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Manejo individualizado de la insensibilidad a andrógenos: adaptando el tratamiento a cada persona

Grapsia

XXII ENCUENTRO GRAPSIA

Montse Amat Bou
Endocrinología Pediátrica
Hospital Sant Joan de Déu
Unidad DSD HSJD - HCB

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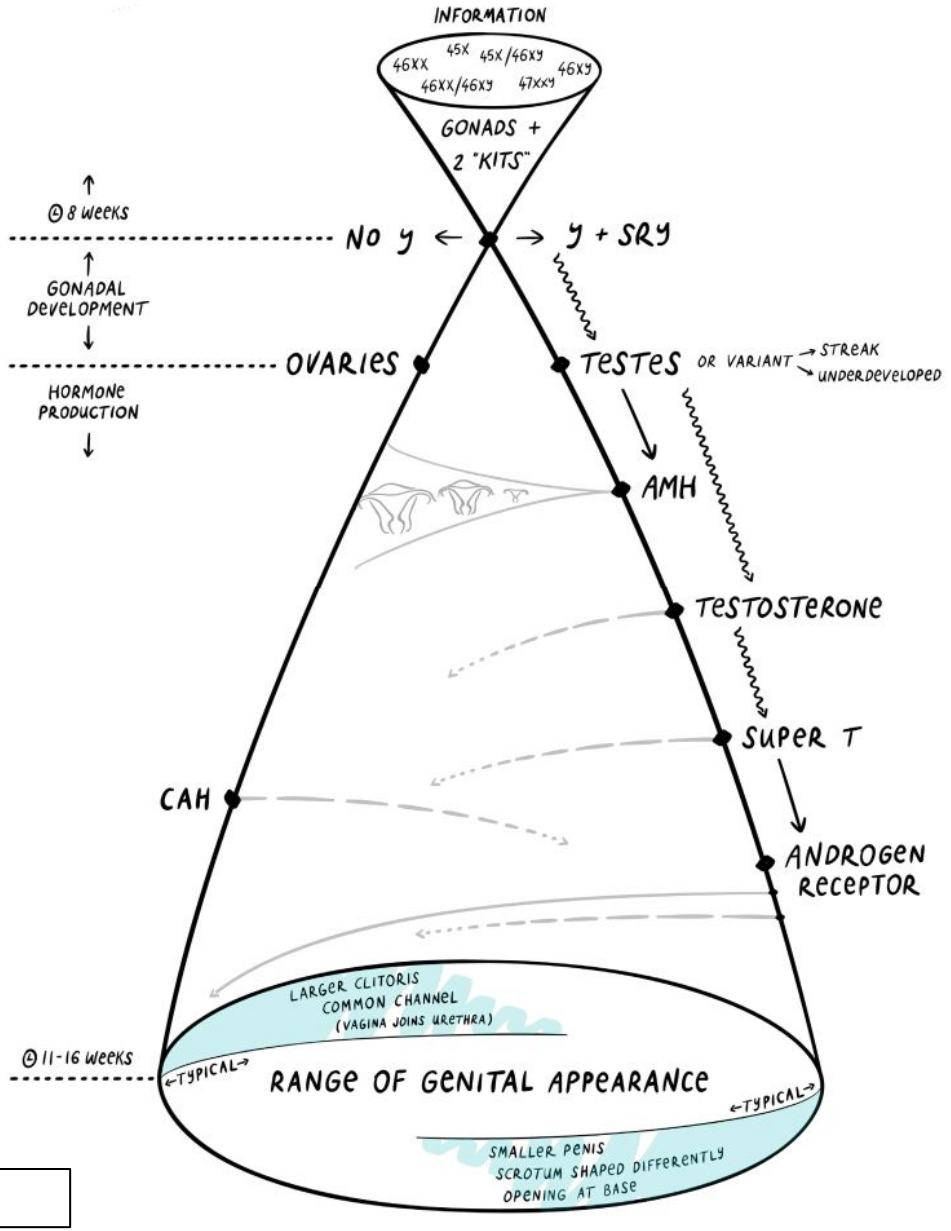
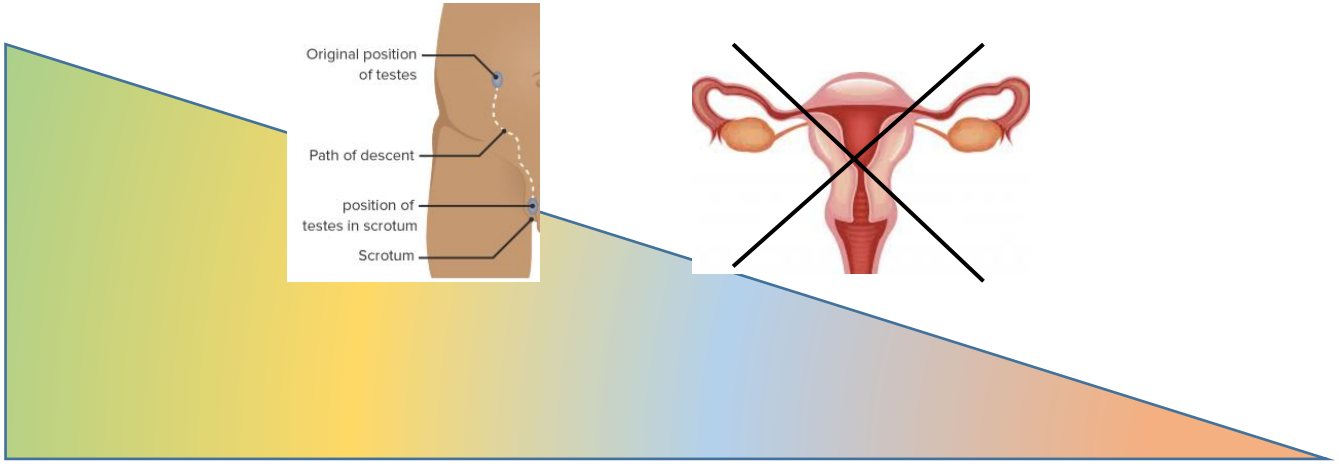
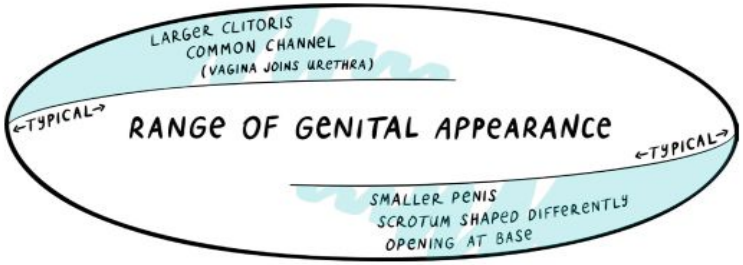


