



Dirección General de Humanización,
Atención y Seguridad del Paciente
CONSEJERÍA DE SANIDAD

El Síndrome de Turner: Detección, Atención y Necesidades de las pacientes

Fecha: 27 de octubre 2025

Horario: 16:00 h a 20:00 h

Modalidad: Presencial

Lugar: Pº de la Castellana nº 280 (SERMAS)
Aula 0-Rs1

Cómo llegar

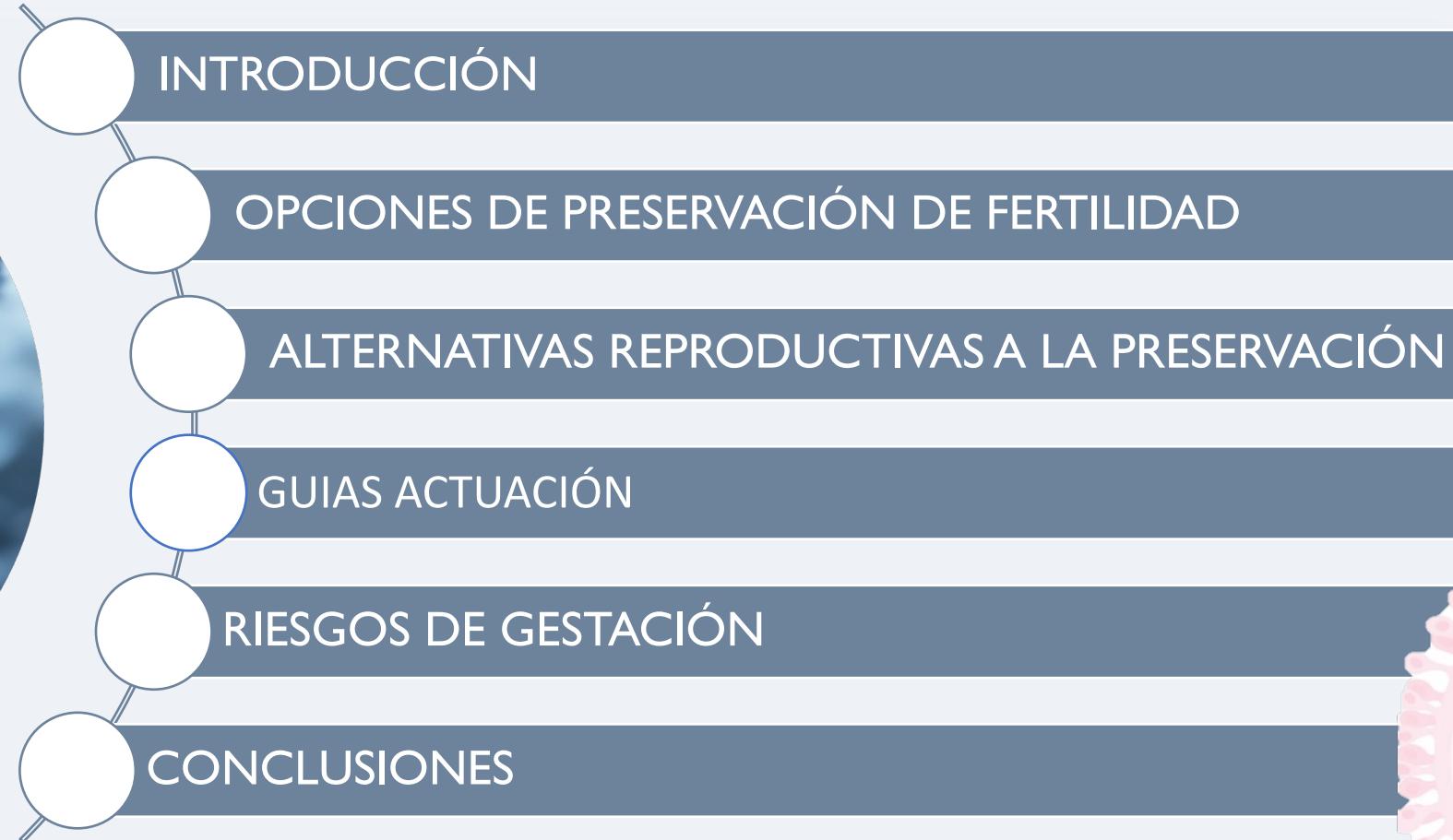


OPCIONES REPRODUCTIVAS EN EL SINDROME DE TURNER

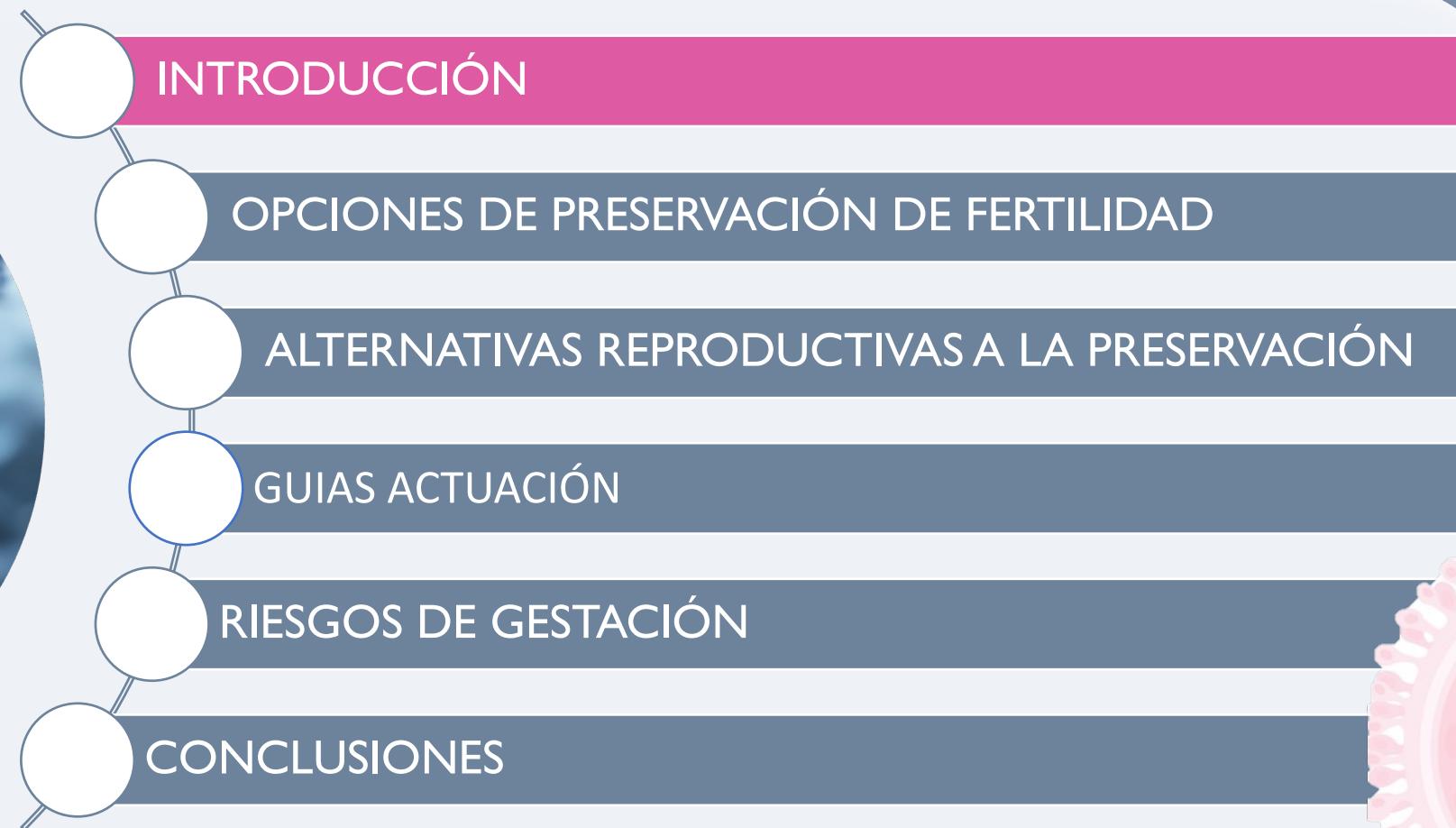
Dra. SILVIA INIESTA PÉREZ
Jefa de Sección Ginecología y Obstetricia
Reproducción Asistida

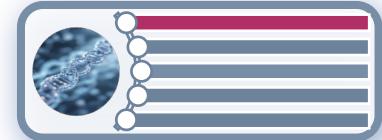
 **Hospital Universitario
La Paz**
Hospital Carlos III
Hospital Cantoblanco

Opciones reproductivas en pacientes con SdR Turner



Opciones reproductivas en pacientes con SdR Turner

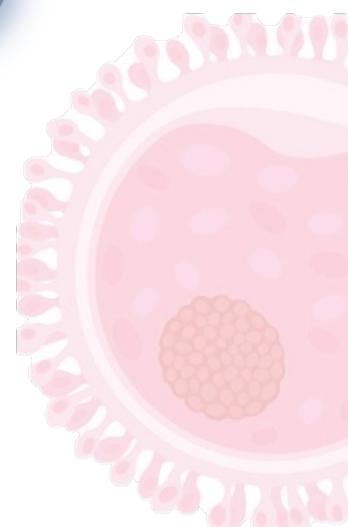


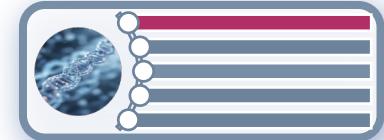


Características del SdR de Turner

- Anomalía cromosómica común

Alteración genética que afecta aproximadamente a 1 de cada 2.000-2.500 mujeres nacidas vivas, caracterizada por la ausencia completa o parcial de un cromosoma X.

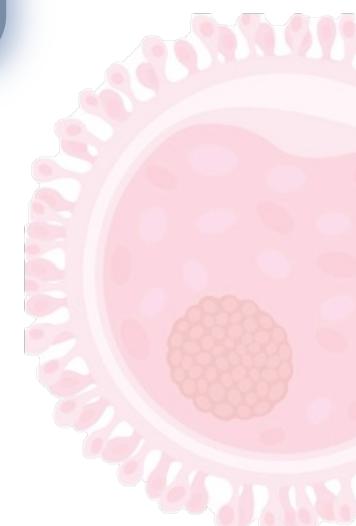


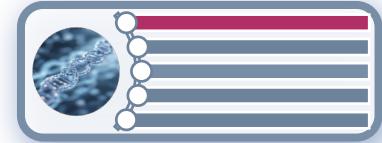


Características del Sdr de Turner

- Anomalía cromosómica común
- Variabilidad genotípica

45 XO completa: disgenesia ovárica. IOP temprana
Mosaico (45X0/46XX): reserva residual variable
Delecciones Xq: asociadas con IOP

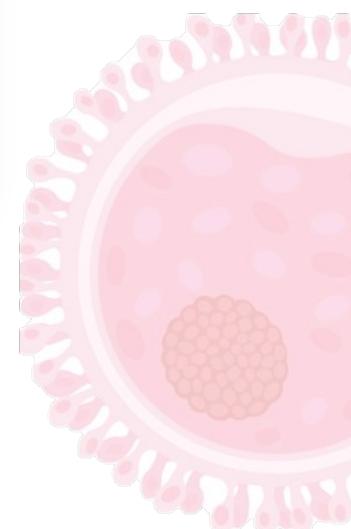


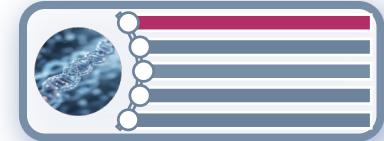


Características del SdR de Turner

- Anomalía cromosómica común
- Variabilidad genotípica
- Variabilidad clínica

Las manifestaciones clínicas varían significativamente según el cariotipo, siendo generalmente menos severas en mujeres con cariotipo mosaico





Características del SdR de Turner

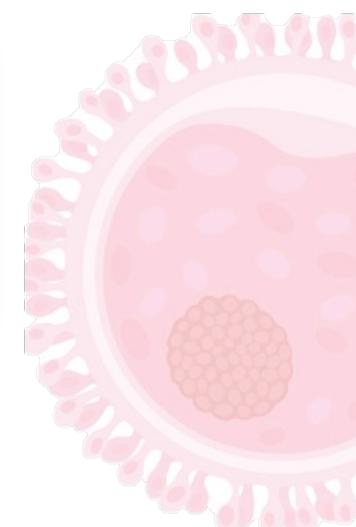
- Anomalía cromosómica común
- Variabilidad genotípica
- Variabilidad clínica
- Insuficiencia ovárica prematura / infertilidad

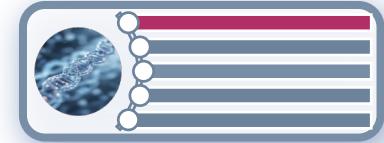
Riesgo extremadamente alto

70-80% sin desarrollo puberal espontáneo

90% presentan amenorrea primaria

Reserva ovárica agotada antes de la edad adulta (mayoría)



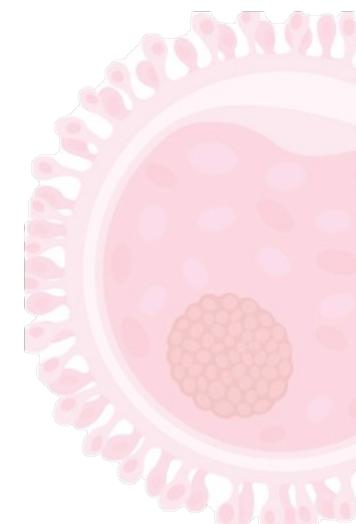


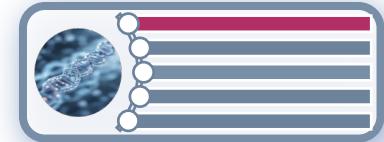
Características del SdR de Turner

- Anomalía cromosómica común
- Variabilidad genotípica
- Variabilidad clínica
- Insuficiencia ovárica prematura / infertilidad
- Mecanismos de pérdida folicular

Células germinales disminuyen significativamente a mitad de la gestación

Apoptosis acelerada de las células germinales





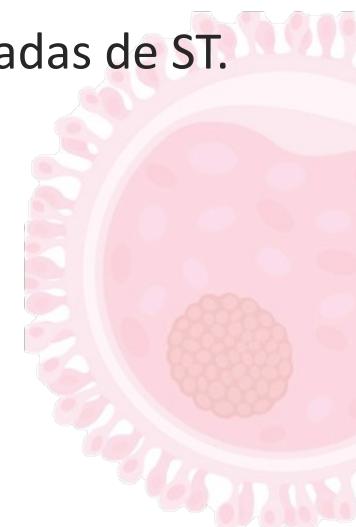
Características del Sdr de Turner

Embarazos espontáneos

<10% (3-5%)

De pacientes con ST logran un embarazo de forma natural

La infertilidad representa un desafío persistente y significativo para las mujeres diagnosticadas de ST.



Reproductive epidemiology

Turner syndrome: fertility, familial clustering, and cancer risk

K. Allen-Brady¹, L.E. Verrilli^{2,3}, B.A. Austin⁴, J.M. Letourneau¹, E.B. Johnstone², and C.K. Welt^{1,4,*}

¹Division of Epidemiology, Department of Internal Medicine, University of Utah School of Medicine, Salt Lake City, UT, USA

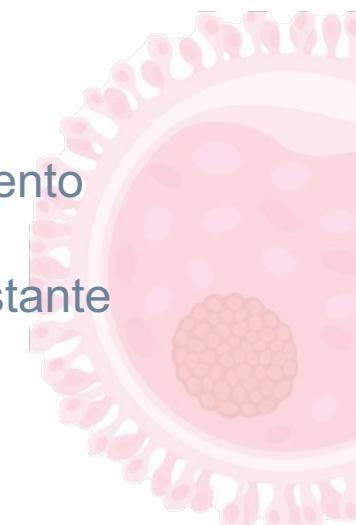
²Division of Reproductive Endocrinology and Infertility, Department of Obstetrics and Gynecology, University of Utah School of Medicine, Salt Lake City, UT, USA

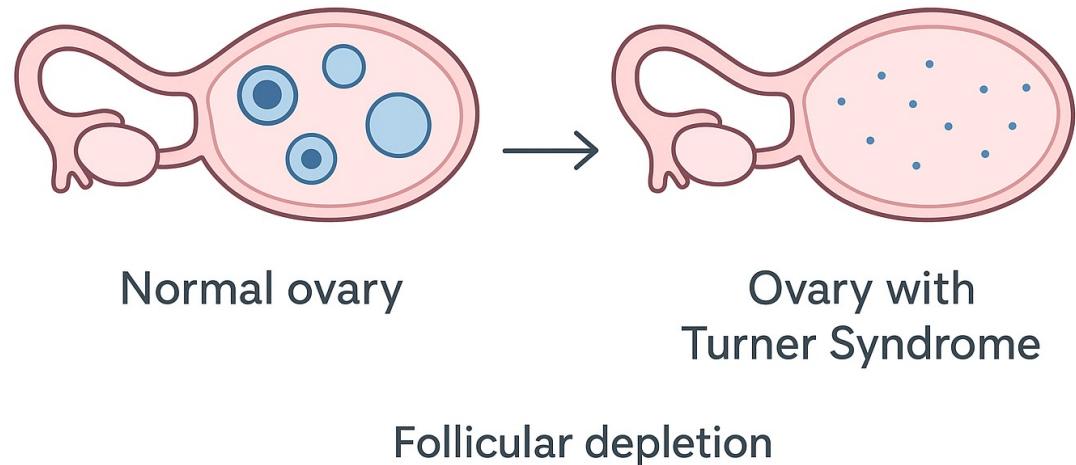
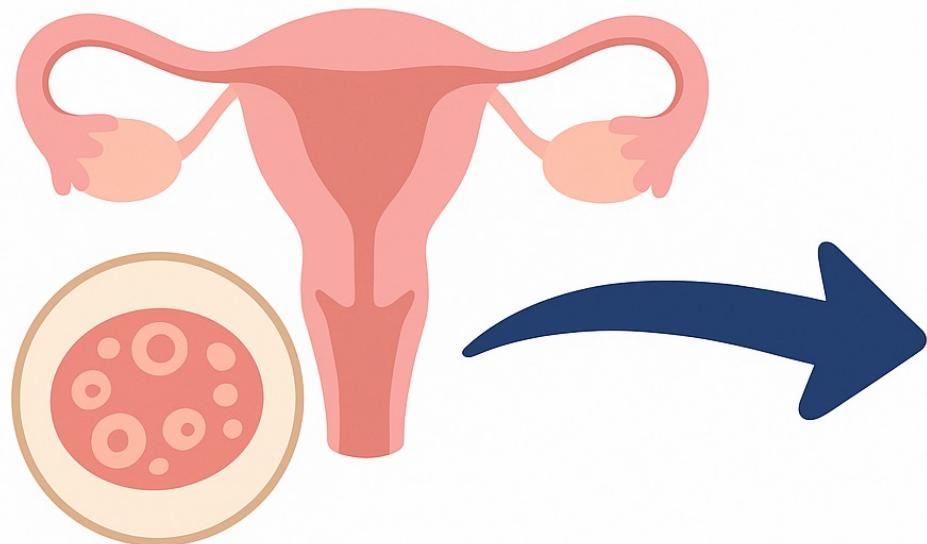
³Intermountain Health, Department of Obstetrics and Gynecology, Murray, UT, USA

⁴Division of Endocrinology, Metabolism and Diabetes, Department of Internal Medicine, University of Utah School of Medicine, Salt Lake City, UT, USA

*Correspondence address. Division of Endocrinology, Metabolism and Diabetes, Department of Internal Medicine, University of Utah School of Medicine, 15 N 2030 E, 2110A, Salt Lake City, UT 84112, USA. E-mail: cwelt@genetics.utah.edu <https://orcid.org/0000-0002-8219-5504>

Fertilidad y Nacimientos Vivos: Menos del 5% de las mujeres con ST tuvieron hijos. Las mujeres con ST que tuvieron un nacido vivo documentado mediante certificados de nacimiento tenían **cariotipos mosaico**. De los nacimientos observados, el 60% se logró mediante embarazos no asistidos (en mujeres de 20 a 30 años con cariotipos mosaico), y el 40% restante tuvo un hijo mediante **donación de ovocitos**.





