moreno-Garcia, S., et al. (2019). CBX2 is required to stabilize the testis pathway by repressing WNT signaling. *PLoS Genetics*, 15(5).

COMPLETE ANDROGEN INSENSITIVITY SYNDROME

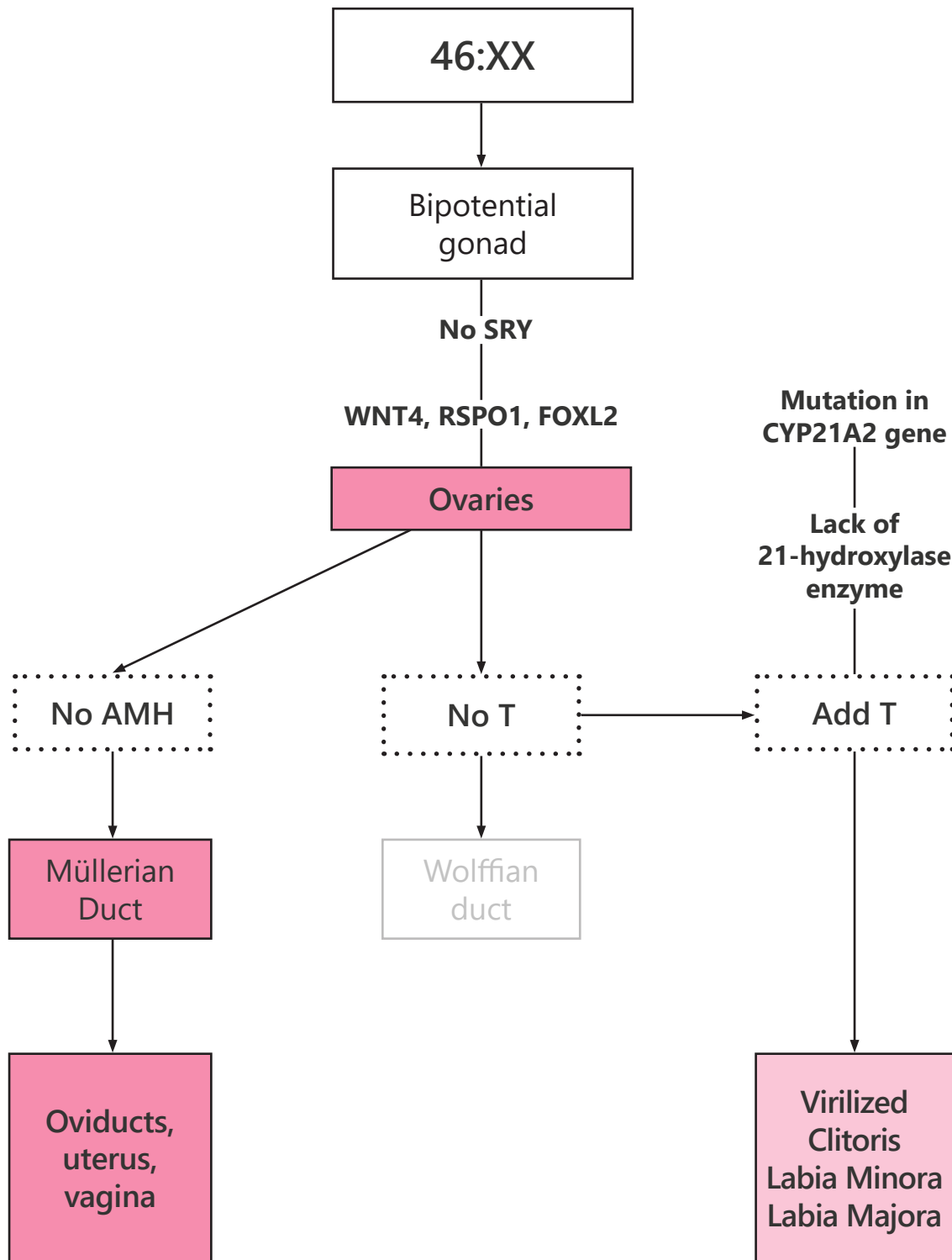
"Male sex rejection" female | 1 in 20,000 | Always infertile



[1] Rey, R., Josso, N., Racine, C. (2020). Sexual differentiation. In: *Endotext*. South Dartmouth, MDText, Inc.
[2] NIH. (2020). Complete androgen insensitivity syndrome. *Genetics Home Reference*, National Library of Medicine.
[3] Oakes, M., et al. (2008). Complete androgen insensitivity syndrome: a review. *J Ped Adolesc Gynec*, 21(6).
[4] Ogilvy-Stuart, A., et al. (2004). Determination of sex: Early assessment of ambiguous genitalia. *Arch Dis Child*.