The Story of Sex development. DSD families.

Variabilidad según resistencia



CAIS PAIS MAIS



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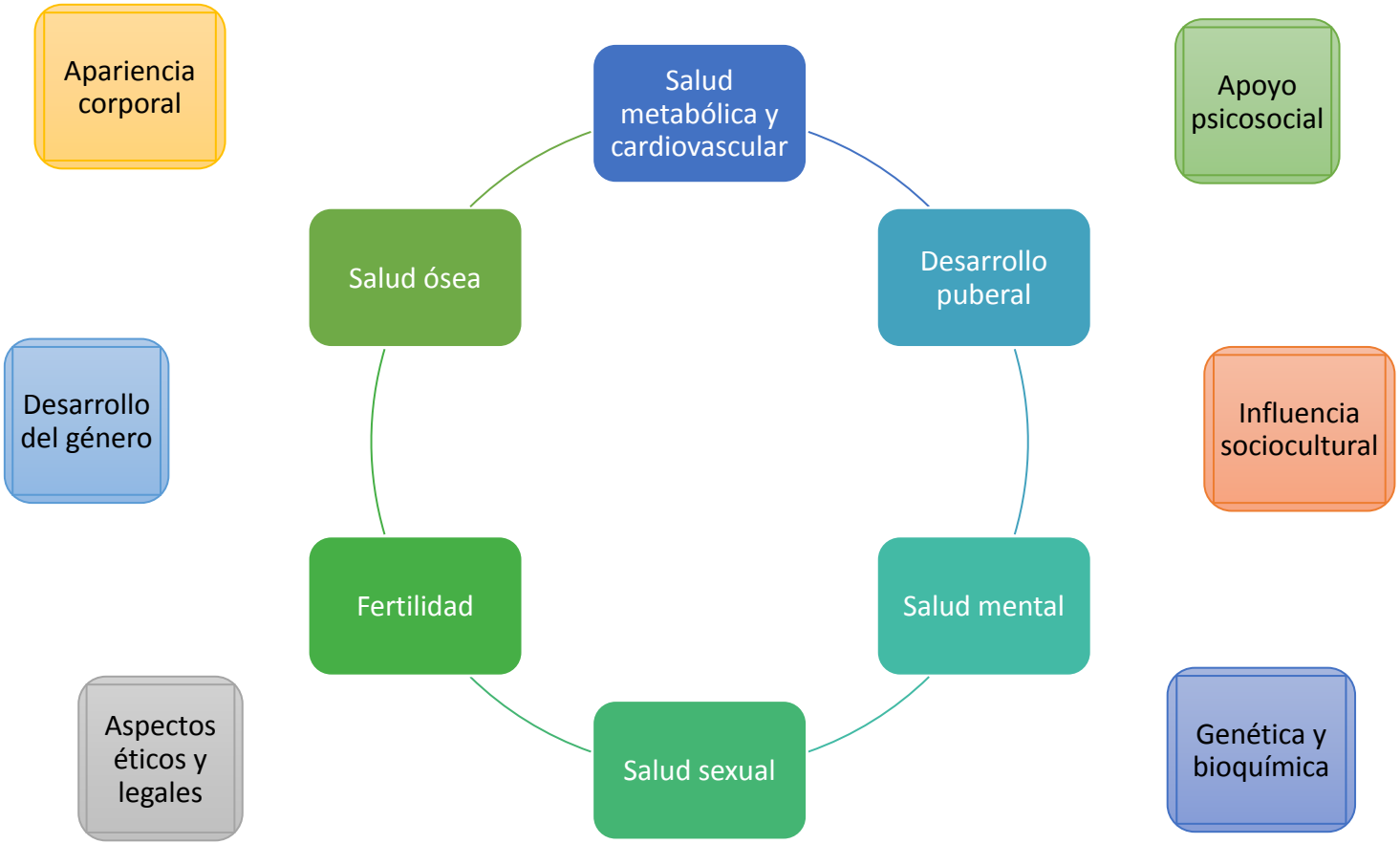
Manejo individualizado