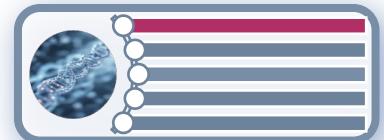
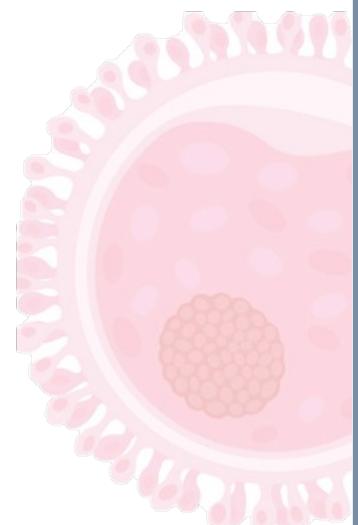
**APOPTOSIS FOLICULAR EXTENSA Y
ACELERADA**

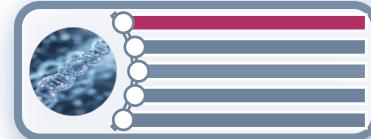
INSUFICIENCIA OVÁRICA PREMATURA

Sólo 1/2 pubertad espontánea

<10% gestación espontánea

Doble tasa de aborto

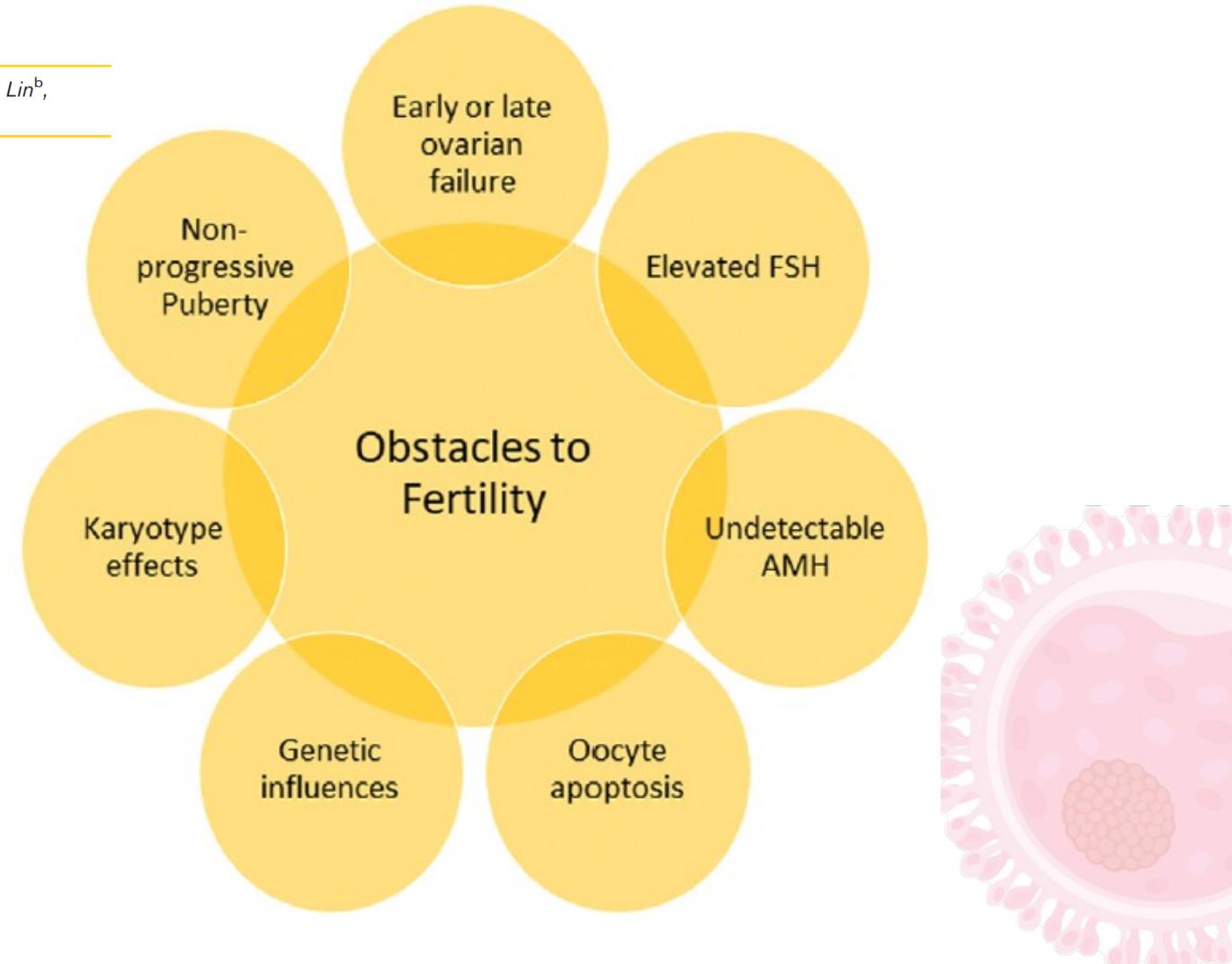


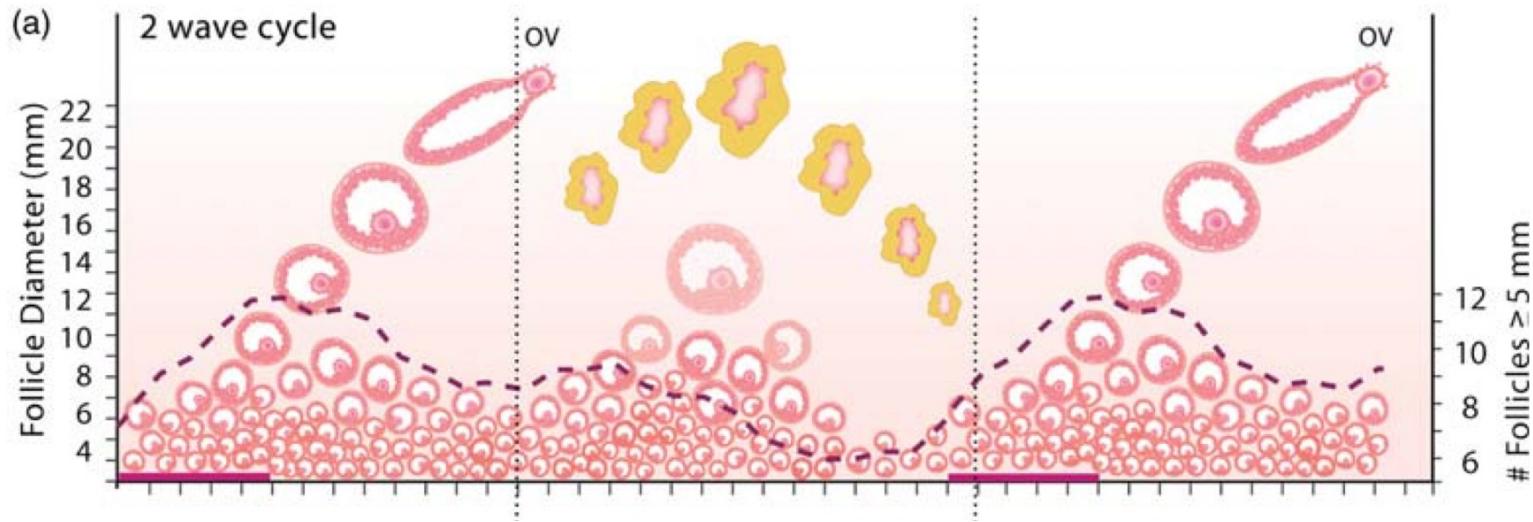
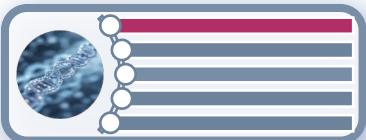
CURRENT
OPINION

Turner syndrome: fertility counselling in childhood and through the reproductive lifespan

Kassie J. Bollig^a, Monica Mainigi^a, Suneeta Senapati^a, Angela E. Lin^b,
Lynne L. Levitsky^c and Vaneeta Bamba^d

PÉRDIDA ACELERADA DE FOLÍCULOS + DIAGNÓSTICO TARDÍO





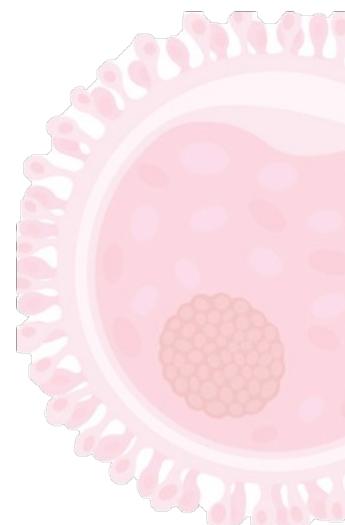
Human Reproduction Update, Vol.18, No.1 pp. 73-91, 2012

Advanced Access publication on November 8, 2011 doi:10.1093/humupd/dmr039

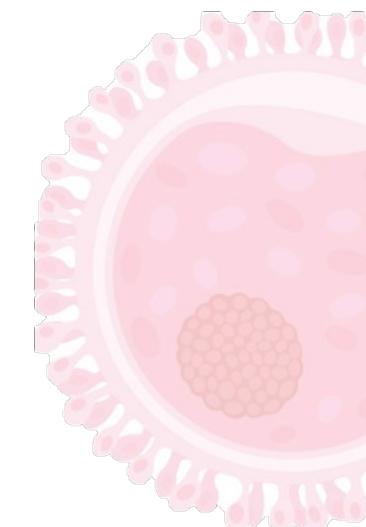
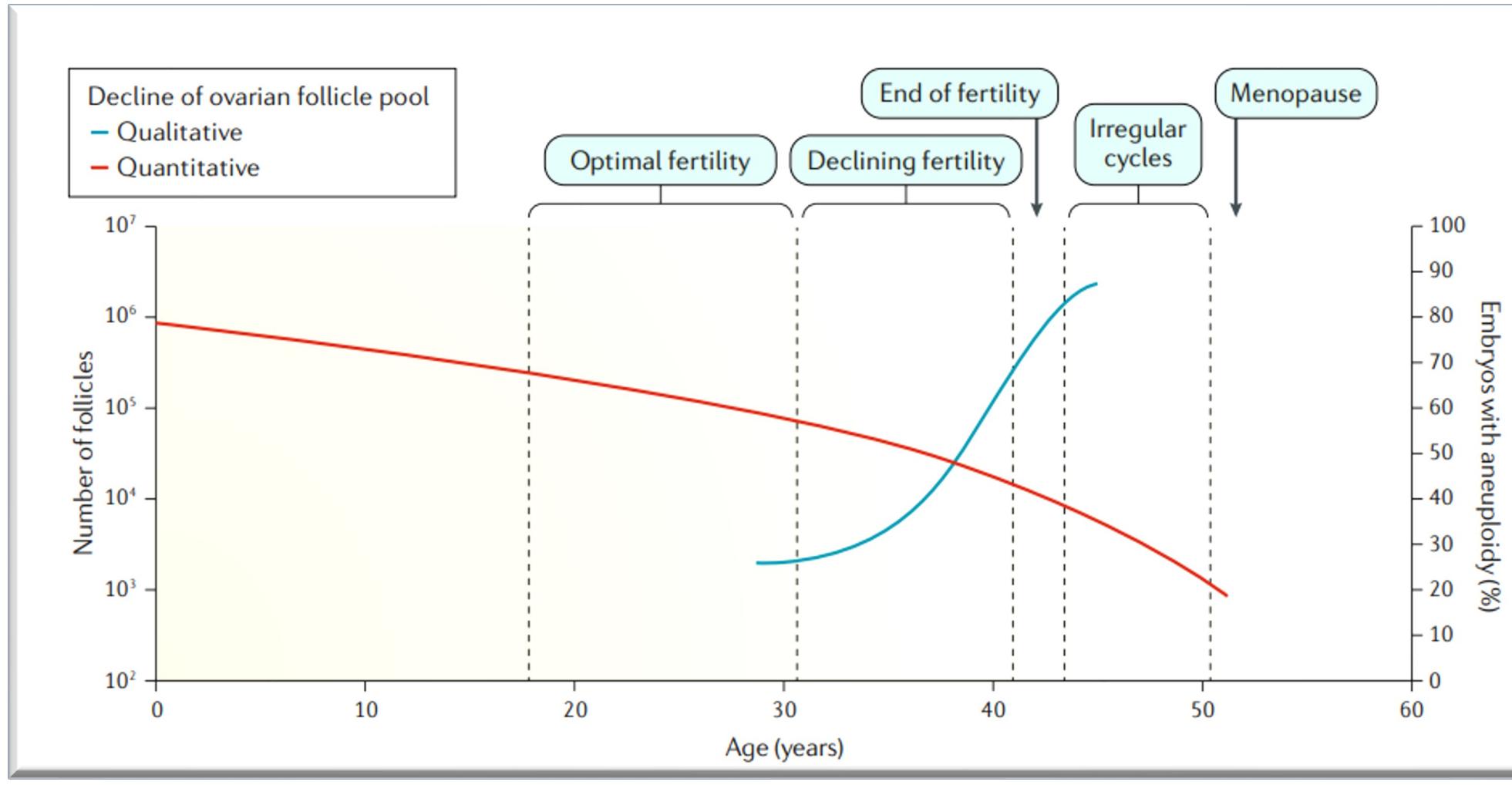
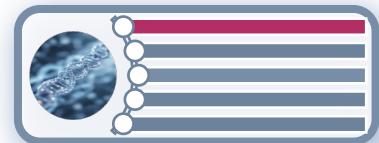
human
reproduction
update

Ovarian antral folliculogenesis during the human menstrual cycle: a review

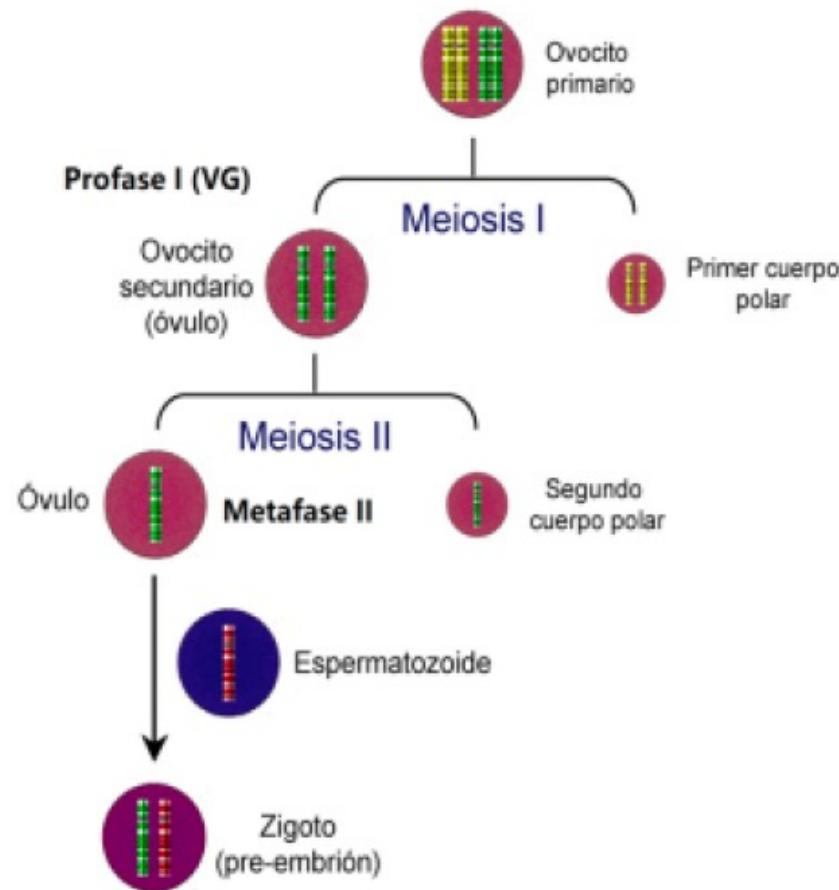
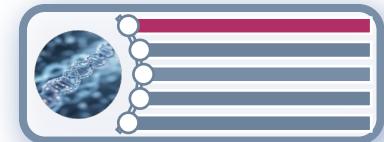
Angela R. Baerwald^{1,*}, Gregg P. Adams², and Roger A. Pierson³



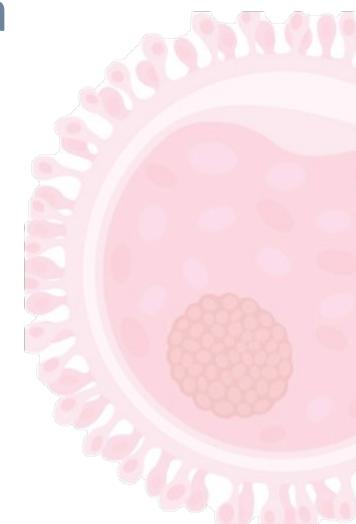
La fertilidad disminuye con el aumento de la edad



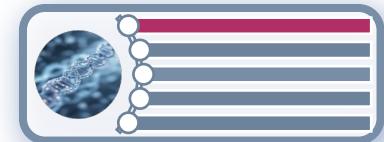
La mayoría de las anomalías cromosómicas de los embriones se producen por errores en la meiosis del ovocito



- Aneuploidías son más frecuentes conforme aumenta la edad materna.
- Pérdida de eficacia en la maquinaria que procesa los cíasmas y la segregación de cromosomas.