CONGENITAL ADRENAL HYPERPLASIA

Female | 1 in 15,000 births | Usually fertile



[1] Rey, R., Josso, N., Racine, C. (2020). Sexual differentiation. In: *Endotext*. South Dartmouth, MDText, Inc.

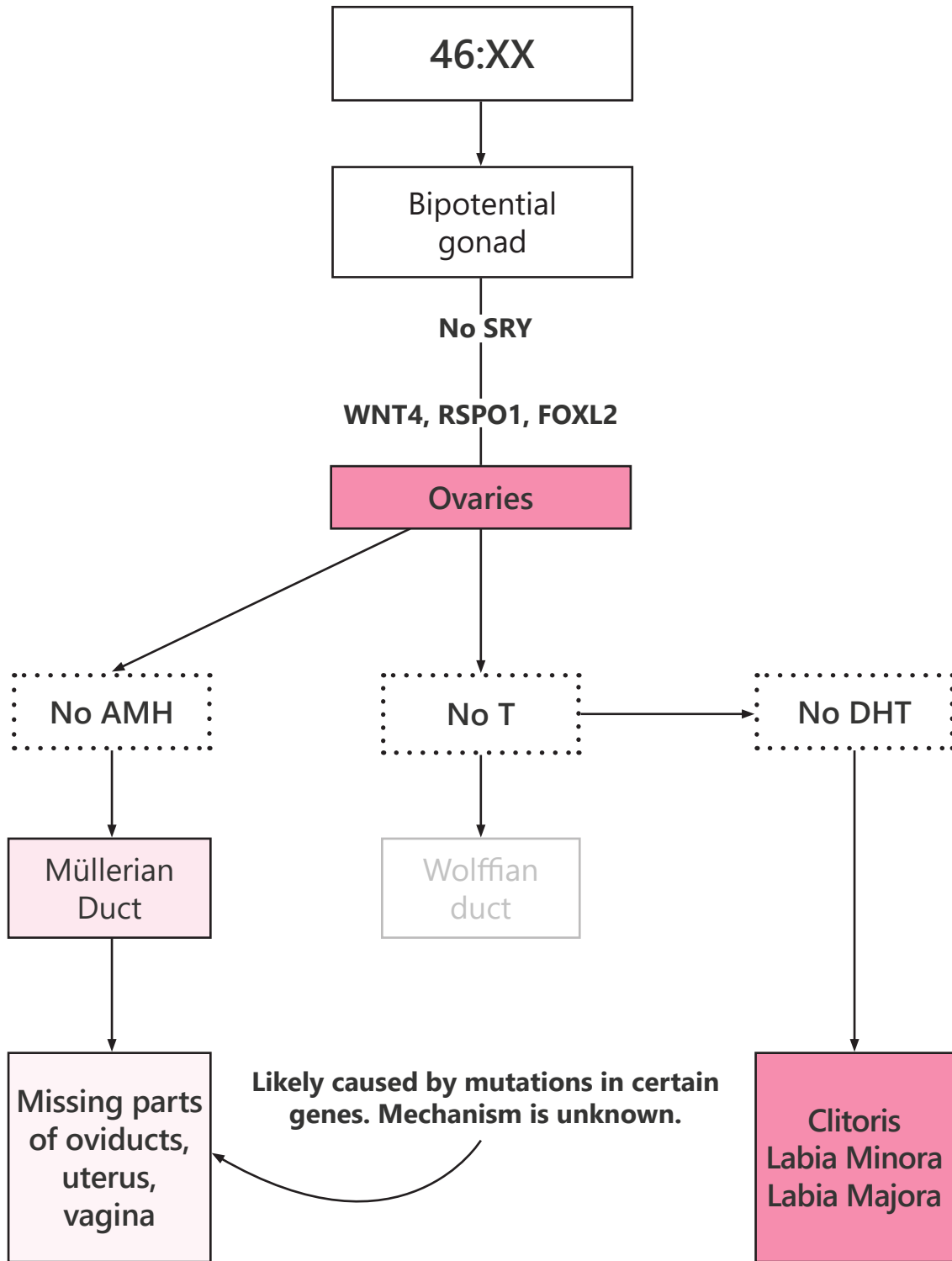
[2] NIH. (2020). 21-hydroxylase deficiency. *Genetics Home Reference*, National Library of Medicine.