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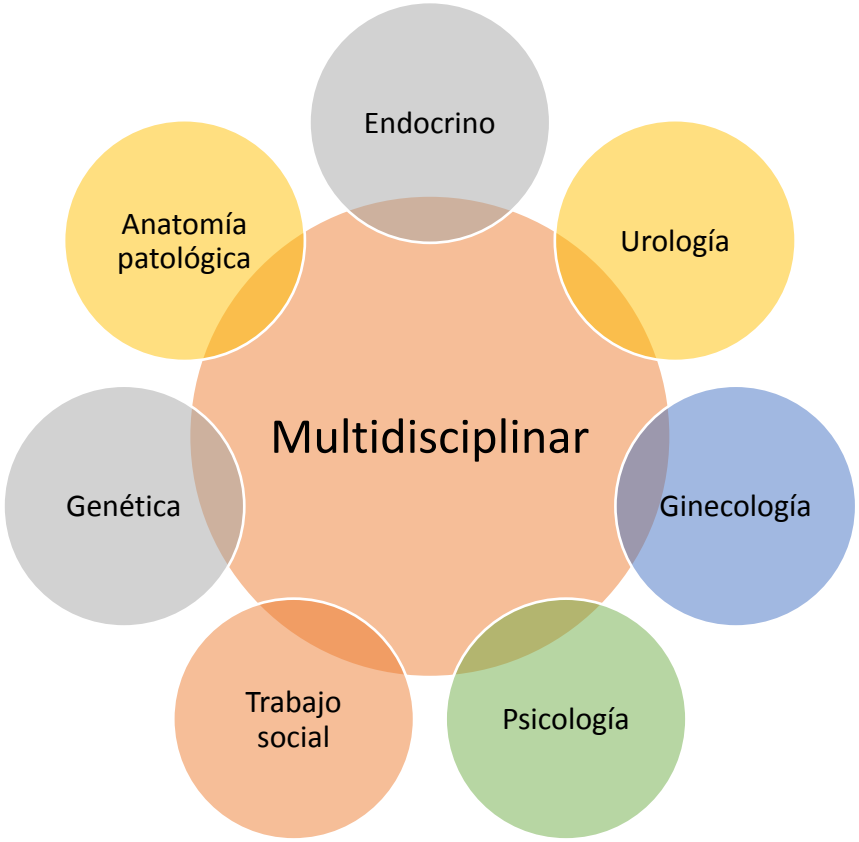
Enfoque holístico



Hiort O et al. Management of DSD. Nat Rev Endocrinol. 2014 Sep;10(9):520-9.

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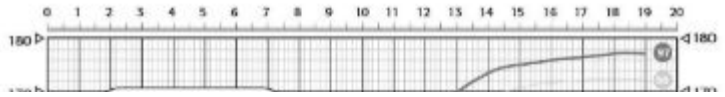
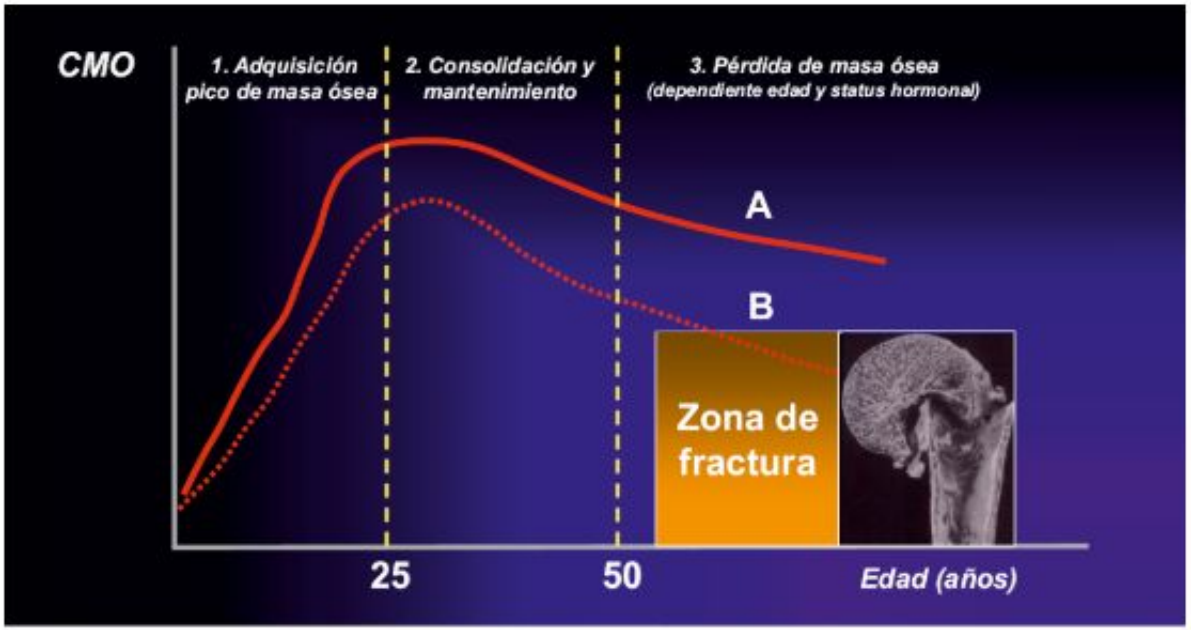
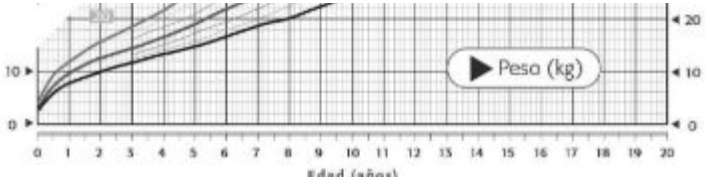
Desarrollo puberal