Implicación de los gametos en los errores cromosómicos

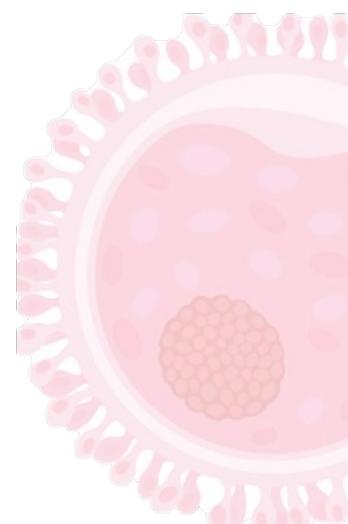


Human Molecular Genetics, 2007, Vol. 16, Review Issue 2 R203–R208
doi:10.1093/hmg/ddm243

The origin of human aneuploidy: where we have been, where we are going

Terry Hassold*, He

	Materna	Paterna	Embrionaria (Mitosis)
Trisomía 13	88%	12%	-
Trisomía 16	100%	-	-
Trisomía 18	89%	-	11%
Trisomía 21	88%	8%	3%
Trisomía 22	97%	3%	-
Trisomía XXX	76%	6%	18%
Trisomía XXY	51%	46%	3%
Trisomía XYY	-	60%	40%
Monosomía X	20%	80%	-



Turner más implicado error en meiosis en gameto masculino

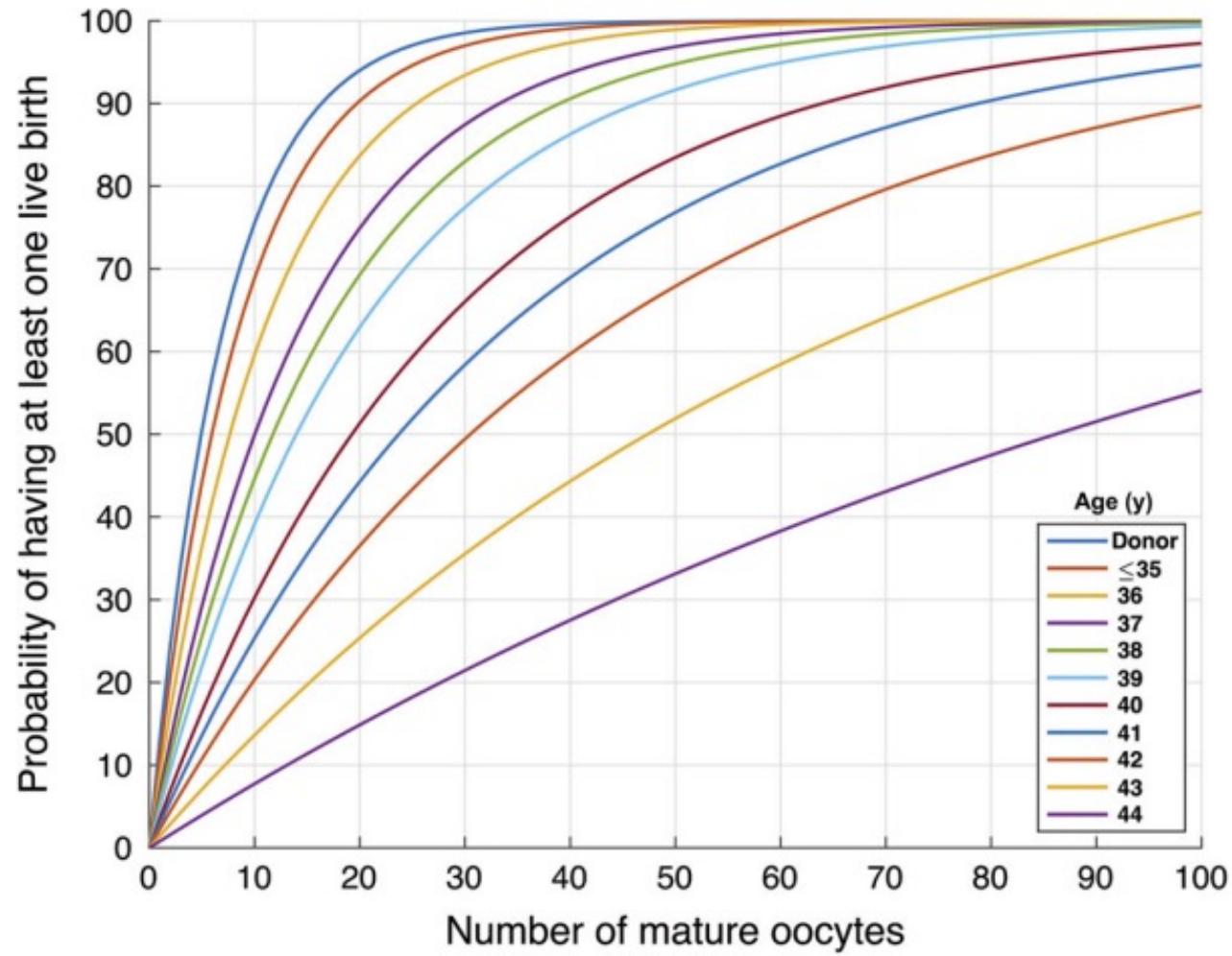
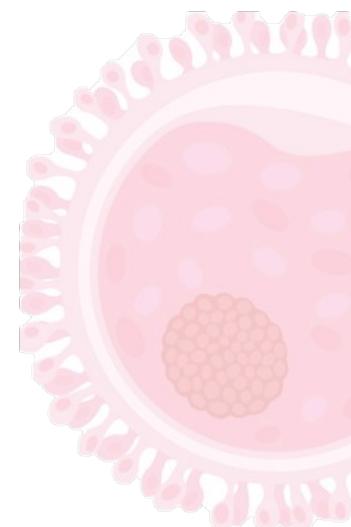
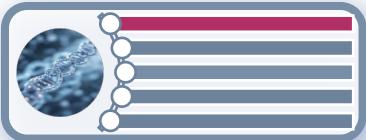


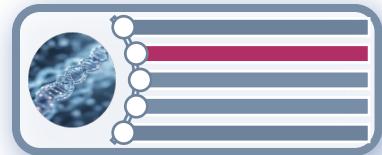
Figure 1 Live birth predictions by age and number of mature oocytes retrieved. Each curve shows the percent likelihood that a patient of a given age will have at least one live birth according to Equation 2, based on the number of mature oocytes retrieved and frozen.



Opciones reproductivas en pacientes con Sdr Turner



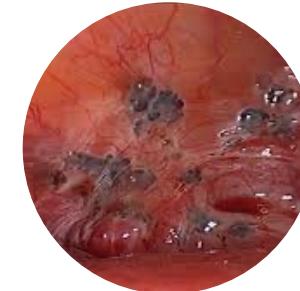
RESERVA FERTILIDAD



SOCIAL

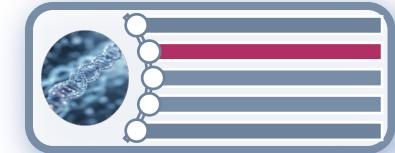


MOTIVOS MÉDICOS
ONCOLÓGICOS



MOTIVOS MÉDICOS NO
ONCOLÓGICOS

Indicaciones de preservación de fertilidad de causa médica no oncológica



- Pacientes con alto riesgo de FALLO OVÁRICO PRECOZ
- ENDOMETRIOSIS
- Otras indicaciones de CIRUGÍA OVÁRICA (quistes dermoides / tumores borderline)
- ENF AUTOINMUNES que precisan quimioterápicos (ciclofosfamida)
- TMO en talasemias, anemia de Fanconi, anemia de células falciforme: aplasia medular como parte del tratamiento y riesgo de fallo ovárico en alto porcentaje de los casos
- ALTERACIONES CROMOSÓMICAS: **Sdr de TURNER** (20% tienen función folicular, preservación en pacientes mosaico) // Portadoras de mutación FMR1 (X frágil), consejo genético // Mutaciones BRCA 1 o 2, menor reserva y cirugía profiláctica //Galactosemia





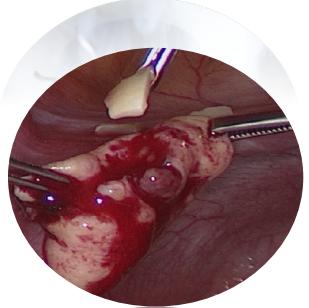
VITRIFICACIÓN OVOCITOS



CRIOPRESERVACIÓN TEJIDO
OVÁRICO

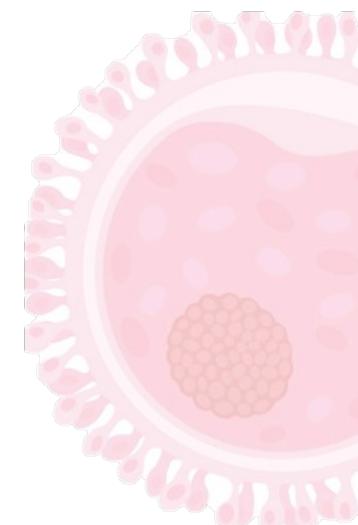
CRIOPRESERVACIÓN DE EMBRIONES

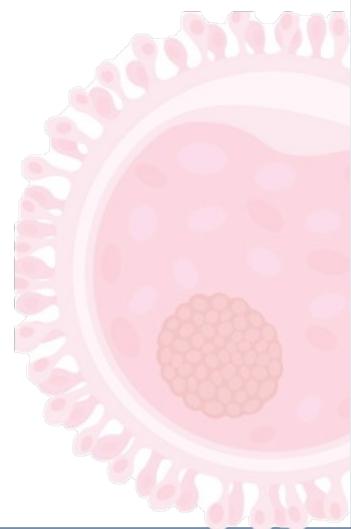
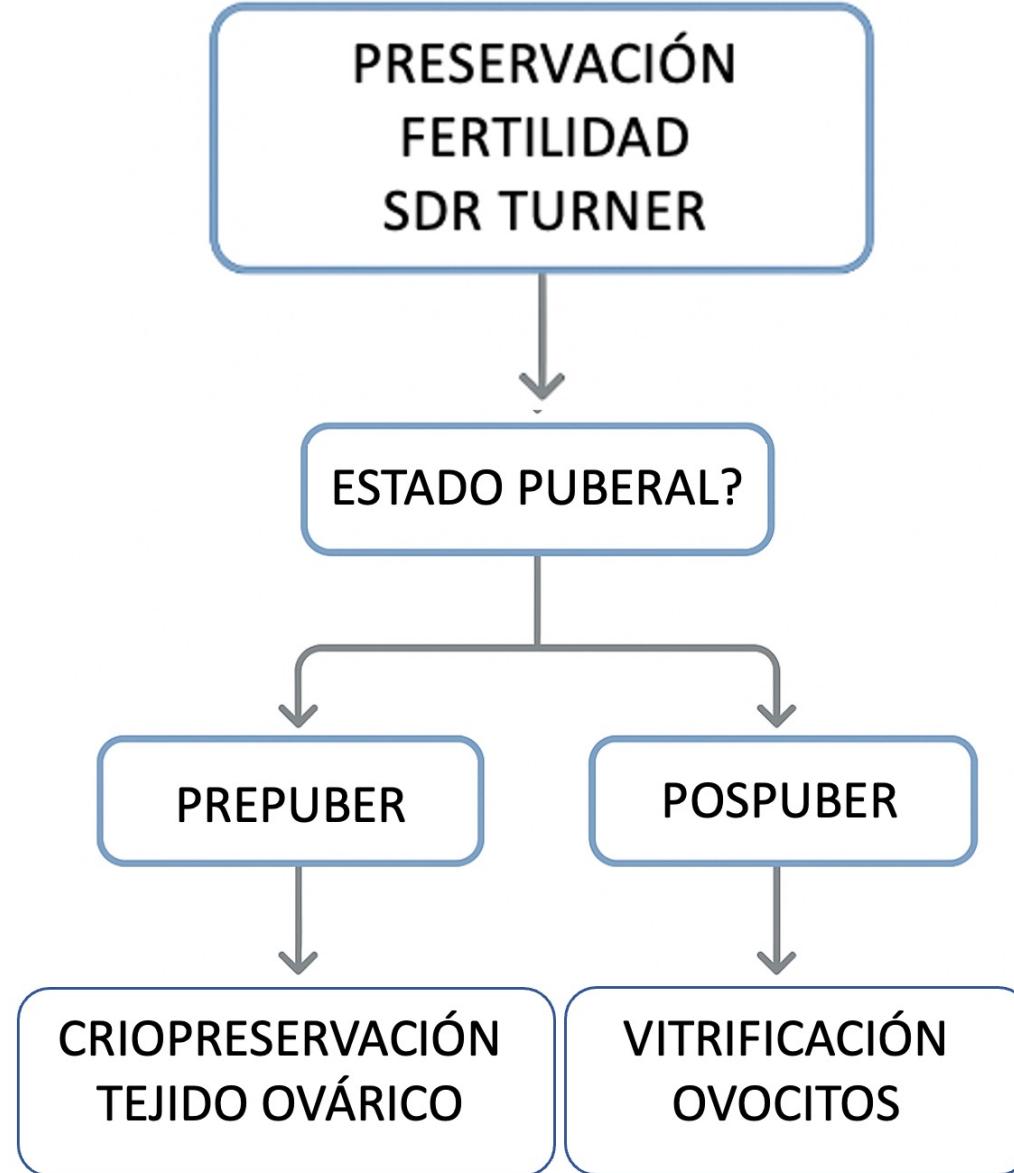
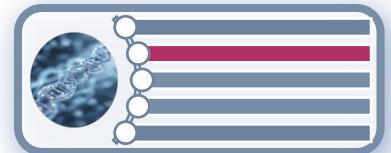


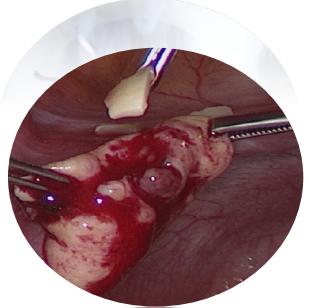


VITRIFICACIÓN OVOCITOS

CRYOPRESERVACIÓN TEJIDO
OVÁRICO

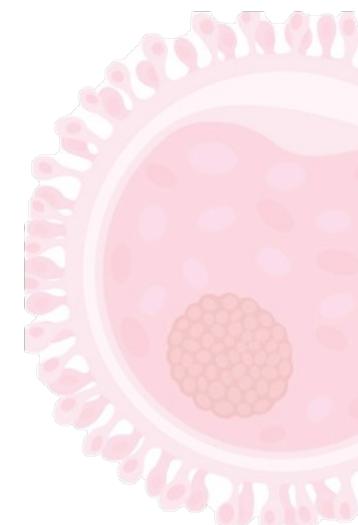






VITRIFICACIÓN OVOCITOS

CRYOPRESERVACIÓN TEJIDO
OVÁRICO

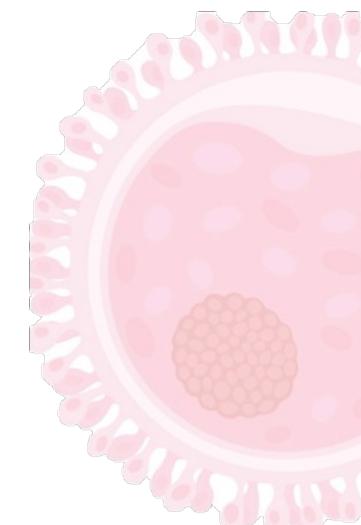
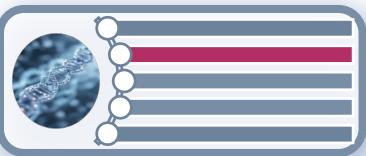


VITRIFICACIÓN OVOCITOS

- PRECISA FUNCIÓN OVÁRICA RESIDUAL
- EDAD, MENARQUIA
- SOLO GAMETO!



- TÉCNICA NO EXPERIMENTAL
- RESULTADOS CONTRASTADOS
- POSIBILIDAD DE PGT A EMBRIONES A FUTURO



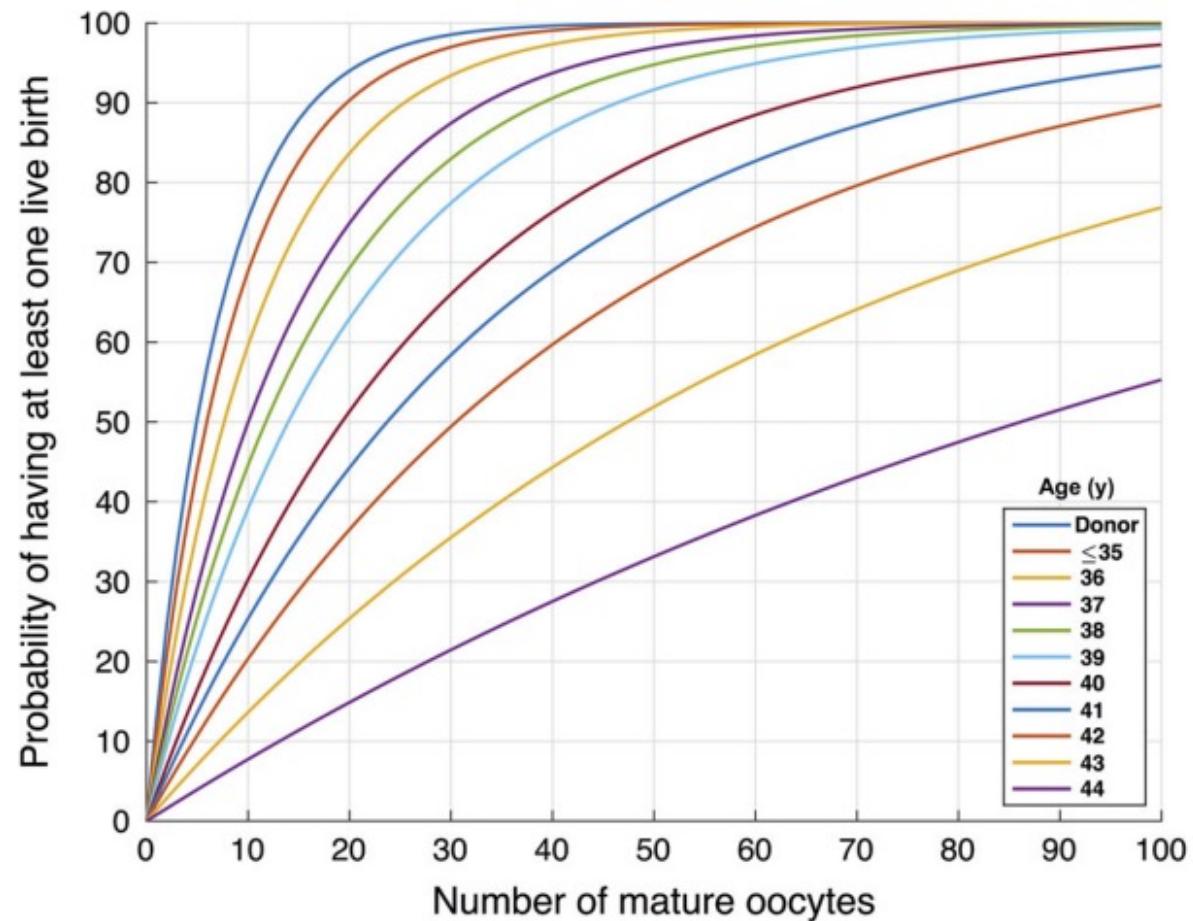
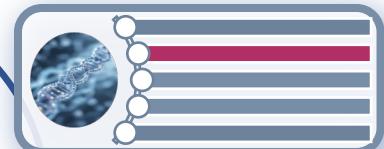


Figure 1 Live birth predictions by age and number of mature oocytes retrieved. Each curve shows the percent likelihood that a patient of a given age will have at least one live birth according to Equation 2, based on the number of mature oocytes retrieved and frozen.