[3] Witchel, S. (2018). Disorders of sex development. *Best Practice and Research in Clinical Obstetrics and Gynecology*.

[4] Ogilvy-Stuart, A., et al. (2004). Determination of sex: Early assessment of ambiguous genitalia. *Arch Dis Child*.

MRKH

Female | 1 in 2,500 births | Fertile ovaries, but unable to gestate offspring



[1] Rey, R., Josso, N., Racine, C. (2020). Sexual differentiation. In: *Endotext*. South Dartmouth, MDText, Inc.

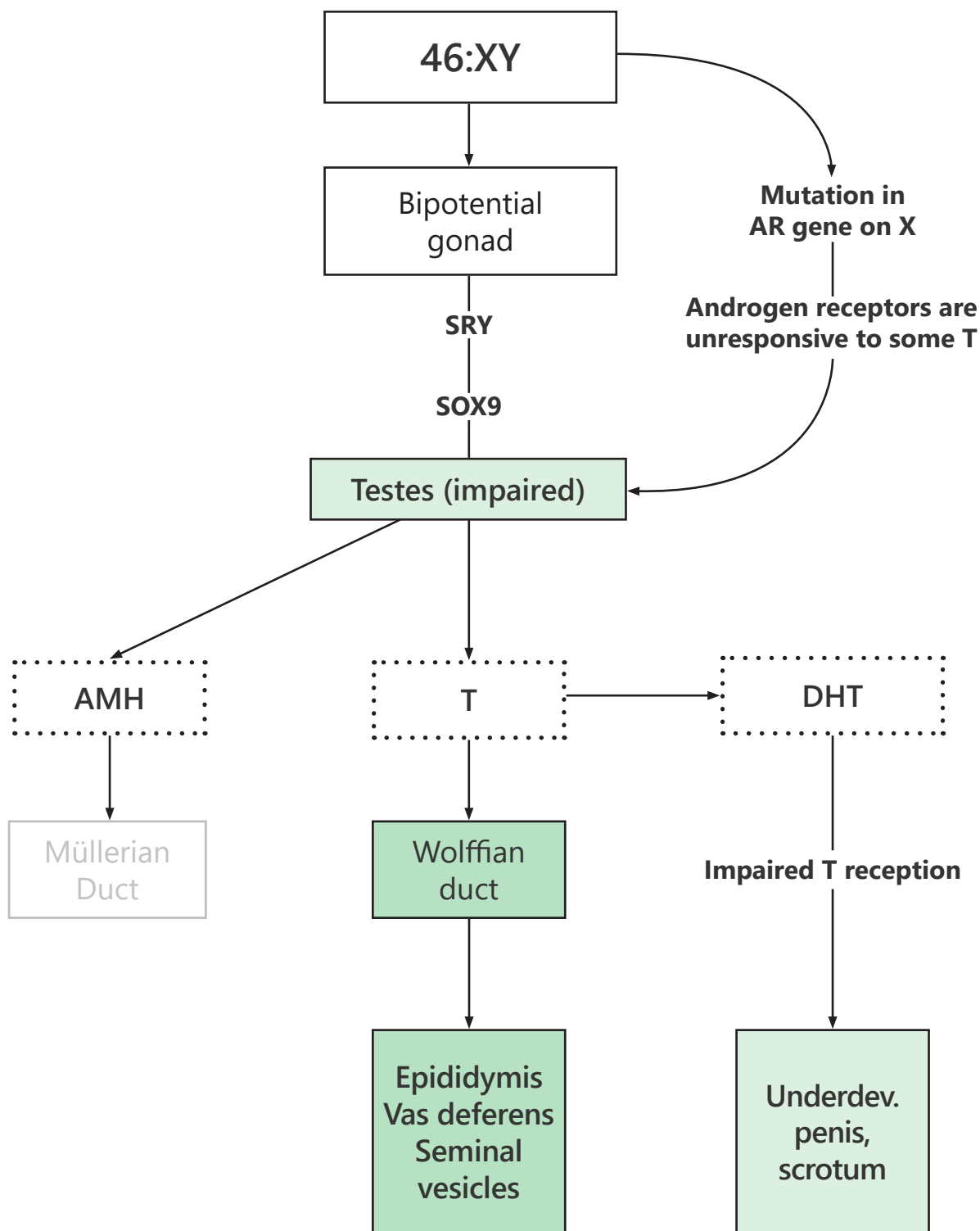
[2] NIH. (2020). Mayer-Rokitansky-Küster-Hausler syndrome. *Genetics Home Reference*, National Library of Medicine.