Caracteres sexuales secundarios







ración exual y social

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Desarrollo puberal

¿Inducción puberal?

- ¿Necesario?
- ¿Cuándo?
- ¿Qué tratamiento?

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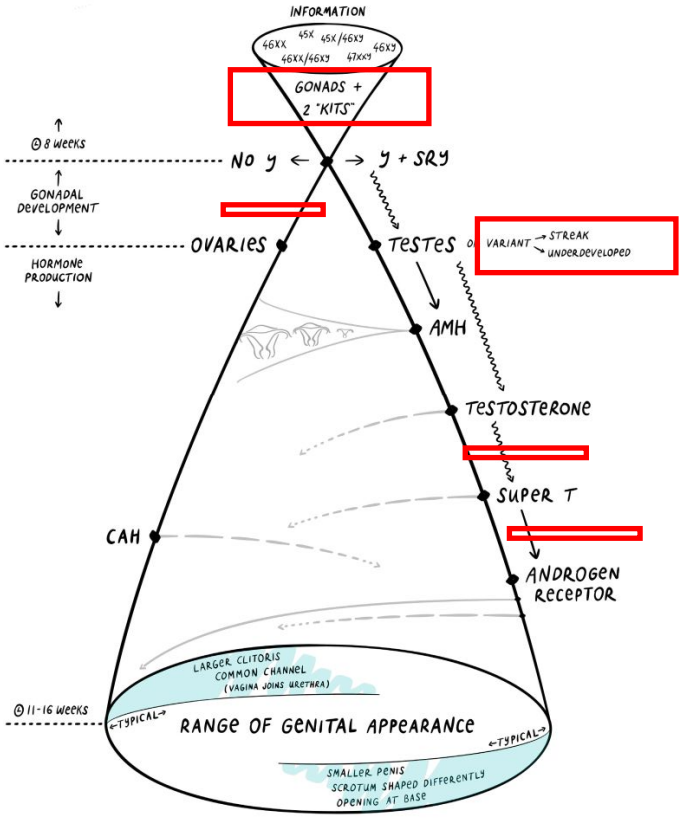
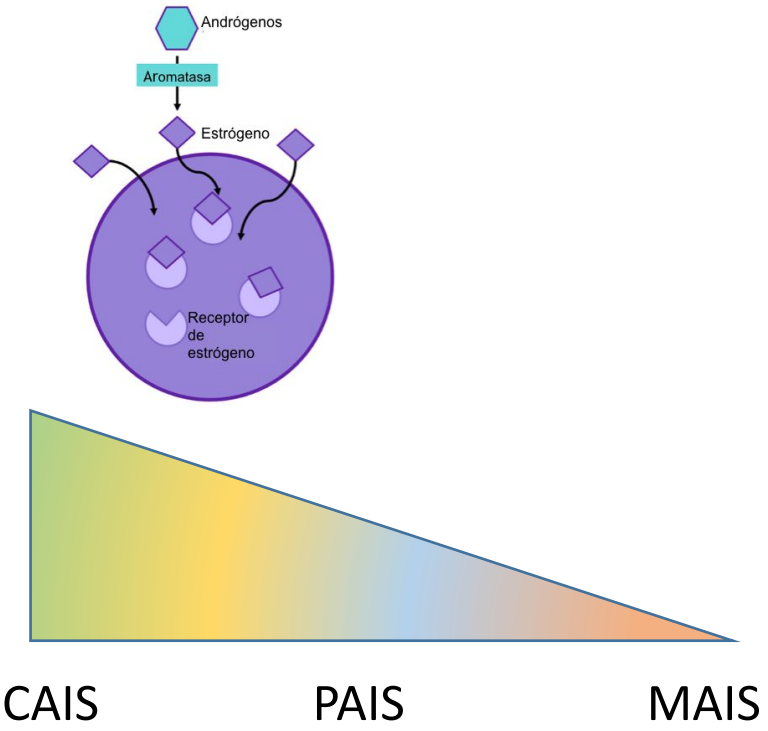
Inducción puberal