Goldman et al. Hum Reprod 2017

iEDAD!



Obstetric babies born from VIT

Ana Cobo, Ph.D., Vicente Serra, M.D., Nicolás Garrido, Ph.D., Iné and José Remohí, M.D.

Fertility and Sterility® Vol. 102, No. 4, October 2014

Fertility and Sterility® Vol. 98, No. 3, September 2012

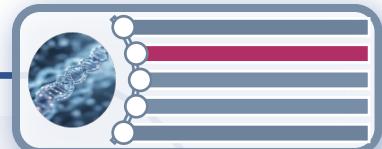
IN VITRO FERTILIZATION

Comparison of concomitant outcome achieved with fresh and cryopreserved donor oocytes vitrified by the Cryotop method

Ana Cobo, Ph.D.,^a Masashige Kuwayama, Ph.D.,^b Sonia Pérez, Ph.D.,^a Amparo Ruiz, M.D.,^a Antonio Pellicer, M.D.,^a and José Remohí, M.D.^a

Oocyte vitrification does not increase the risk of embryonic aneuploidy or diminish the implantation potential of blastocysts created after intracytoplasmic sperm injection: a novel, paired randomized controlled trial using DNA fingerprinting

Eric J. Forman, M.D.,^{a,b} Xinying Li, Ph.D.,^a Kathleen M. Ferry, B.S.,^a Katherine Scott, M.S.,^a Nathan R. Treff, Ph.D.,^{a,b} and Richard T. Scott Jr., M.D.^{a,b}



Is oocyte cryopreservation effective and safe for FP?



Oocyte cryopreservation is an **established option** for fertility **STRONG**
preservation



Women with a partner may cryopreserve unfertilized oocytes or to split the oocytes to attempt both embryo and oocyte cryopreservation.

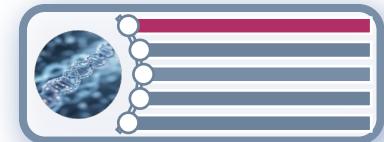
GPP

Women should be informed of **accurate, centre-specific expertise** and live birth rates. They should also be informed that success rates after cryopreservation of oocytes at the time of a cancer diagnosis may be lower than in women without cancer.

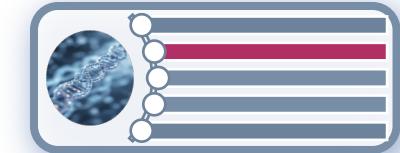
GPP



¿Qué haríamos con esos ovocitos
vitrificados a futuro?



Oocyte vitrification for fertility preservation for both medical and nonmedical reasons



Fertility and Sterility® Vol. 115, No. 5, May 2021 0015-0282/\$36.00

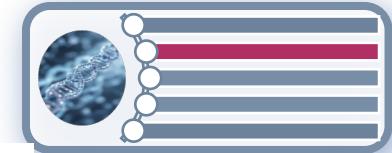
Ana Cobo, Ph.D.,^a Juan Antonio García-Velasco, M.D.,^b José Remohí, M.D.,^a and Antonio Pellicer, M.D.^a



- Opción más viable en **postmenárgicas**
- Realizada en niñas **mosaico** a partir de menarquia
- Requiere estimulación ovárica controlada (FSH, LH)
- Requiere niveles de FSH/LH normales y de AMH aceptables
- Número de ovocitos objetivo > 10
- Preocupaciones: aneuploidía ovocitaria; éxito desconocido en esta cohorte. **PGT**.



Criopreservación de ovocitos



Journal of Assisted Reproduction and Genetics (2022) 39:543–549
<https://doi.org/10.1007/s10815-022-02420-4>

FERTILITY PRESERVATION



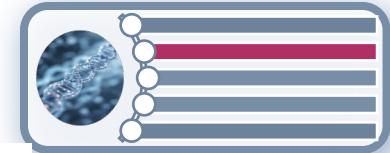
First live birth after fertility preservation using vitrification of oocytes in a woman with mosaic Turner syndrome

L. Strypstein¹ · E. Van Moer¹ · J. Nekkebroeck¹ · I. Segers¹ · H. Tournaye^{1,2} · I. Demeestere^{3,4} · M.-M. Dolmans^{5,6} · W. Verpoest^{1,7} · M. De Vos^{1,2,8}

En los estudios de cohortes y revisiones multicéntricas, aunque varias mujeres con síndrome de Turner han vitrificado ovocitos como estrategia de preservación de la fertilidad, **ninguna otra paciente ha retornado para utilizar sus ovocitos vitrificados** y, por lo tanto, no se han reportado más embarazos en este contexto.



Criopreservación de ovocitos



Journal of Assisted Reproduction and Genetics (2022) 39:543–549
<https://doi.org/10.1007/s10815-022-02420-4>

FERTILITY PRESERVATION

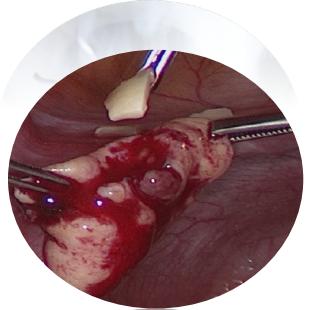


First live birth after fertility preservation using vitrification of oocytes in a woman with mosaic Turner syndrome

L. Strypstein¹ · E. Van Moer¹ · J. Nekkebroeck¹ · I. Segers¹ · H. Tournaye^{1,2} · I. Demeestere^{3,4} · M.-M. Dolmans^{5,6} · W. Verpoest^{1,7} · M. De Vos^{1,2,8}

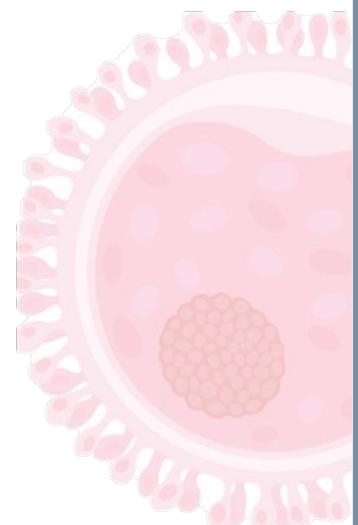
La evidencia actual indica que la vitrificación de ovocitos es factible en mujeres con Turner, especialmente en aquellas con mosaicismo y reserva ovárica suficiente, pero el uso clínico de estos ovocitos para lograr embarazo sigue siendo excepcional y extremadamente raro.





VITRIFICACIÓN OVOCITOS

CRYOPRESERVACIÓN TEJIDO
OVÁRICO

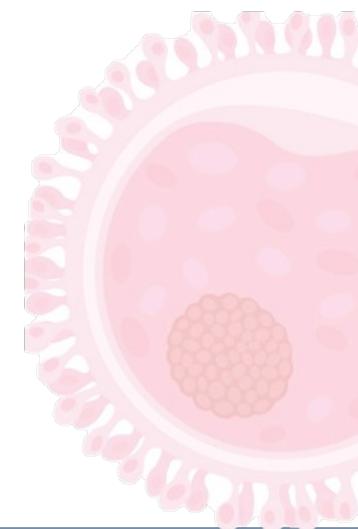




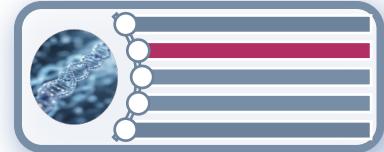
VITRIFICACIÓN OVOCITOS



CRIOPRESERVACIÓN TEJIDO
OVÁRICO



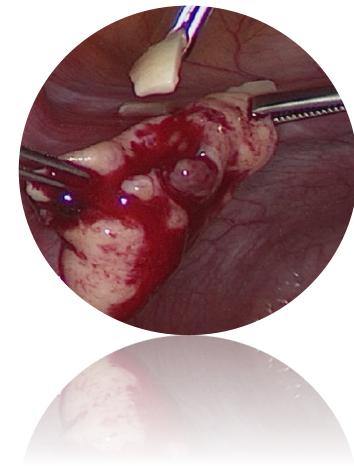
CRIOPRESERVACIÓN TEJIDO OVÁRICO



Única alternativa en prepúberes o pacientes que precisan inicio inmediato de tratamiento

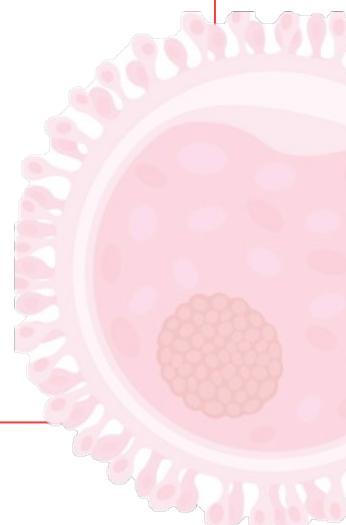
PROS:

- Técnica sencilla, baja curva de aprendizaje
- Beneficios: fertilidad y función endocrina
- Posibilidad gestación espontánea /varias gestaciones
- Se puede combinar con vitrificación de ovocitos

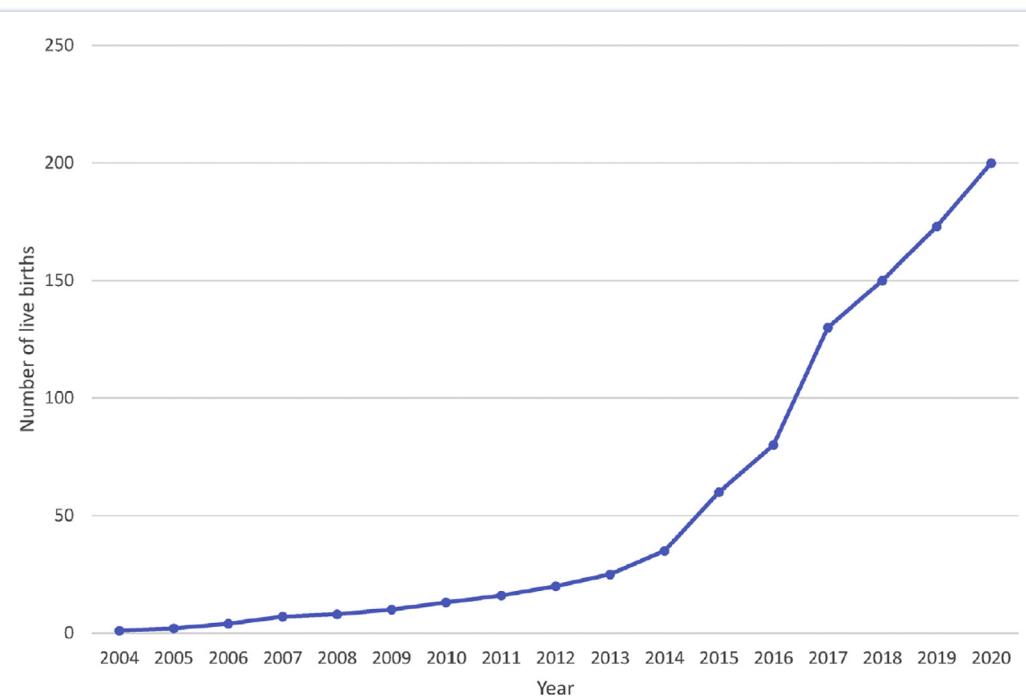
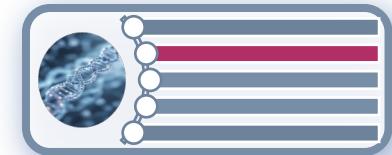


CONTRAS:

- Técnica considerada EXPERIMENTAL ESHRE. Desde 2020 ASRM no experim.
- Necesidad banco de tejidos
- Medios para criopreservar (congelación lenta mayoría)
- Riesgo de reimplante

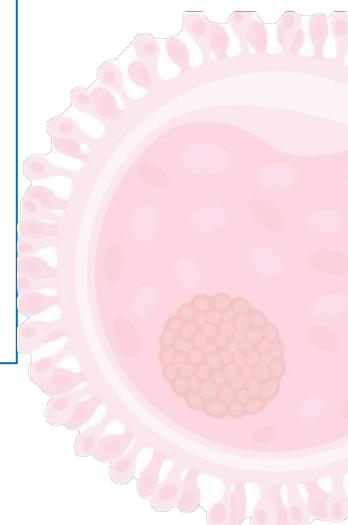


CRIOPRESERVACIÓN TEJIDO OVÁRICO

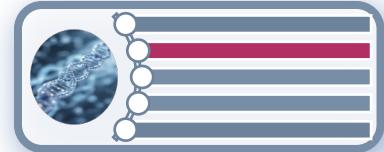


- Tras reimplante 95% recupera regla
- Duración media de implante 2-5 años (se podría realizar más de un implante en función de cantidad de tejido criopreservado)
- Criterios de selección: edad < 35 años, supervivencia razonable a > 5 años, riesgo de FOP 50%, no administración de agentes gonadotóxicos previos
- Tasa de gestación 29-45%, con tasa de RN vivo 23-66% según series
- 140 RNV tras reimplantación (2020)

¡Más del 50% espontáneas!



CRIOPRESERVACIÓN TEJIDO OVÁRICO

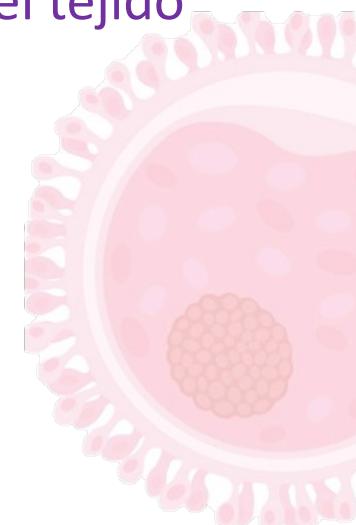
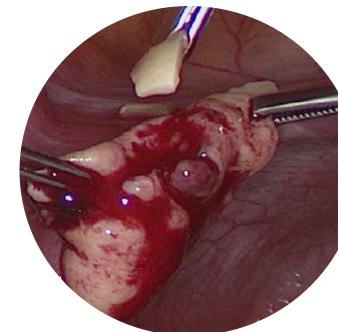


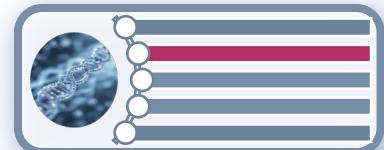
LIMITACIONES

- Reintroducción de células malignas con el reimplante. Importante el estudio del tejido previo al reimplante.
- Destrucción de la cohorte folicular por el efecto hipoxia- reperfusión.



- Aislar folículos in vitro + inserción en OVARIO ARTIFICIAL
- Mejorar la supervivencia del tejido tras el reimplante





Should ovarian tissue cryopreservation (OTC) be used for FP?

Offer OTC in patients undergoing moderate/high risk gonadotoxic treatment where oocyte/embryo cryopreservation is not feasible, or at patient preference

The efficiency of OTC procedure is questionable above 36 years of age.

The GDG considers that OTC is an **innovative** method in post pubertal women.

OTC/Ovarian tissue transplantation (OTT) can be considered in patients with POI-associated genetic and chromosomal disorders but requires genetic counselling and should be performed within a research protocol.



STRONG



WEAK

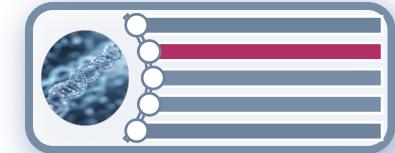


GPP

**RESEARCH
ONLY**

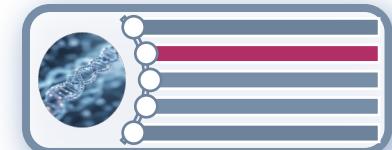


CRIOPRESERVACIÓN TEJIDO OVÁRICO



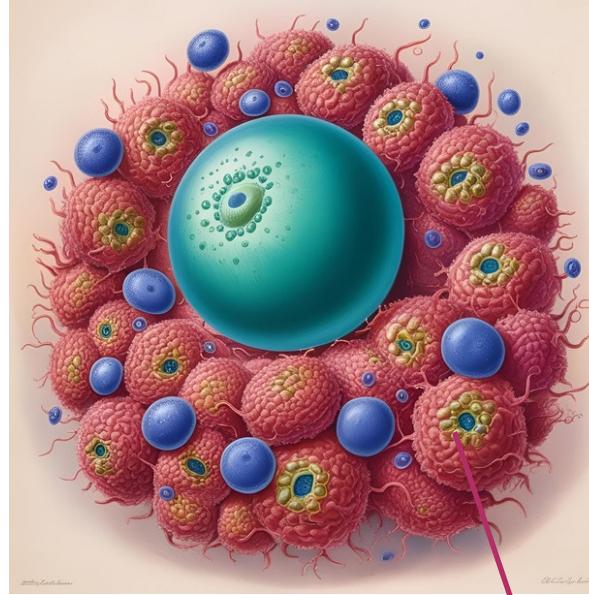
- Técnica experimental.
- Única opción potencial para niñas prepúberes con reserva baja/temprana
- Recomendado extirpar el ovario completo debido a reserva ya baja
- Autotrasplante ortotópico o heterotópico
- Éxito desconocido en pacientes con Turner: se requieren más datos
- Folículos encontrados: más frecuente en mosaicos (86%)
- Puede reimplantarse en la edad adulta para restaurar función hormonal y fertilidad.