[3] Witchel, S. (2018). Disorders of sex development. *Best Practice and Research in Clinical Obstetrics and Gynecology*.

[4] Ogilvy-Stuart, A., et al. (2004). Determination of sex: Early assessment of ambiguous genitalia. *Arch Dis Child*.

MILD ANDROGEN INSENSITIVITY SYNDROME

Male | Unknown rate | Often infertile



[1] Rey, R., Josso, N., Racine, C. (2020). Sexual differentiation. In: *Endotext*. South Dartmouth, MDText, Inc.

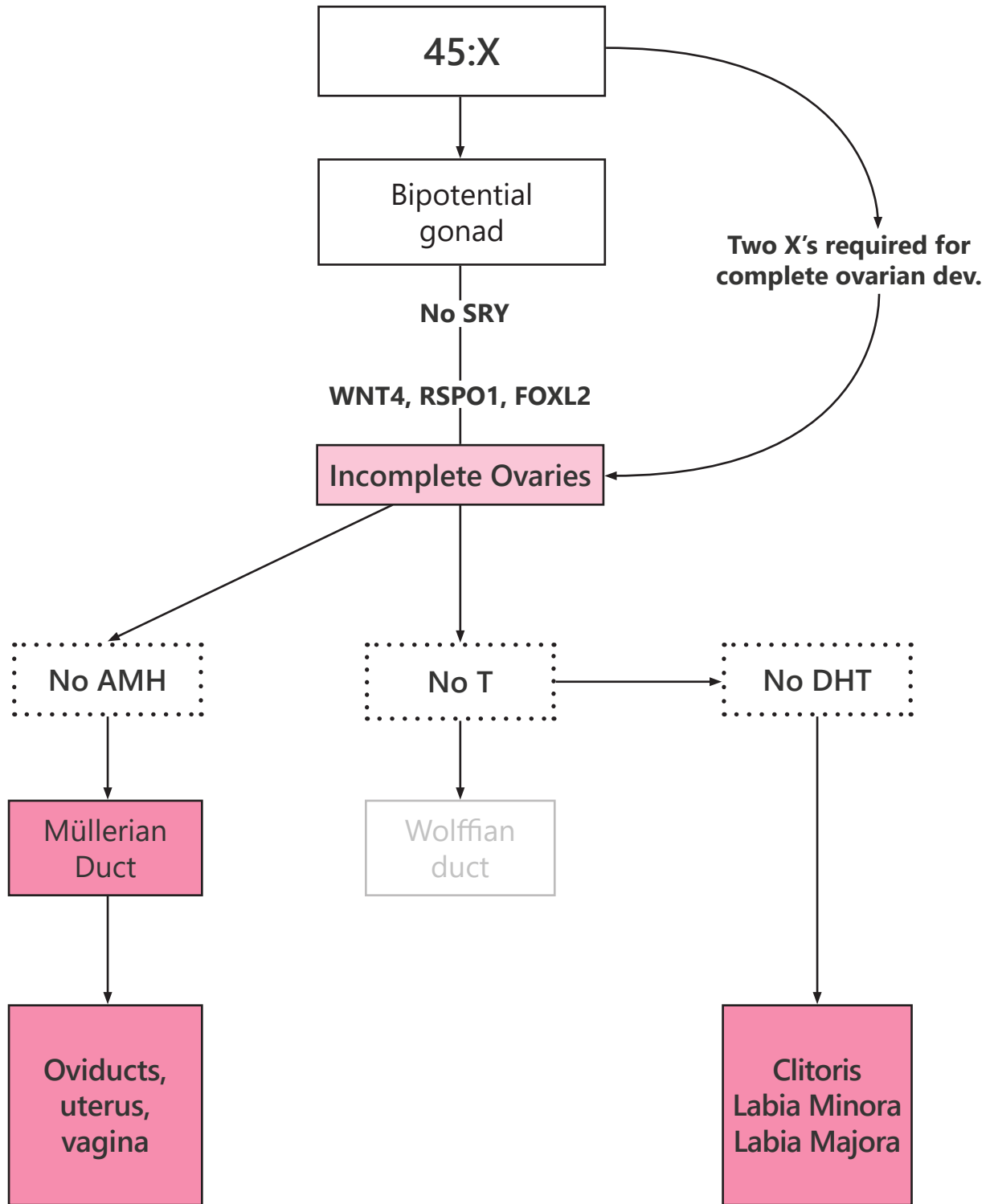
[2] NIH. (2020). Androgen insensitivity syndrome. *Genetics Home Reference*, National Library of Medicine.