Inducción puberal

Clinical Practice Guideline | A Nordenström and others | Pubertal induction: a clinical guideline | 186:6 | G9–G49

Pubertal induction and transition to adult sex hormone replacement in patients with congenital pituitary or gonadal reproductive hormone deficiency: an Endo-ERN clinical practice guideline

A Nordenström[1], S F Ahmed[2], E van den Akker[3], J Blair[4], M Bonomi[5,6], C Brachet[7], L H A Broersen[8], H L Claahsen-van der Grinten[9], A B Dessens[10,11], A Gawlik[12], C H Gravholt[13,14], A Juul[15,16], C Krausz[17], T Raivio[18], A Smyth[19], P Touraine[20,21], D Vitali[22] and O M Dekkers[23,24]

Pubertal induction in CAIS

In whom to consider puberty induction?

R 6.1 We recommend allowing girls with CAIS and retain gonads to go through spontaneous puberty.

Timing of puberty induction, treatment approach and monitoring

R. 6.2 We recommend to start puberty induction at the age of 11 years in girls with CAIS who underwent gonadectomy. We recommend the same treatment protocols (for pubertal induction in girls with CAIS who have been gonadectomised) as for other girls (R2.2–2.3, R2.6–2.8, R2.11). (+OOO)

Considerar tratamiento si pubertad retrasada o falta de progresión puberal

*Pueden haber niveles bajos de E2 y DMO.

Inducción puberal

Clinical Practice Guideline | A Nordenström and others | Pubertal induction: a clinical guideline | 186:6 | G9–G49

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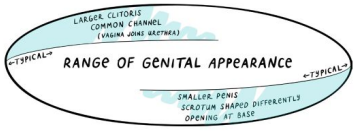
Pubertal induction in PAIS

In whom to consider puberty induction?

R. 7.1 We recommend that in all patients with PAIS, evaluation of gender identity should take place before considering puberty induction.

Evaluation of pubertal development

R. 7.2 We recommend that careful evaluation of patients' endocrine profile, phenotype and genotype is undertaken and that this information is used to consider the potential for virilisation during puberty (in patients with a male or undecided/binary identity).



Treatment approach and monitoring

R. 7.3 In children with PAIS identifying as as girls, the general recommendations for pubertal induction in CAIS as formulated under R 6.2–6.3 apply.

R 7.4 In boys with PAIS, we suggest considering additional testosterone treatment in mid puberty depending on the clinical and biochemical assessment of pubertal development. If clinical signs of hypoandrogenism such as micropenis and gynaecomastia are present, we suggest treating with the addition of testosterone for 6 months and then evaluating the effect. (+OOO)

Inducción puberal

Clinical Practice Guideline | A Nordenström and others | Pubertal induction: a clinical guideline | 186:6 | G9–G49

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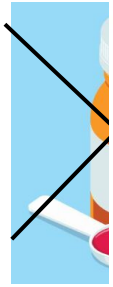
Imitar la pubertad

R 2.6 We recommend to use 17β-oestradiol for puberty induction or to sustain puberty in girls (++OO).