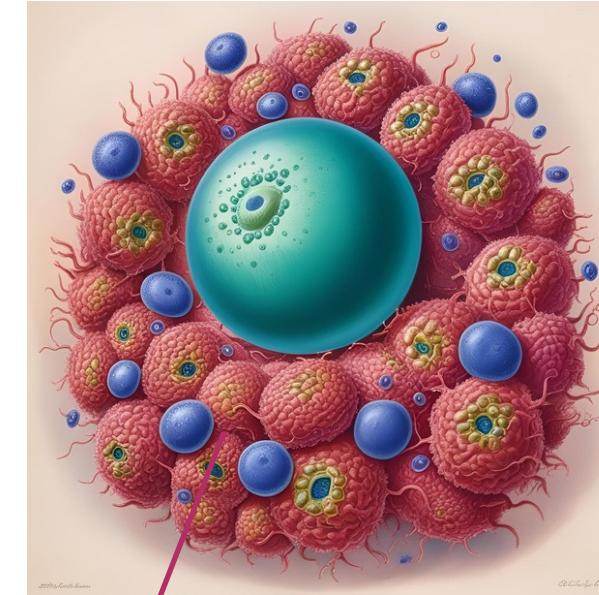


Gene expression analysis of ovarian follicles and stromal cells in girls with Turner syndrome

Ron Peek ^{1,*}, Sanne van der Coelen¹, and Marie-Madeleine Dolmans  ^{2,3}



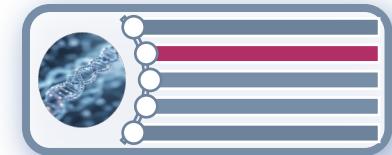
PREVIO A INJERTO
OVOCITOS 97% 46 XX; 3 % 45 XO
CÉLULAS DE LA GRANULOSA 85 % 45 X0



POSTERIOR AL INJERTO
CÉLULAS DE LA GRANULOSA 48,8% 45 XO



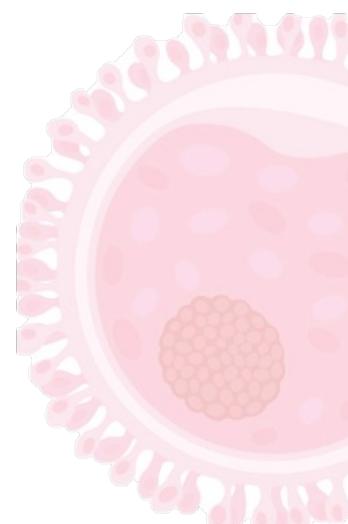
Puberty progression in girls with Turner syndrome after ovarian tissue cryopreservation



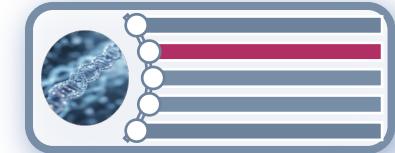
Fertil Steril® Vol. 123, No. 4, April 2025

Sanne van der Coelen, M.D.,^a Sapthami Nadesapillai, M.D.,^a Ronald Peek, Ph.D.,^a Didi Braat, M.D., Ph.D.,^a Gianni Bocca, M.D., Ph.D.,^b Martijn Finken, M.D., Ph.D.,^c Sabine Hannema, M.D., Ph.D.,^d Sandra de Kort, M.D., Ph.D.,^e Theo Sas, M.D., Ph.D.,^f Saartje Straetemans, M.D., Ph.D.,^g Vera van Tellingen, M.D.,^h Annemarie Verrijn Stuart, M.D., Ph.D.,ⁱ Kathrin Fleischer, M.D., Ph.D.,^j and Janielle van der Velden, M.D., Ph.D.^k

- **Impacto a Corto Plazo:** Los hallazgos sugieren que la ovariectomía unilateral para OTC **no tiene un impacto significativo en el desarrollo puberal a corto plazo** en la mayoría de las niñas con ST.
- **Relevancia Clínica:** El estudio demuestra que la mayoría de las niñas experimentaron un desarrollo puberal espontáneo continuo después de la OTC, lo cual es importante para el bienestar psicosocial.



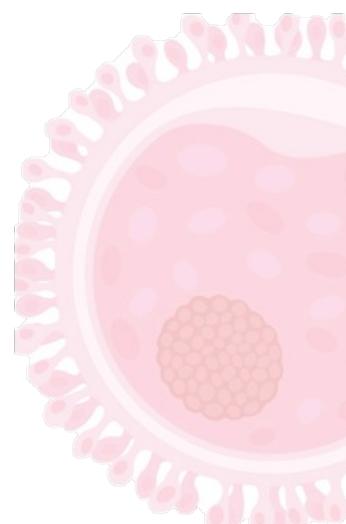
Puberty progression in girls with Turner syndrome after ovarian tissue cryopreservation



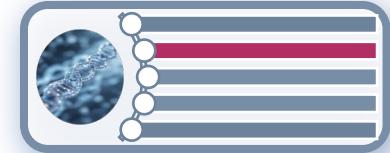
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- Hasta la fecha, solo se han reportado a nivel mundial tres casos de trasplante de tejido ovárico criopreservado en mujeres con ST, con solo uno resultando en un embarazo seguido de un aborto espontáneo. Por lo tanto, se aconseja que las niñas con ST que consideren la OTC sean asesoradas e incluidas en un entorno de investigación hasta que se establezca su eficacia.
- **Otras Opciones de Fertilidad:** La vitrificación de ovocitos es la técnica de preservación de la fertilidad preferida, más utilizada y más exitosa hasta la fecha, y debe discutirse con las niñas con ST cuando su madurez psicológica lo permita.



Fertility preservation in girls with Turner syndrome: to do or not to do?

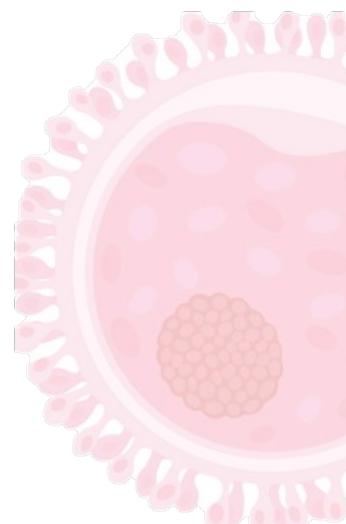


Jacques Donnez M.D., Ph. D. y Marie-Madeleine Dolmans M.D., Ph. D.

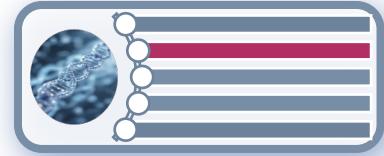
Fertility and Sterility, 2023-11-01, Volumen 120, Número 5, Páginas 993-994, Copyright © 2023 American Society for Reproductive Medicine

Although some experts still consider the technique experimental in patients with TS, we have always believed that they could be candidates for OTC as long as there is a reasonable chance of finding primordial follicles in their ovarian tissue. In our opinion, OTC should be performed before puberty (at least before age 12) to avoid depletion of the ovarian reserve as best we can.

- Mosaicos 45X0/46XX.
- Menores de 12 años
- **Antes de la pubertad, si hay posibilidad razonable de encontrar folículos.**
- Niveles de FSH < 10 UI/ml y AMH > 0.1 ng/ml



Fertility preservation in girls with Turner syndrome: to do or not to do?

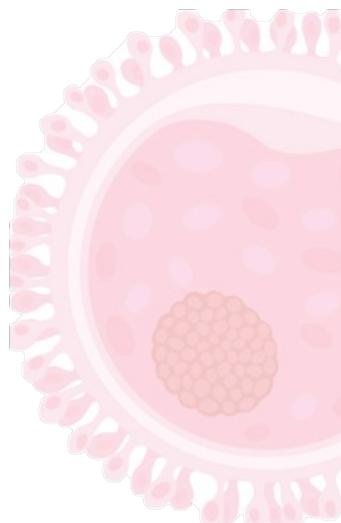


Jacques Donnez M.D., Ph. D. y Marie-Madeleine Dolmans M.D., Ph. D.

Fertility and Sterility, 2023-11-01, Volumen 120, Número 5, Páginas 993-994, Copyright © 2023 American Society for Reproductive Medicine

Young patients should be psychologically prepared for OTC, as they need to make a decisión they may not be emotionally or mentally ready for.

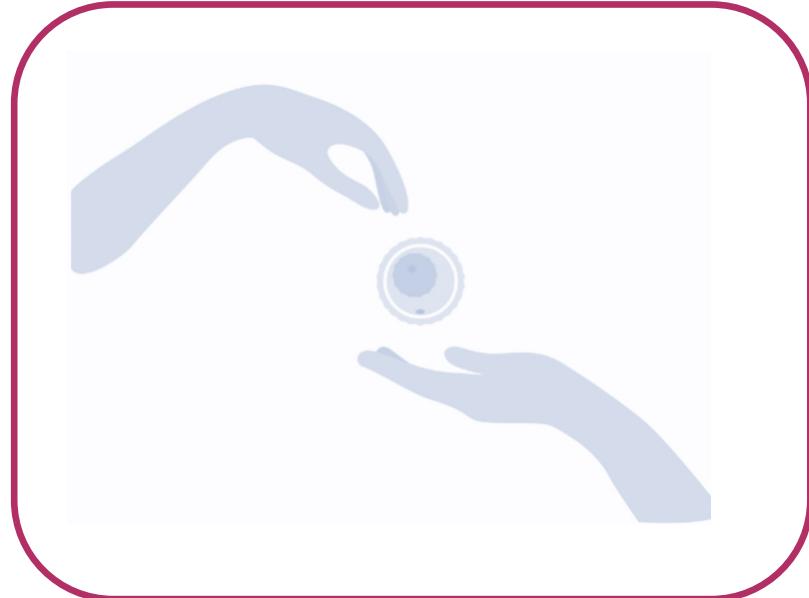
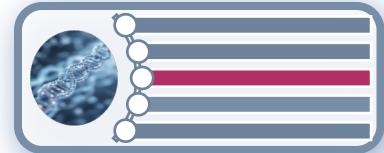
- Mosaicos 45X0/46XX.
- Menores de 12 años
- **Antes de la pubertad, si hay posibilidad razonable de encontrar folículos.**
- Niveles de FSH < 10 UI/ml y AMH > 0.1 ng/ml
- Asegurar una adecuada evaluación psicológica y CI firmado
- Realizar cribado cardiológico antes de la preservación de fertilidad



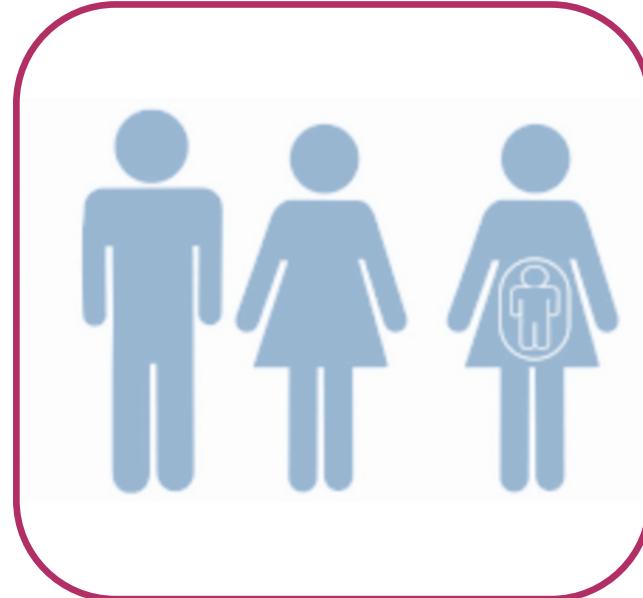
Opciones reproductivas en pacientes con Sdr Turner



Alternativas reproductivas



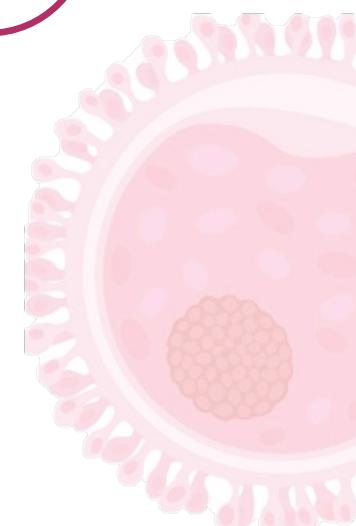
Donación ovocitos o embriones



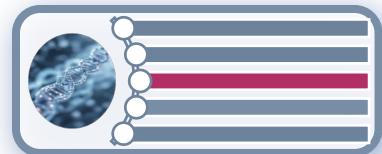
Subrogación uterina



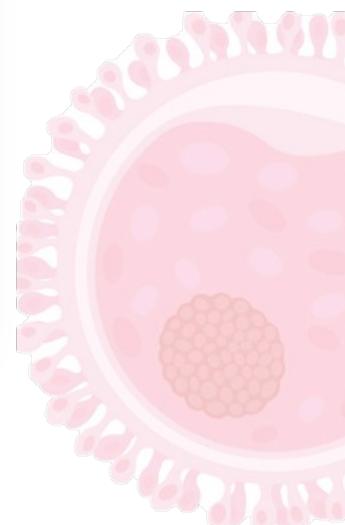
Adopción



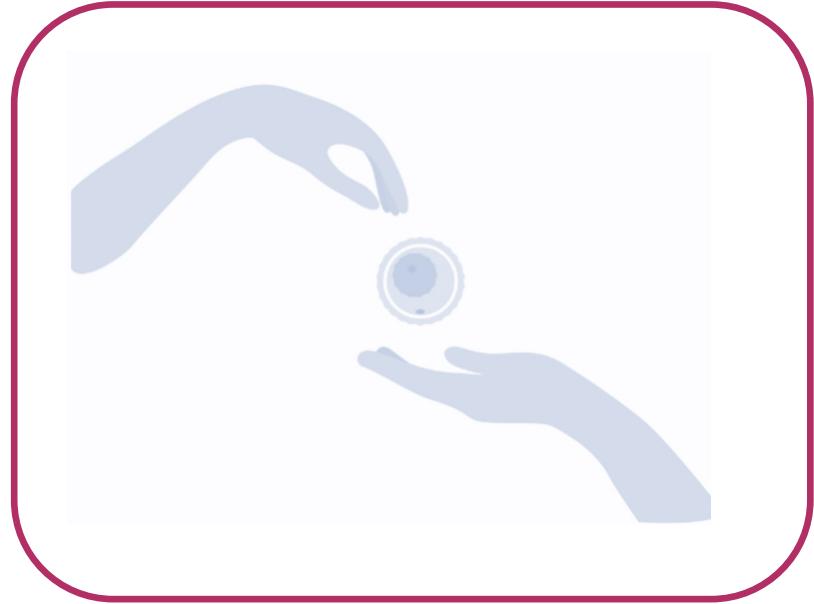
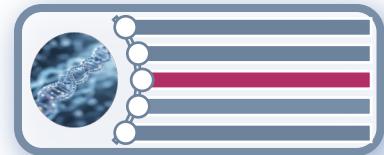
Alternativas reproductivas



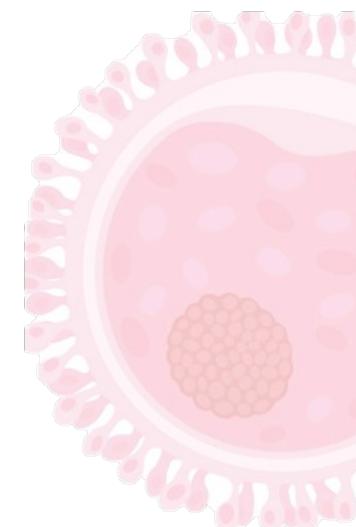
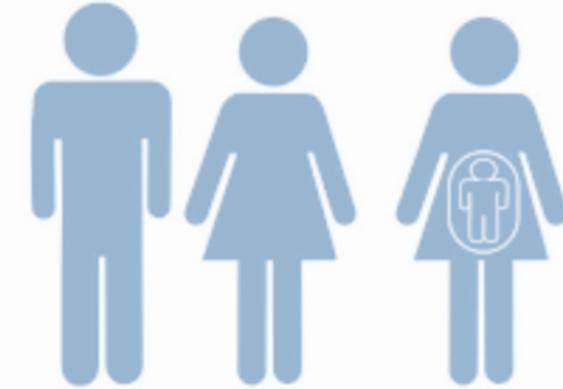
Característica	Donación de ovocitos	Adopción	Gestación subrogada
Vínculo genético	La madre receptora no tiene vínculo. El padre sí (si se usa su semen).	Ninguno de los padres tiene vínculo.	Los futuros padres pueden tenerlo (si usan sus propios óvulos/esperma). La gestante no.
Vínculo gestacional	La madre receptora gesta al bebé.	El niño ya ha nacido.	La gestante lleva el embarazo.
Marco legal en España	Legal.	Legal.	Prohibida.
Procedimiento	Reproducción asistida con transferencia de embrión.	Proceso legal y administrativo.	Acuerdo con gestante.
Control	Los padres receptores no eligen a la donante.	Los padres no eligen al menor.	Los futuros padres eligen a la gestante en el extranjero.



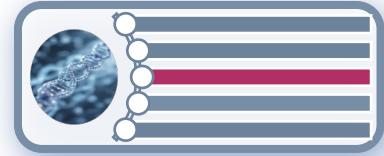
Alternativas reproductivas



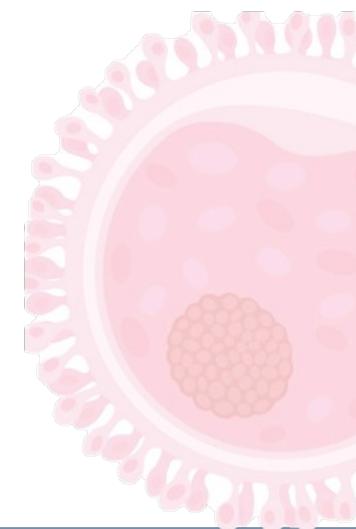
**Donación ovocitos o
embriones**



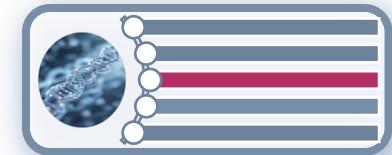
Donación de ovocitos / embriones



- Opción más eficaz para lograr embarazo en ST.
- Tasas de aborto algo mayores.
- Considerar salud uterina y riesgo cardiovascular.