[3] Gottlieb, B., et al. (2017). Androgen Insensitivity Syndrome. *Gene Reviews (NIH)*.

[4] Ogilvy-Stuart, A., et al. (2004). Determination of sex: Early assessment of ambiguous genitalia. *Arch Dis Child*.

MONOSOMY X (TURNER SYNDROME)

Female | 1 in 2,500 births | Usually infertile



[1] Rey, R., Josso, N., Racine, C. (2020). Sexual differentiation. In: *Endotext*. South Dartmouth, MDText, Inc.

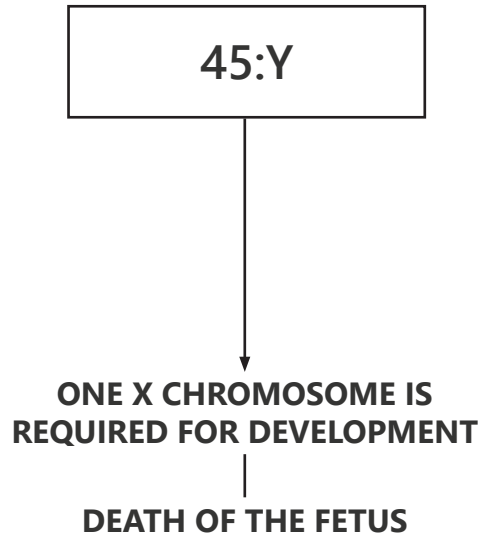
[2] NIH. (2020). Turner Syndrome. *Genetics Home Reference*, National Library of Medicine.

[3] Witchel, S. (2018). Disorders of sex development. *Best Practice and Research in Clinical Obstetrics and Gynecology*.

[4] Ogilvy-Stuart, A., et al. (2004). Determination of sex: Early assessment of ambiguous genitalia. *Arch Dis Child*.

MONOSOMY Y

Not viable



[1] Rey, R., Josso, N., Racine, C. (2020). Sexual differentiation. In: *Endotext*. South Dartmouth, MDText, Inc.