Preparation	Doses available	Starting dose of puberty	Increase approximately every 6 months* to adult dosing
E_2: transdermal options TD (some brands examples)		3–7 µg/day	25–100 µg/day
Menostar	14 µg	Part of patch twice weekly	Only used for low dosing situations, not fully hypogonadal replacement
Vivelle Dot	25, 37.5, 50, 75, 100 µg	Part of patch twice weekly or 1 patch per month (no patch for 3 weeks)**	25–100 µg twice weekly
Vivelle Mini	25, 37.5, 50, 75, 100 µg	Part of patch twice weekly or 1 patch per month (no patch for 3 weeks)**	25–100 µg twice weekly
Generic (different brands in different countries; e.g. Oesclim Estradot, Evorel, Systen, Climara, Demestril)	25, 37.5, 50, 75, 100 µg	Part of patch twice weekly or 1 patch per month (no patch for 3 weeks)**	25–100 µg twice weekly
Estraderm MX	25, 50, 100 µg	Part of patch twice weekly or 1 patch per month (no patch for 3 weeks)**	25–100 µg twice weekly

en plasma más
ıibilidad, disminuye el
o)

menos rápido la

che vs oral

Diferencias en el efecto en salud ósea y función sexual comparado con otras personas

Inducción puberal

¿Testosterona?

Randomized Controlled Trial > Lancet Diabetes Endocrinol. 2018 Oct;6(10):771-780.
doi: 10.1016/S2213-8587(18)30197-9. Epub 2018 Jul 31.

Oestrogen versus androgen in hormone-replacement therapy for complete androgen insensitivity syndrome: a multicentre, randomised, double-dummy, double-blind crossover trial

Wiebke Birnbaum [1], Louise Marshall [1], Ralf Werner [1], Alexandra Kulle [2], Paul-Martin Holterhus [2], Katharina Rall [3], Birgit Köhler [4], Annette Richter-Unruh [5], Michaela F Hartmann [6], Stefan A Wudy [6], Matthias K Auer [7], Anke Lux [8], Siegfried Kropf [8], Olaf Hiort [9]

¿Gonadectomía?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DOCUMENTO DE CONSENSO

Guía de consenso sobre la gonadectomía profiláctica en el desarrollo sexual diferente

Julio Guerrero-Fernández[a,b,*], Pilar González-Peramato[c], Amaia Rodríguez Estévez[d], María José Alcázar Villar[a,e], Laura Audí Parera[a,f], María Cristina Azcona San Julián[a,g], Atilano Carcavilla Urquí[a,b], Luis Antonio Castaño González[a,h], José María Martos Tello[a,i], Cristina Mora Palma[a,b], María Francisca Moreno Macián[a,j], Diego Yeste Fernández[a,k] y Manuel Nistal[l]

***CAIS*: Bajo riesgo de TCG** (riesgo variable entre estudios: 1-3% con un riesgo acumulado de un 3,6% a los 25 años, y 33% a los 50 años, otros autores estiman un riesgo del 15% pasada la pubertad

Infancia y pubertad INFORMACIÓN

Adolescencia/edad adulta

¿Preferencia?

No tenemos herramientas no invasivas para detección precoz

Riesgo 10-15%

Riesgo de progresión bajo

Necesidad de THS para siempre

¿Gonadectomía?


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***PAIS*: Intermedio- Alto riesgo** (15 -20%). Menor, si localización escrotal; estudios recientes reducen este riesgo sustancialmente.

Infancia y pubertad

INFORMACIÓN

Identidad de género

Adolescencia/edad adulta

¿Preferencia?