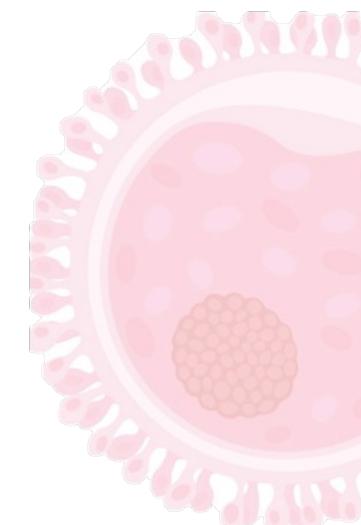


Donación de ovocitos

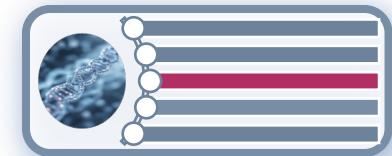


CRITERIOS SELECCIÓN DONANTES

- 18-35 años
- Máximo de 6 recién nacidos vivos
- Sin historia de enfermedad genética
- Buen estado psicofísico
- Serologías negativas
- Otras: cariotipo, screening de enfermedades monogénicas



Donación de ovocitos

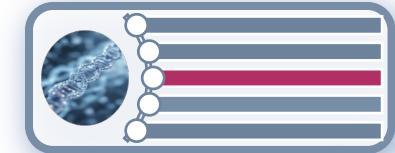


España fue el primer país con una Ley separada sobre técnicas de reproducción asistida

- Donación debe ser ANÓNIMA
- Se debe garantizar el máximo de similitud y compatibilidad fenotípica e inmunológica entre donante / receptora
- NO se paga por la donación (pero se compensa por las molestias que supone)



Donación de ovocitos

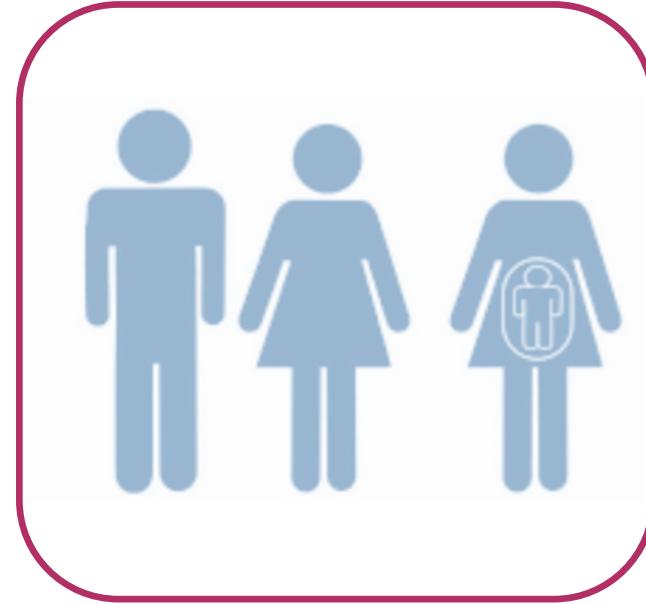
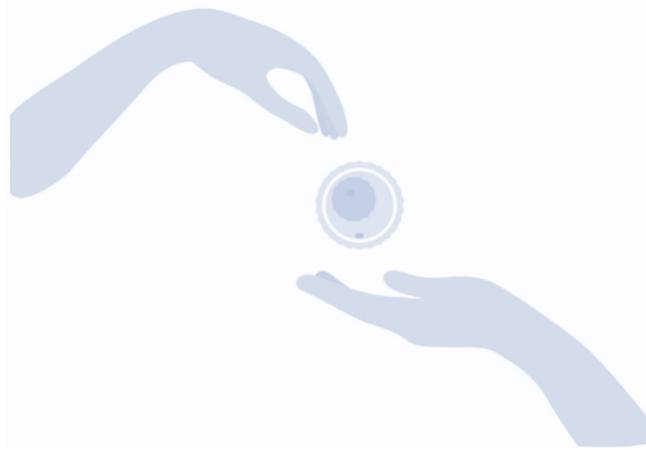
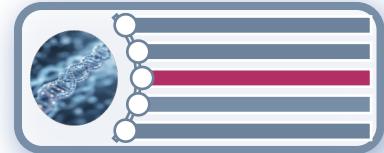


Compatibilidad

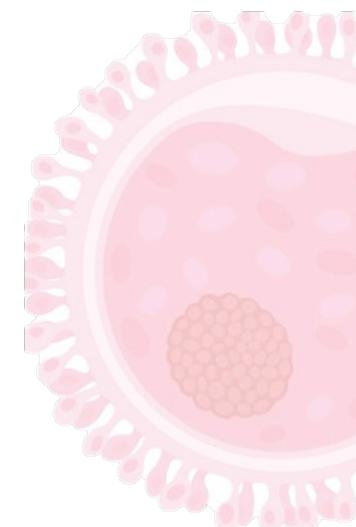
- Raza / grupo sanguíneo
- Raza / grupo sanguíneo / características físicas
- Raza / grupo sanguíneo / características físicas / características genéticas



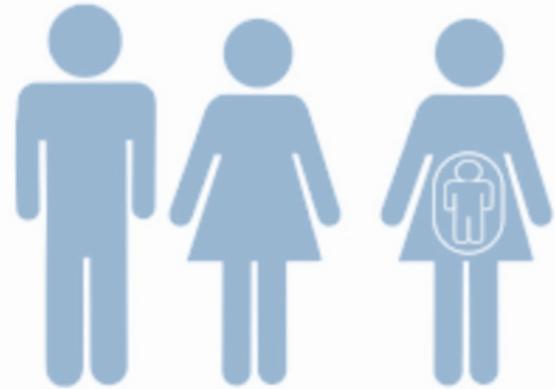
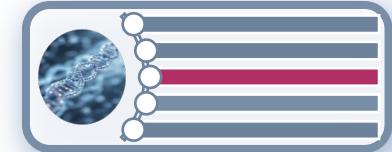
Alternativas reproductivas



Subrogación uterina



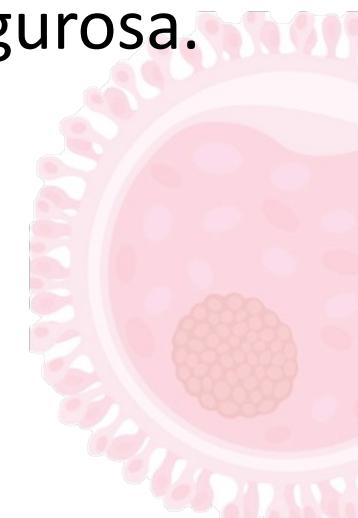
Gestación subrogada



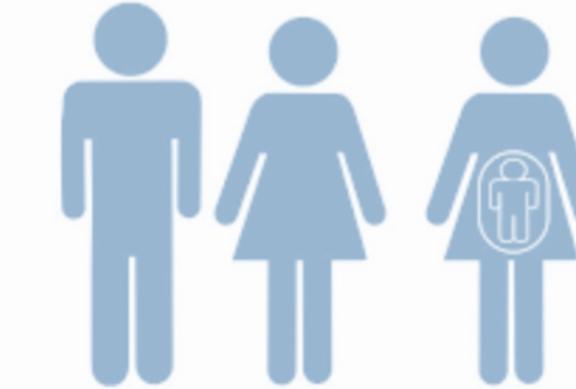
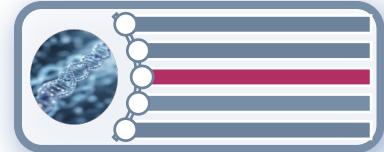
- Alternativa recomendada para mujeres con riesgo cardiovascular (contraindicación embarazo).
- Permite maternidad genética con ovocitos propios (si los tiene) o donados.
- Requiere evaluación médica y legal rigurosa.



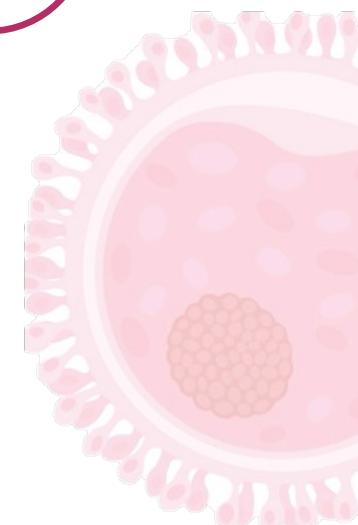
¡NO LEGAL EN ESPAÑA!



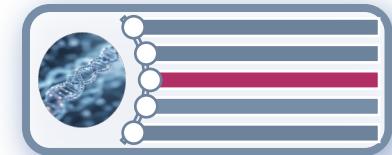
Alternativas reproductivas



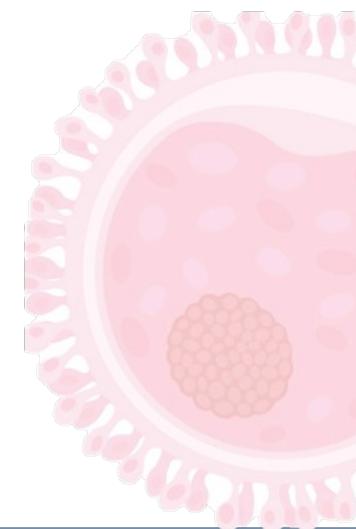
Adopción



Adopción como alternativa

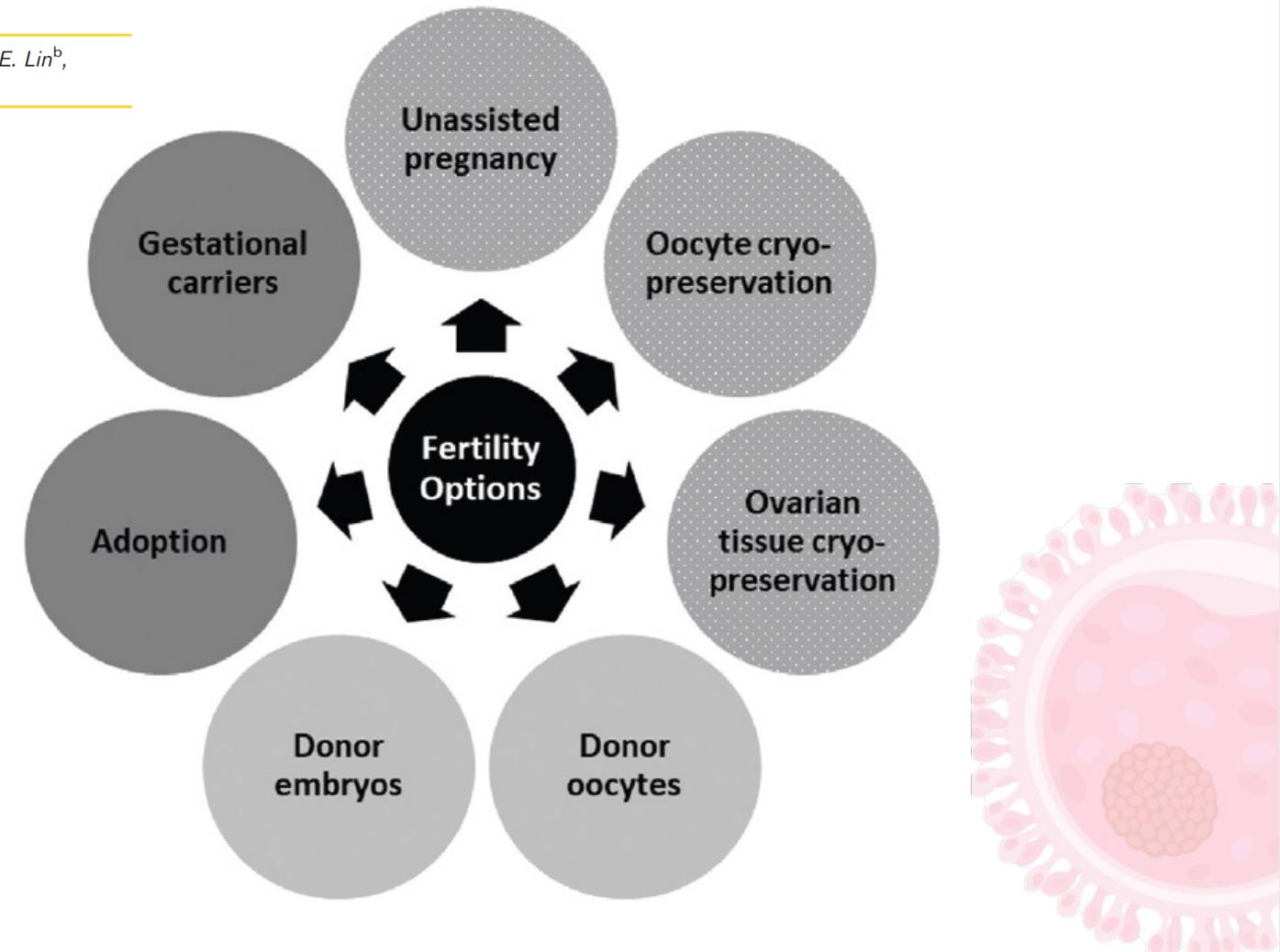


- Vía segura y viable para mujeres con ST.
- Posibilidad de adopción nacional o internacional.
- No hay restricciones específicas para ST.

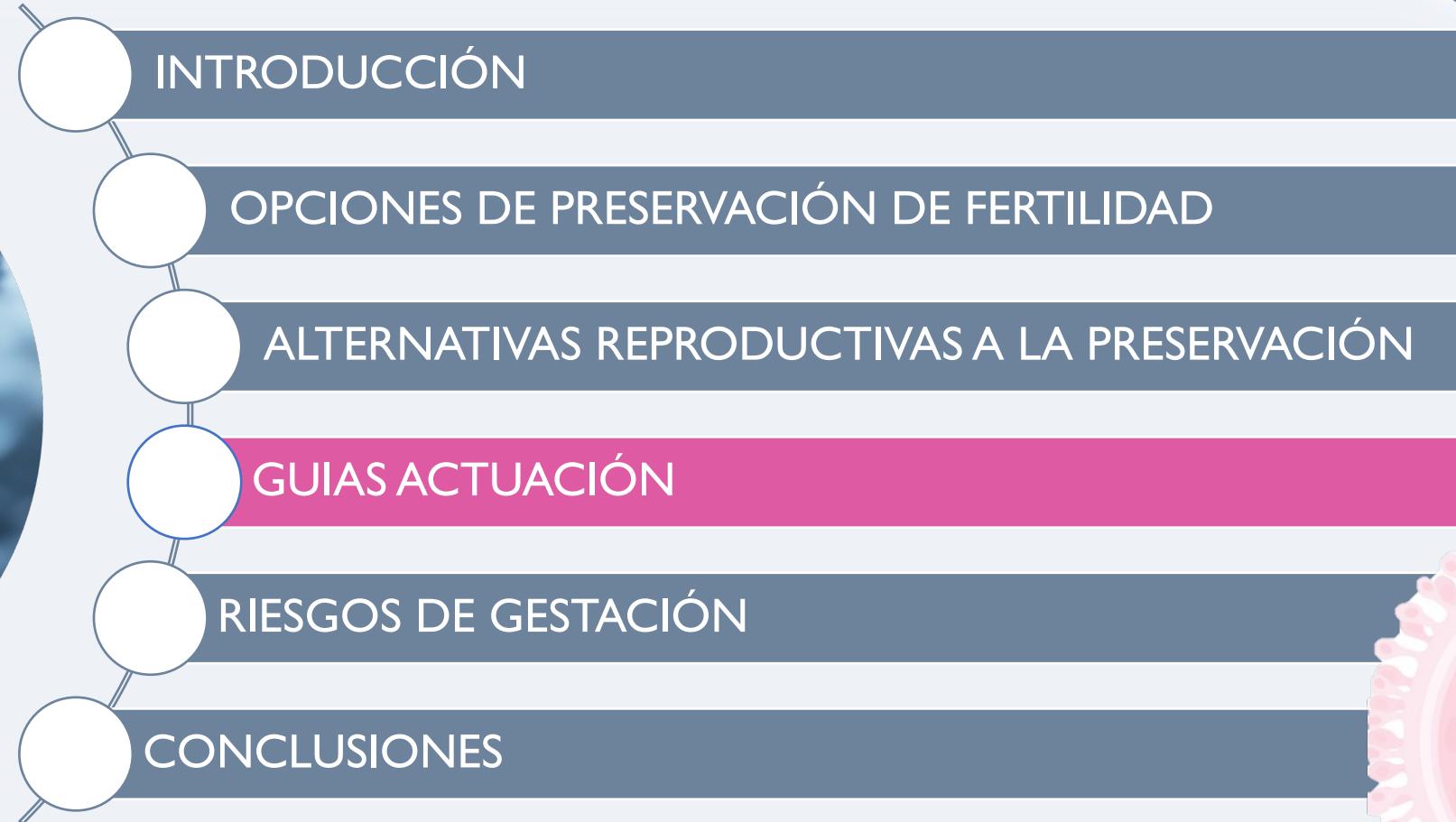


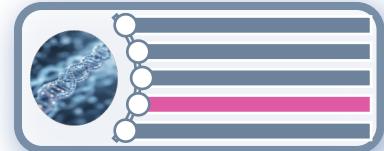
Turner syndrome: fertility counselling in childhood and through the reproductive lifespan

Kassie J. Bollig^a, Monica Mainigi^a, Suneeta Senapati^a, Angela E. Lin^b,
Lynne L. Levitsky^c and Vaneeta Bamba^d



Opciones reproductivas en pacientes con SdR Turner





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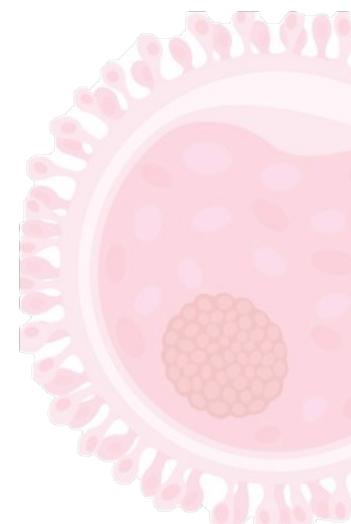
Clinical Practice Guideline



Clinical practice guidelines for the care of girls and women with Turner syndrome

Proceedings from the 2023 Aarhus International Turner Syndrome Meeting

Claus H. Gravholt,^{1,2,3,*} Niels H. Andersen,⁴ Sophie Christin-Maitre,⁵ Shanlee M. Davis,^{6,7} Anthonie Duijnhouwer,⁸ Aneta Gawlik,⁹ Andrea T. Maciel-Guerra,¹⁰ Iris Gutmark-Little,¹¹ Kathrin Fleischer,¹² David Hong,^{13,14} Karen O. Klein,¹⁵ Siddharth K. Prakash,¹⁶ Roopa Kanakatti Shankar,¹⁷ David E. Sandberg,^{18,19} Theo C.J. Sas,^{20,21} Anne Skakkebæk,^{2,3,22} Kirstine Stochholm,^{1,23} Janielle A. van der Velden²⁴ The International Turner Syndrome Consensus Group[†]; and Philippe F. Backeljauw^{11,*}



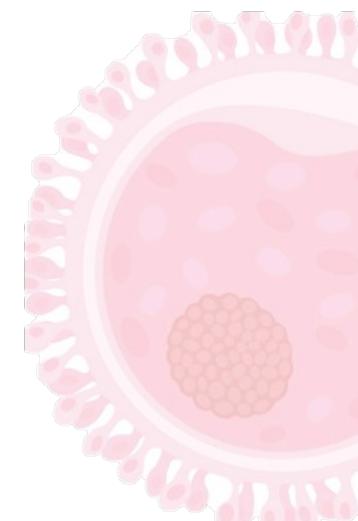
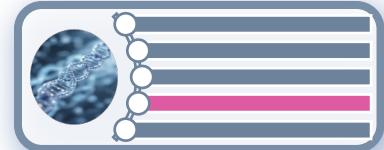
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6 Fertility assessment, monitoring, and counselling