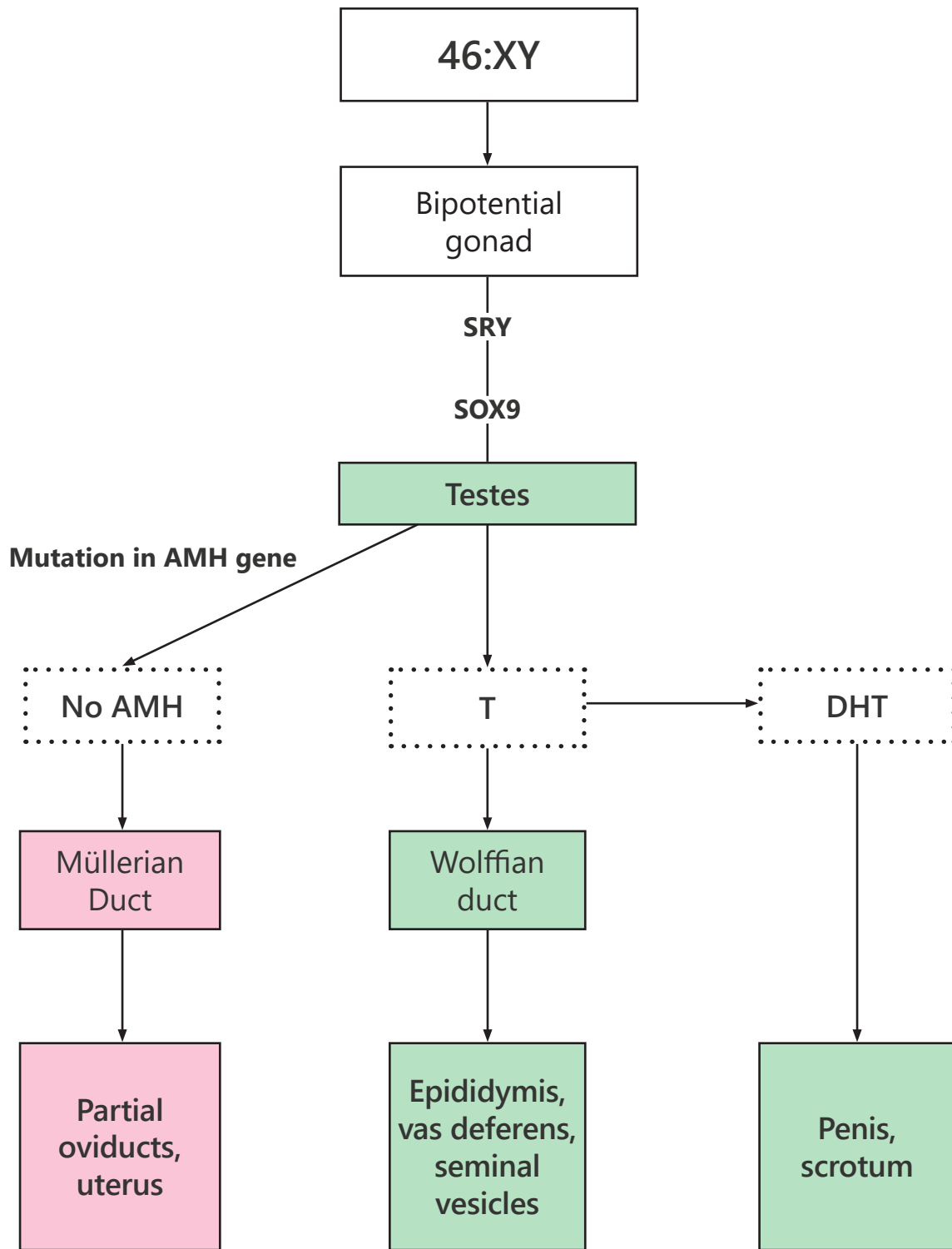
[2] Jones, R., Lopez, K. (2014). *Human Reproductive Biology*, 4th edition. Elsevier.

[3] Wilhelm, D., et al. (2007). Sex determination and gonadal development in mammals. *Physiol Rev*, 87(1-28).

[4] Ogilvy-Stuart, A., et al. (2004). Determination of sex: Early assessment of ambiguous genitalia. *Arch Dis Child*.