No tenemos herramientas no invasivas para detección precoz

Riesgo 10-15% (depende de la localización)

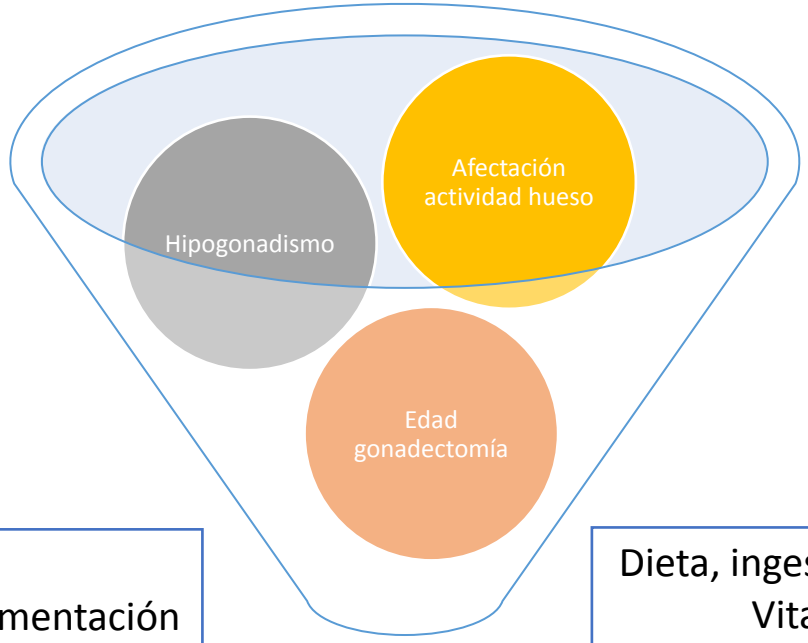
Virilización

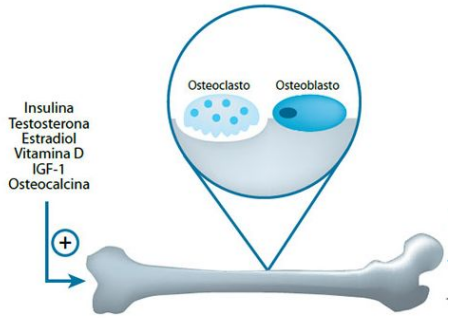
Riesgo de progresión bajo

Necesidad de THS para siempre

Salud ósea

Disminución INSL-3
Aumento de FSH
THS inadecuado



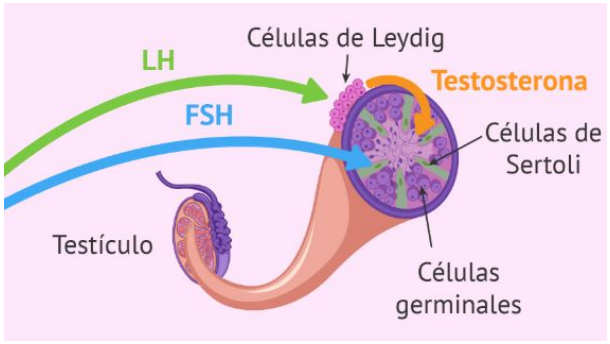


THS
Tipo, dosis, cumplimentación

Dieta, ingesta de calcio, actividad física, Vitamina D, tabaquismo

Empeoramiento de salud ósea en CAIS

Fertilidad



CAIS — PAIS — MAIS

Hiperplasia de células de Leydig
Aumento de fibrosis
Pocas espermatogonias
Espermatogénesis incompleta

Acción residual del RA

Manejo multidisciplinar

Xarxes d'unitats d'expertesa clínica (XUEC) en malalties minoritàries (MM)

Desenvolupament i maduració sexual

Centros, Servicios y Unidades de Referencia del Sistema Nacional de Salud

Desarrollo sexual diverso

SJD Sant Joan de Déu
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Gracias.

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