- **R 6.1** We recommend developmentally appropriate disclosure of the potential for reduced fertility in individuals with TS. We recommend disclosing that the probability to conceive is primarily associated with the presence of a 46, XX cell line and spontaneous menarche, and that there is increased risk of maternal and fetal complications in pregnancy compared to the general population (⊕⊕⊕○).
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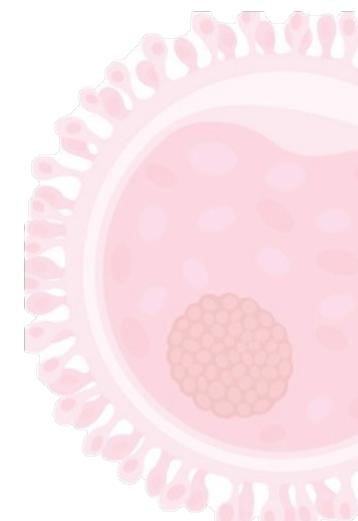
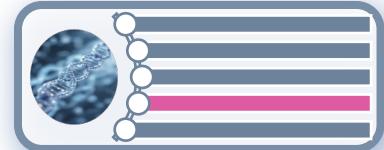
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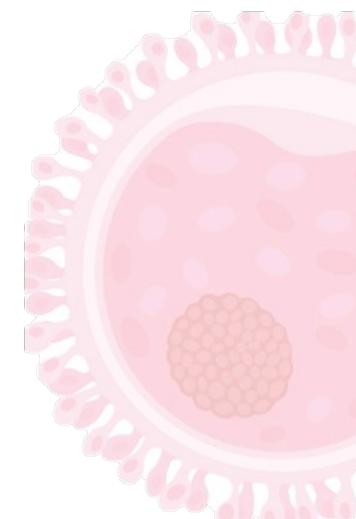
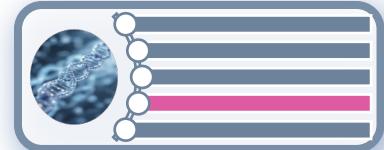
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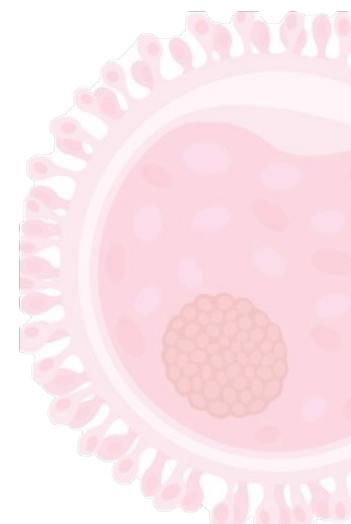
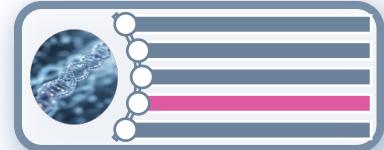
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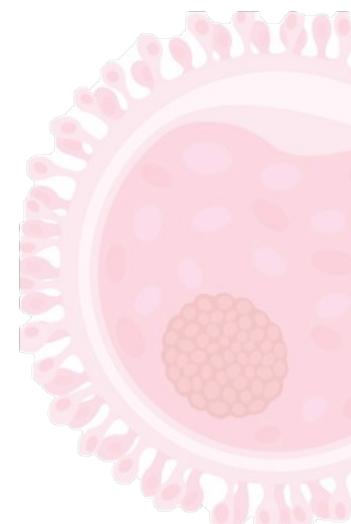
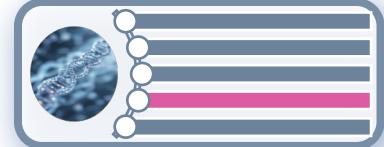
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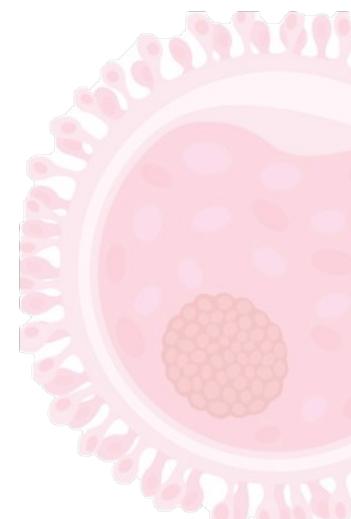
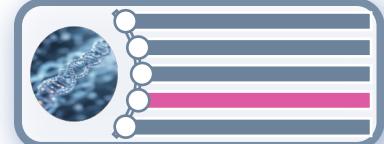
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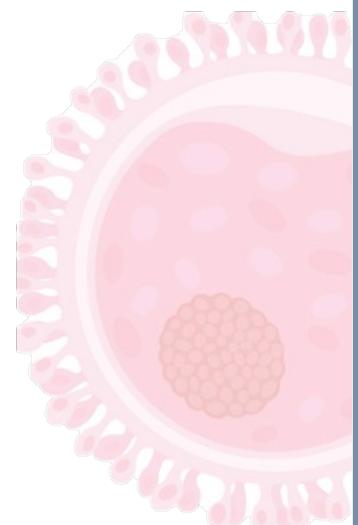
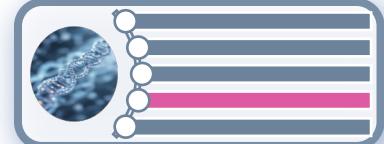
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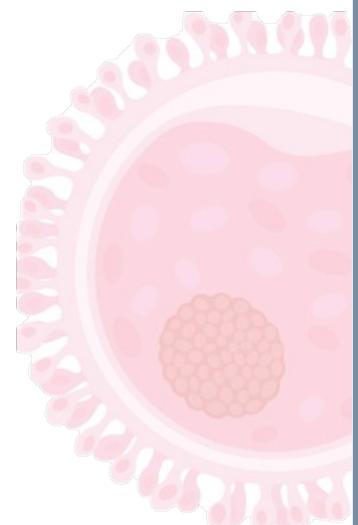
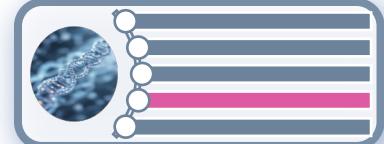
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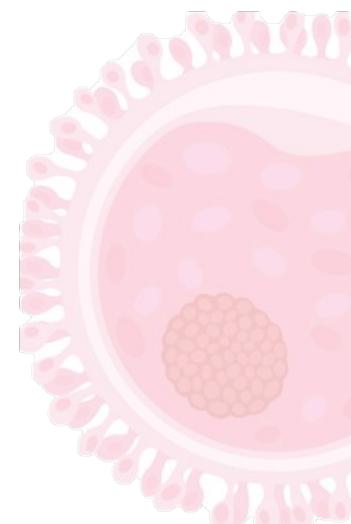
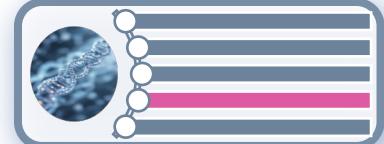
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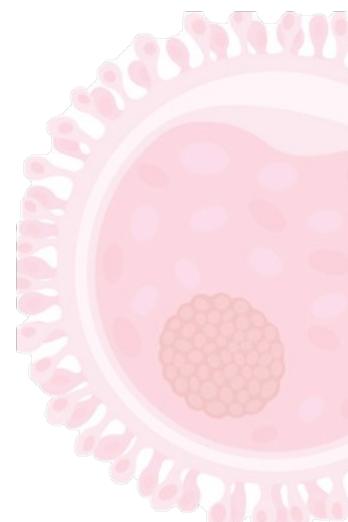
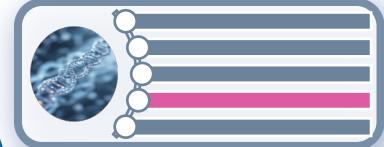
Clinical practice guidelines with Turner syndrome: Proceedings from the Meeting

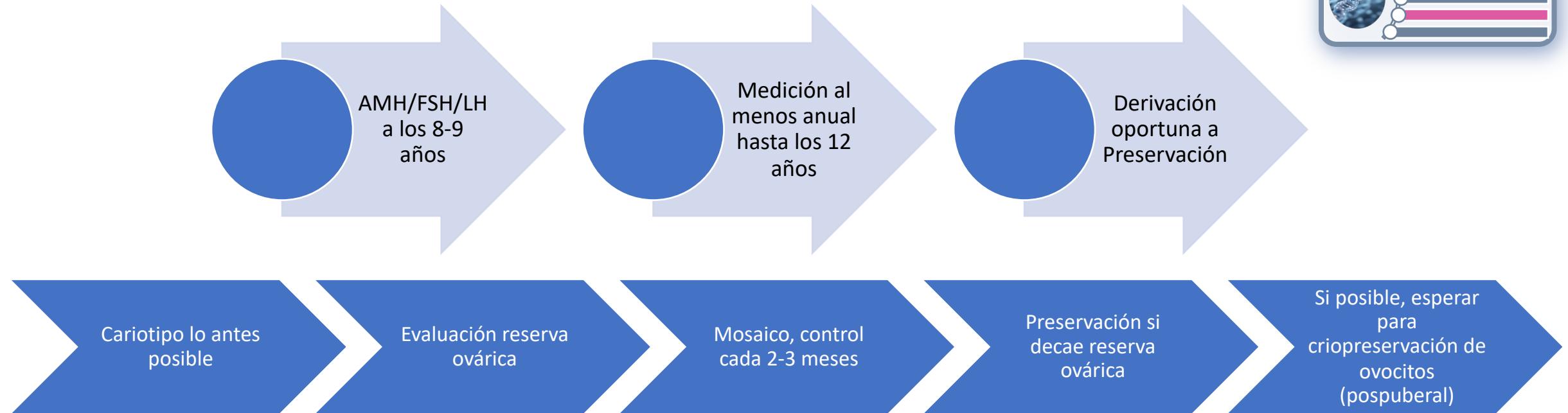
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1.2.2 Pre-implantation genetic testing

- R 1.13 We recommend that preimplantation testing can be offered to individuals with TS who want to use their own oocytes for pregnancies. TS individuals with mosaicism (45,X/46,XX), who become pregnant spontaneously, should be offered prenatal diagnostic testing (⊕⊕⊕○).

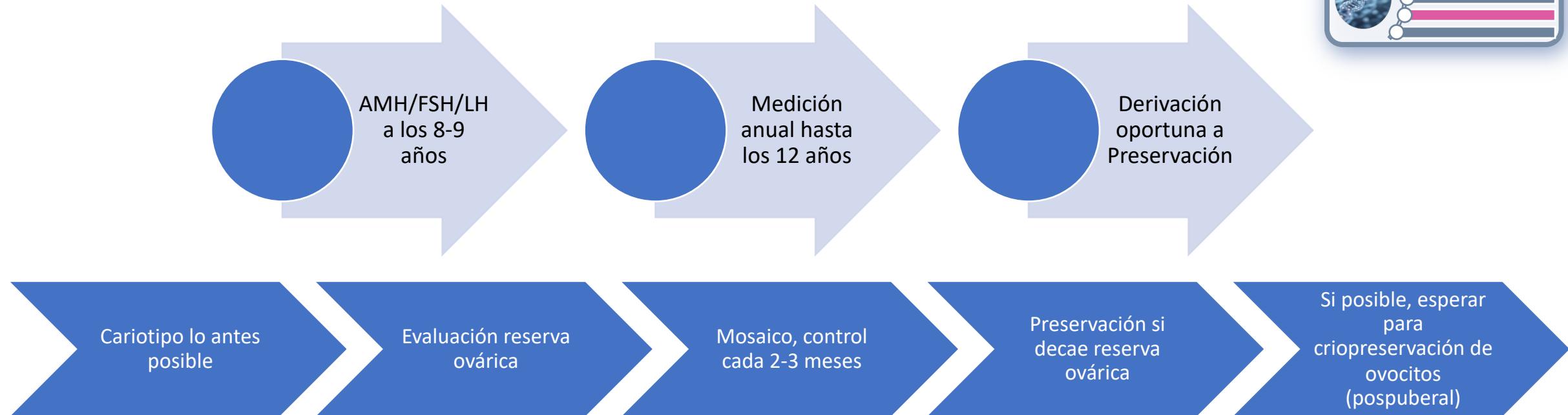
Pre-implantation genetic testing is currently offered to women with recurrent pregnancy loss or repetitive implantation failure after in vitro fertilization procedures although the clinical benefit is an ongoing topic of discussion.^{74,75} Pre-implantation genetic testing can be offered in case of a women with TS and the desire to have children.⁷⁶ However, a sufficient ovarian reserve to obtain sufficient embryos for testing is a prerequisite for applying pre-implantation genetic testing.





Para las pacientes no mosaico (donde la reserva ovárica puede agotarse en los primeros años de vida), la referencia y la discusión sobre la PF deben ocurrir tan pronto como se haga el diagnóstico, incluso en la infancia.

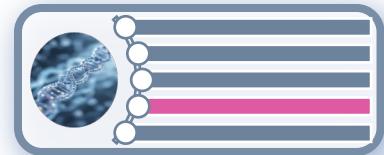




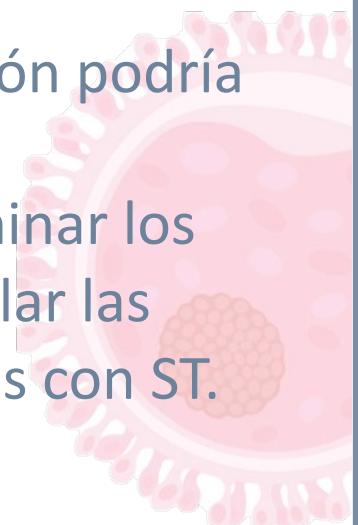
Niveles elevados de FSH + niveles indetectables de AMH + ausencia de folículos antrales visibles + visualización cintillas ováricas → fallo ovárico ya establecido. Preservación no es posible.



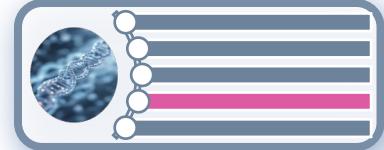
Criopreservación de ovocitos



- **Candidatas:** Se debe ofrecer la criopreservación de ovocitos a las **niñas postmenárquicas** (típicamente de 13 años o más) y lo suficientemente maduras psicológicamente para tolerar los procedimientos de estimulación ovárica.
- **Evaluación y Asesoramiento:** Se recomienda que todas las niñas postmenárquicas con ST sean evaluadas para la reserva ovárica y se les asesore sobre la criopreservación de ovocitos como una opción viable.
- **Riesgos Genéticos:** Debido al potencial de aneuploidía ovocitaria (una fracción podría carecer del cromosoma X), si se utilizan ovocitos autólogos, se deben ofrecer tecnologías como el **diagnóstico genético preimplantacional (DGP)** para examinar los embriones resultantes antes de la transferencia. El consentimiento debe detallar las posibles limitaciones, incluida la falta de tasas de éxito específicas en pacientes con ST.



Criopreservación de tejido ovárico



- **Candidatas:** Esta opción puede ofrecerse a niñas con ST que tienen reserva ovárica adecuada, pero que **no pueden esperar hasta tener la madurez suficiente para someterse a la criopreservación de ovocitos**. También se puede considerar para aquellas inmaduras sexual y/o psicosocialmente o incapaces de tolerar la estimulación ovárica.
- **Estado Experimental:** La criopreservación de tejido ovárico es una técnica que sigue considerándose **experimental** en adultos y para las niñas con ST. La probabilidad de éxito es desconocida.
- **Proceso:** Se recomienda extirpar tanto tejido como sea posible, generalmente un ovario entero, ya que la reserva ovárica en estas niñas ya es baja.
- **Consentimiento:** Si se lleva a cabo, debe ser bajo protocolos experimentales aprobados por la junta de revisión institucional (IRB). Se debe obtener el **asentimiento verbal** de los niños mayores de nueve años, además del consentimiento aprobado por los padres.



Otras opciones de gestación



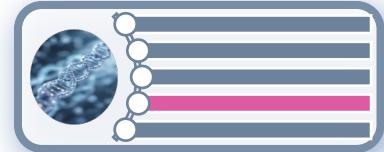
DONACIÓN DE OVOCITOS

Para la mayoría de las pacientes con ST, la opción preferida que ofrece la mayor probabilidad de éxito para tener hijos es el embarazo **mediante ovocitos de donante**.

- **Riesgos Adicionales:** A pesar del uso de ovocitos de donante, las tasas de aborto espontáneo son ligeramente más altas en las receptoras con ST en comparación con la población general de receptoras. También es más probable que el embarazo se complique por disfunción tiroidea, obesidad, diabetes y trastornos hipertensivos (incluida la preeclampsia, hasta el 40%).



Otras opciones de gestación



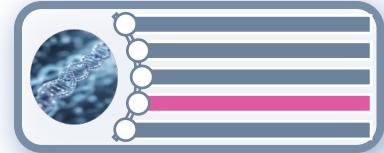
SUBROGACIÓN UTERINA

Debido a las conocidas complicaciones cardíacas y médicas potenciales (incluyendo aquellas que surgen incluso con una evaluación cardíaca normal), se recomienda que **todas las pacientes con ST sean asesoradas sobre la GS y la adopción como alternativas al embarazo.**

- La GS permite la posibilidad de paternidad biológica (usando ovocitos propios) o puede servir para llevar a término un embarazo resultante de ovocitos o embriones de donantes.

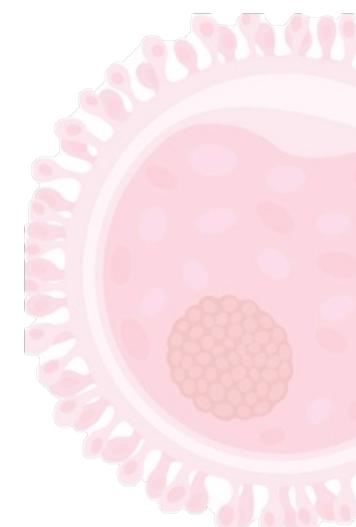


Otras opciones de gestación

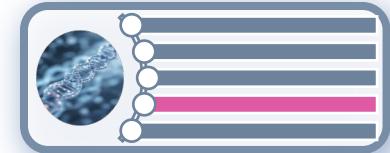


ADOPCIÓN

La adopción es una opción viable para las mujeres con ST que desean ser padres. No hay casos documentados de mujeres con ST que hayan sido restringidas para la adopción debido a requisitos de salud.

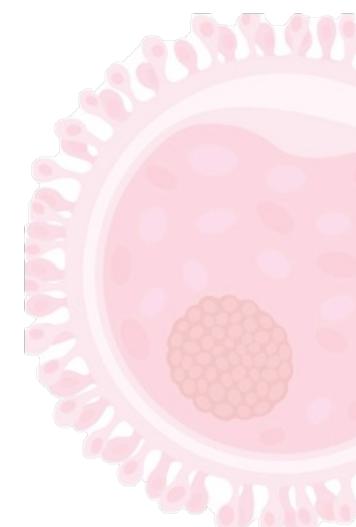


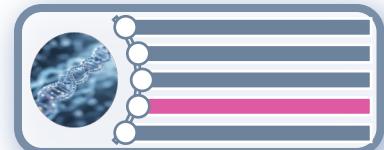
Otras opciones de gestación



GESTACIÓN ESPONTÁNEA

Aunque rara (2%-5%), si ocurre, las mujeres con ST deben considerar las pruebas prenatales o la fertilización in vitro (FIV) con pruebas genéticas preimplantacionales para evitar la aneuploidía, ya que las tasas de anomalías cromosómicas y abortos espontáneos son altas (hasta el 50%).