PERSISTENT MÜLLERIAN DUCT SYNDROME

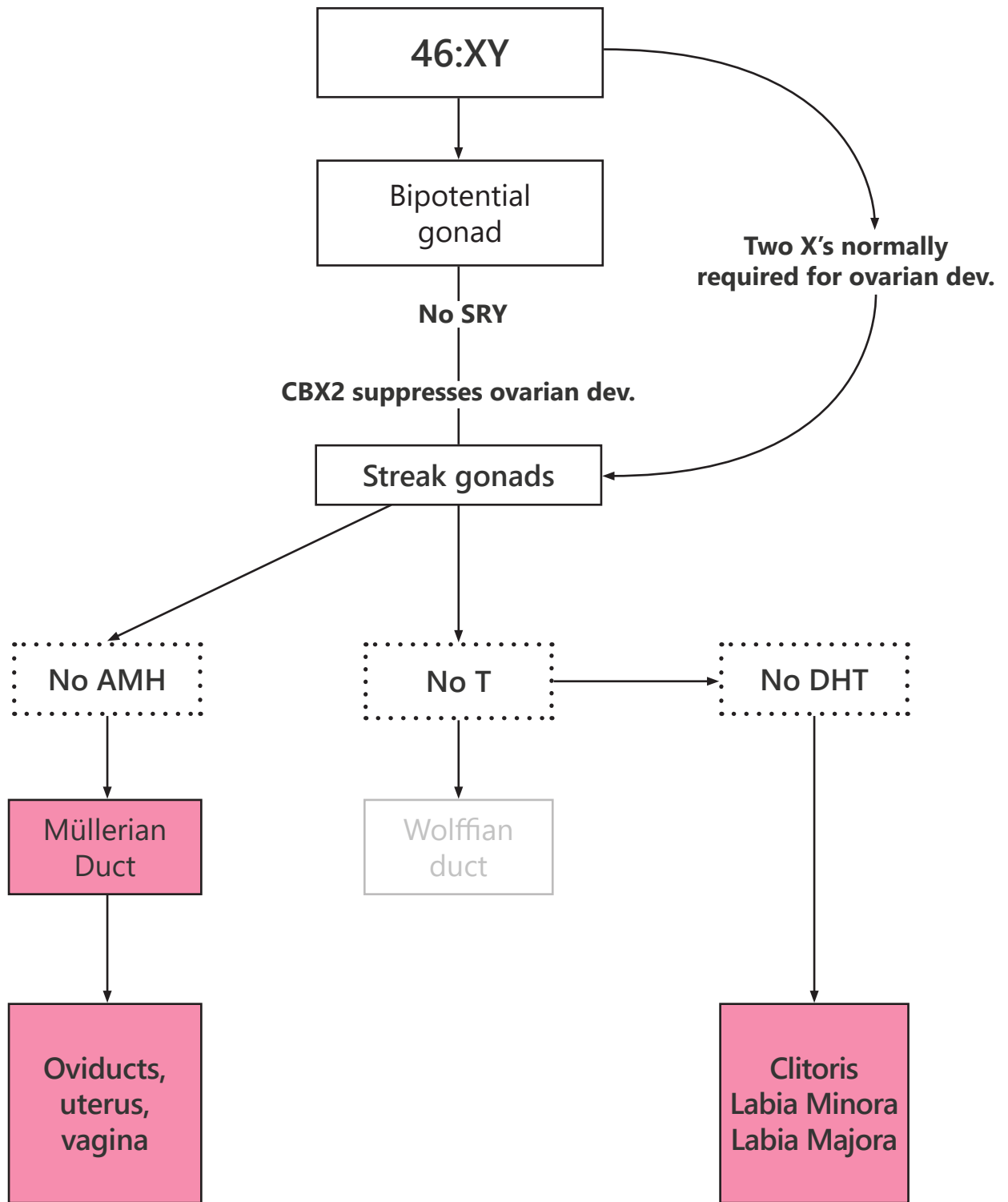
Male | Unknown birth rate | Often infertile



- [1] Rey, R., Josso, N., Racine, C. (2020). Sexual differentiation. In: *Endotext*. South Dartmouth, MDText, Inc.
- [2] NIH. (2020). Persistent mullerian duct syndrome. *Genetics Home Reference*, National Library of Medicine.
- [3] Witchel, S. (2018). Disorders of sex development. *Best Practice and Research in Clinical Obstetrics and Gynecology*.
- [4] Ogilvy-Stuart, A., et al. (2004). Determination of sex: Early assessment of ambiguous genitalia. *Arch Dis Child*.

SWYER SYNDROME

Female | 1 in 80,000 births | Always infertile



[1] Rey, R., Josso, N., Racine, C. (2020). Sexual differentiation. In: *Endotext*. South Dartmouth, MDText, Inc.

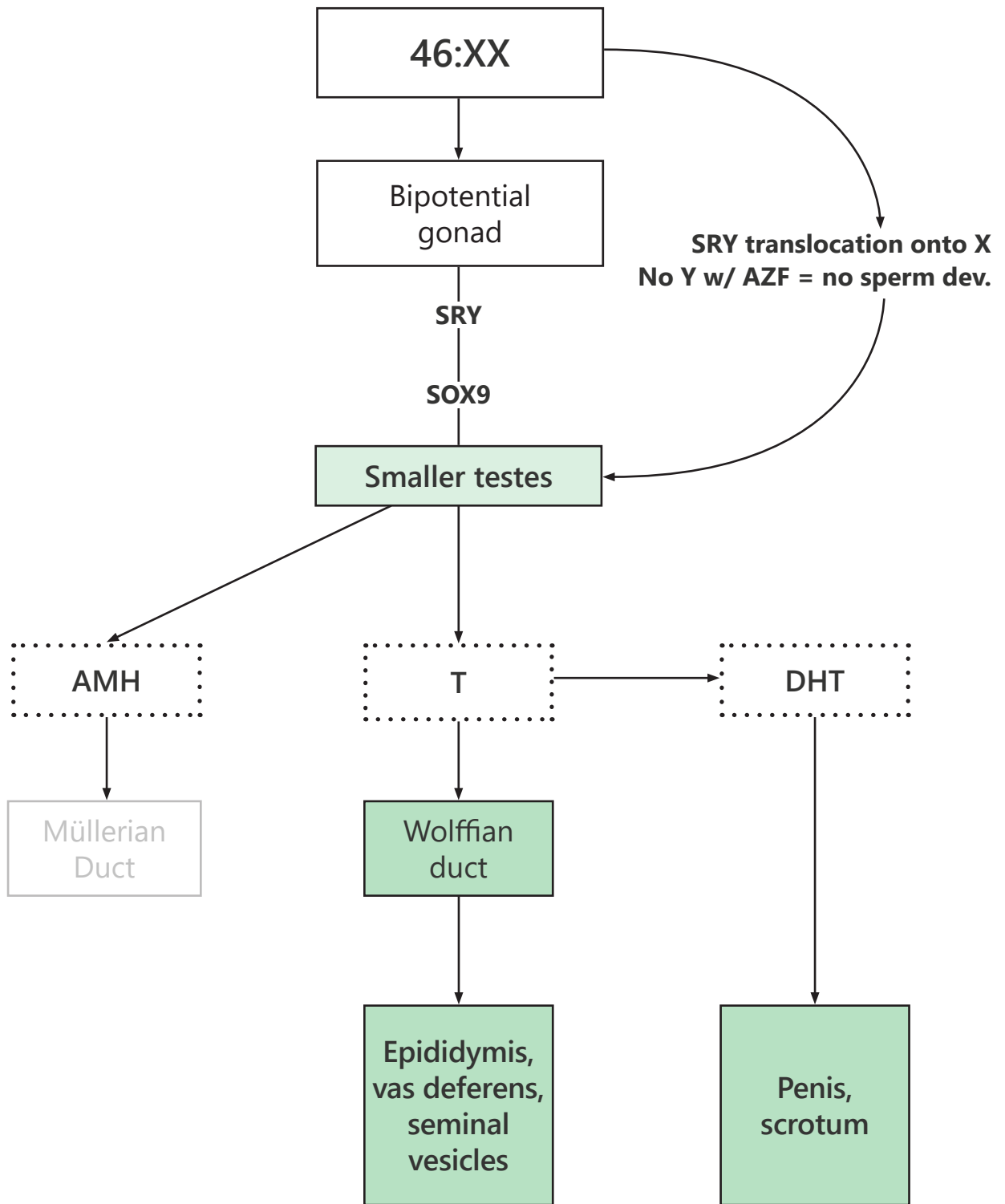
[2] NIH. (2020). Swyer Syndrome. *Genetics Home Reference*, National Library of Medicine.

[3] Witchel, S. (2018). Disorders of sex development. *Best Practice and Research in Clinical Obstetrics and Gynecology*.

[4] Ogilvy-Stuart, A., et al. (2004). Determination of sex: Early assessment of ambiguous genitalia. *Arch Dis Child*.

XX MALE SYNDROME

Male | 1 in 20,000 births | Always infertile



[1] Rey, R., Josso, N., Racine, C. (2020). Sexual differentiation. In: *Endotext*. South Dartmouth, MDText, Inc.

[2] NIH. (2020). 46:XX testicular disorder of sex development. *Genetics Home Reference*, National Library of Medicine.

[3] Witchel, S. (2018). Disorders of sex development. *Best Practice and Research in Clinical Obstetrics and Gynecology*.

[4] Ogilvy-Stuart, A., et al. (2004). Determination of sex: Early assessment of ambiguous genitalia. *Arch Dis Child*.