Journal Pre-proof

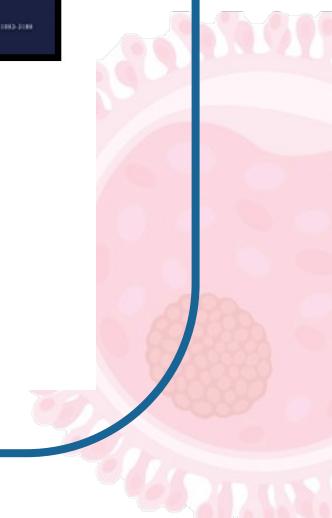
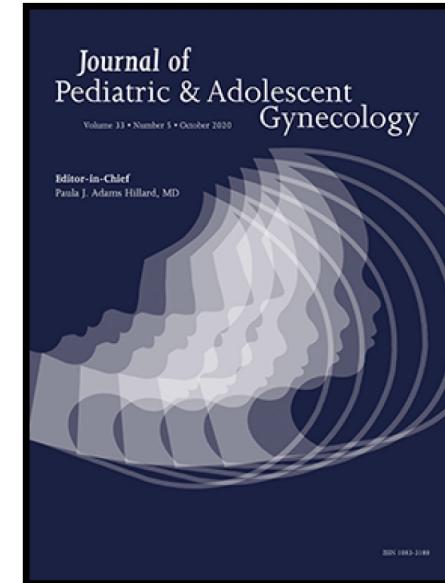
Perception and Attitude towards Fertility and Fertility Preservation Options in Parents of Children with Turner Syndrome: A Qualitative Survey Study

Victoria H. Bustamante Velez MD , Tazim Dowlut-McElroy MD , Roopa Kanakatti Shankar MBBS, MS

PII: S1083-3188(25)00316-X
DOI: <https://doi.org/10.1016/j.jpag.2025.07.008>
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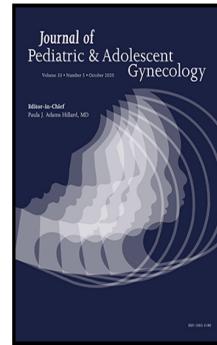
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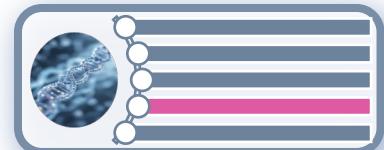
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TOMA DECISIONES COMPLEJAS POR FAMILIARES EN MENORES DE EDAD



COSTE

FINANCIACIÓN

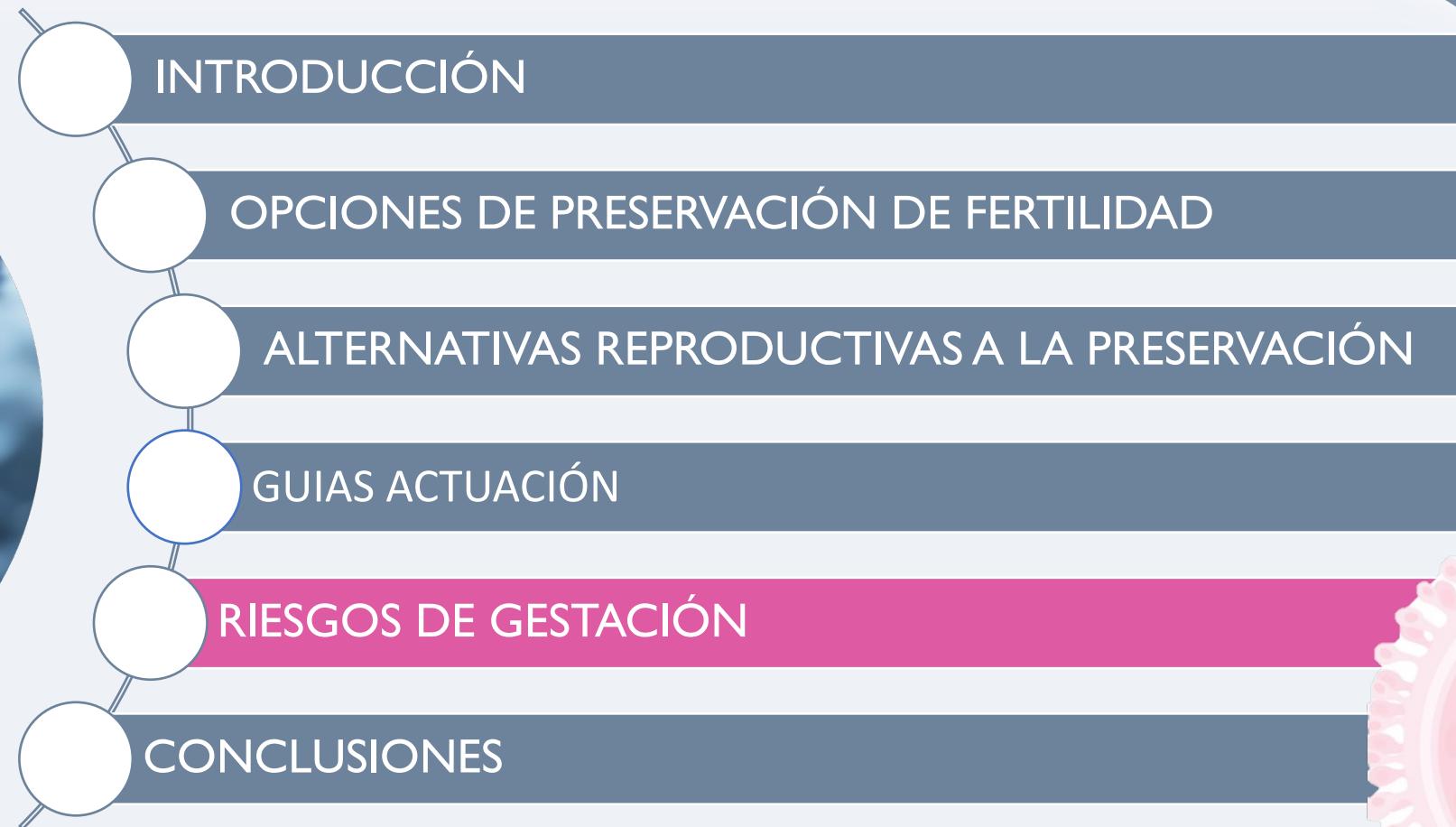
ÉTICA CRIOPRESERVAR OVOCITOS Q PUDIERAN ESTAR ALTERADOS

ESTADO VARIABLE DE MADUREZ PSICOLÓGICA Y SOCIAL

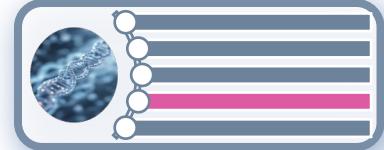
Los **factores socioeconómicos, culturales y éticos** tienen un impacto significativo en las decisiones parentales y personales relativas a las estrategias de preservación de la fertilidad (PF) para pacientes con Síndrome de Turner (ST), especialmente cuando se consideran opciones complejas como la criopreservación de gametos y embriones.



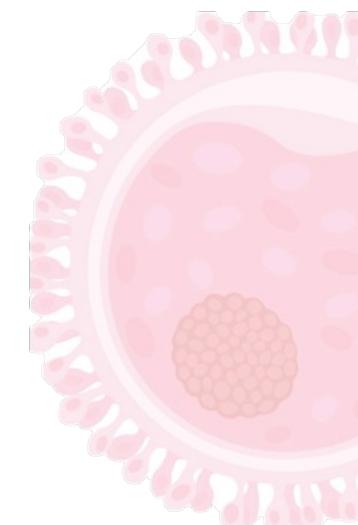
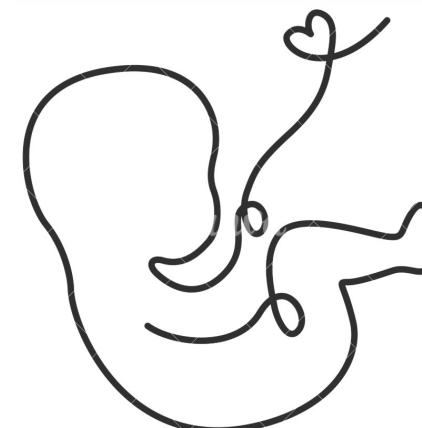
Opciones reproductivas en pacientes con SdR Turner



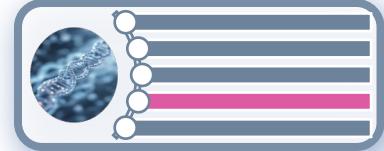
Riesgos y Complicaciones del Embarazo en ST



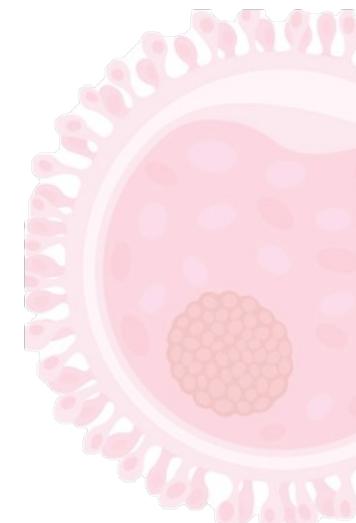
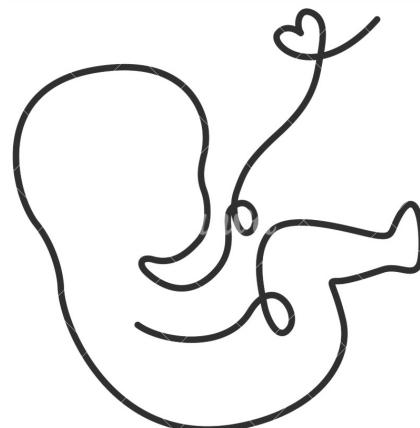
- Alta tasa de **preeclampsia**: hasta en el 21% de los embarazos, en comparación con el 5% en la población general.
- Aumento de **parto prematuro** (10-15%), restricción de crecimiento (**CIR**) (23%).
- Riesgo de **disección aórtica** (mortalidad 1-2%).
- Mayor tasa de **aborted** 48% en mujeres con ST.
- Mayor tasa de **cesáreas** (60-70%)



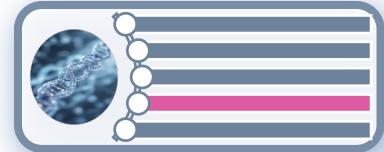
Riesgos y Complicaciones del Embarazo en ST



No se han observado diferencias[?] significativas en los resultados fetales entre embarazos espontáneos y por reproducción asistida, aunque los embarazos por ART tienden^a a finalizar por cesárea y a edades gestacionales ligeramente menores

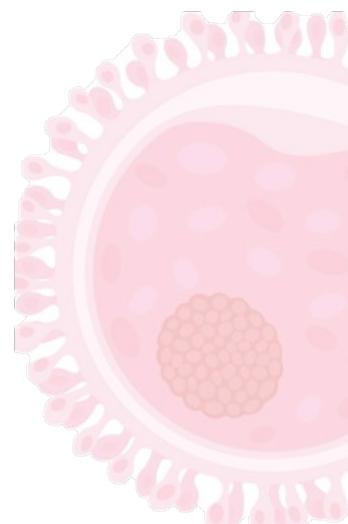
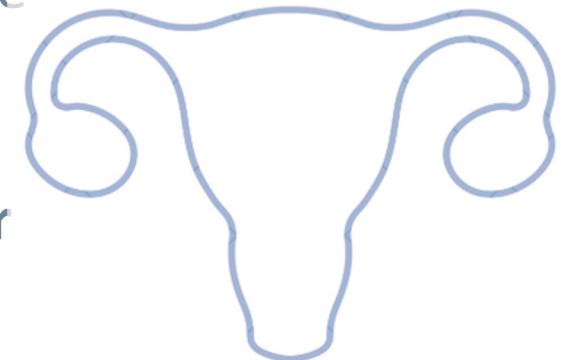


Riesgos y Complicaciones del Embarazo en ST



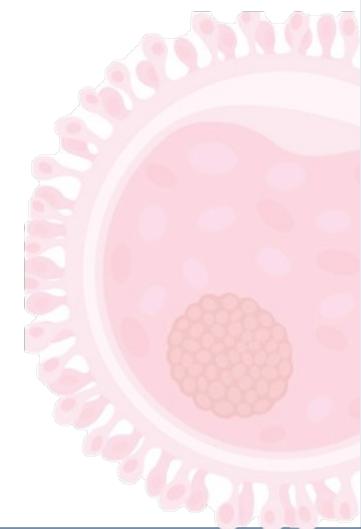
Existe una relación directa entre el **tamaño uterino** y los **resultados de los tratamientos reproductivos** en pacientes con síndrome de Turner.

Un útero de tamaño reducido o **hipoplásico** se asocia con **menor tasa de implantación embrionaria, mayor riesgo de aborto espontáneo y complicaciones obstétricas**, especialmente en el contexto de reproducción asistida y **ovodonación**. Un **tamaño uterino adecuado tras terapia hormonal** es esencial para obtener **buenos resultados en los tratamientos reproductivos en síndrome de Turner**; la hipoplasia uterina limita severamente el éxito y aumenta los riesgos obstétricos.



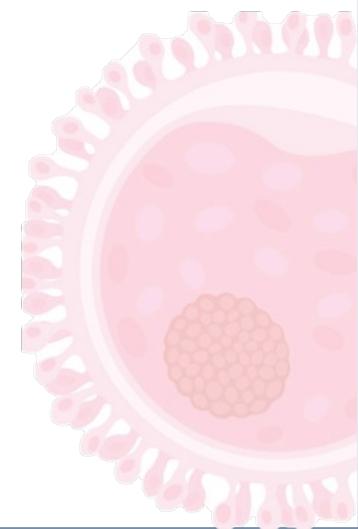
Evaluación preconcepcional recomendada

- Ecocardiograma y resonancia aórtica.
- Evaluar función renal, tiroidea, glucémica.
- Consejo genético y psicológico.
- Abordaje multidisciplinar.



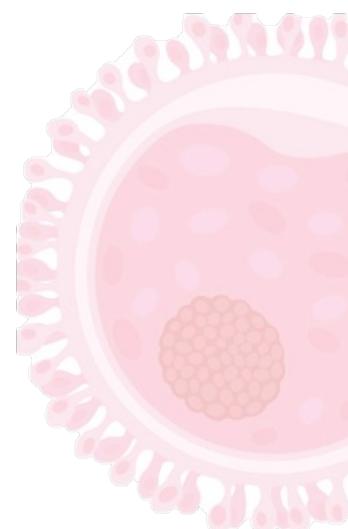
Evaluación preconcepcional recomendada

- Se debe transferir un solo embrión, ya que las gestaciones múltiples pueden plantear riesgos potencialmente mortales incluso en pacientes con estado cardíaco normal.
- Se debe ofrecer anticoncepción si existe alguna preocupación de embarazo espontáneo pero no deseado.



Evaluación preconcepcional recomendada

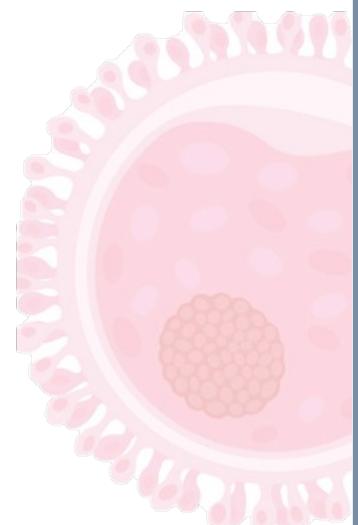
Abordar los riesgos individuales, discutir alternativas reproductivas (gestación subrogada, adopción) y asegurar que las pacientes comprendan la importancia del seguimiento cardiovascular.



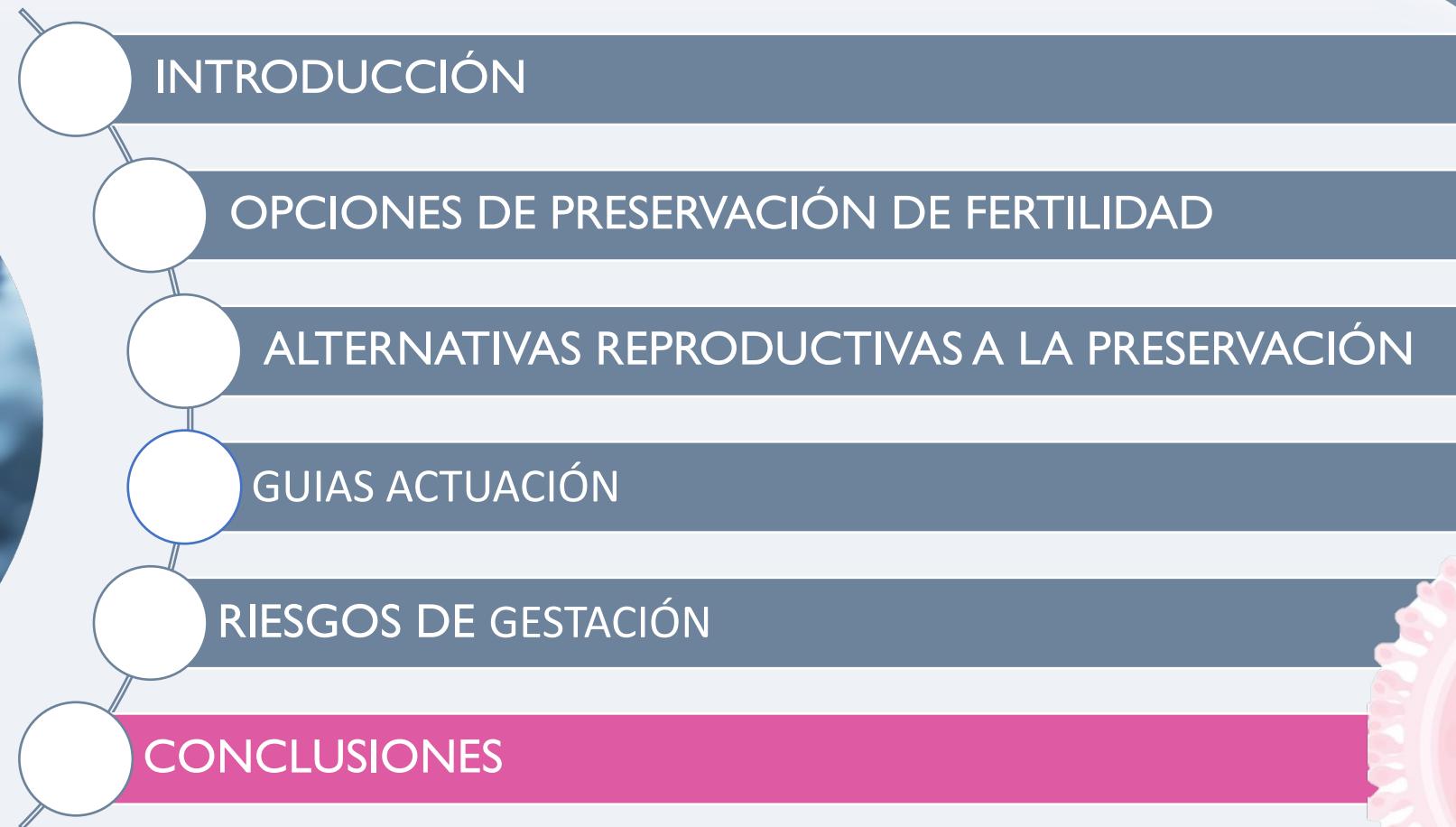
Evaluación gestación



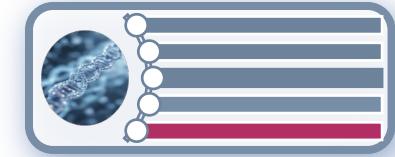
Necesidad de un equipo multidisciplinar (cardiología, obstetricia, endocrinología), control con imágenes seriadas de la aorta y estricto de la presión arterial durante el embarazo y el posparto



Opciones reproductivas en pacientes con SdR Turner



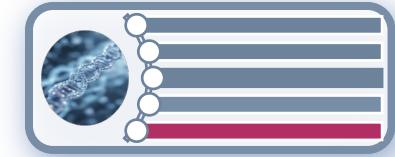
Conclusiones



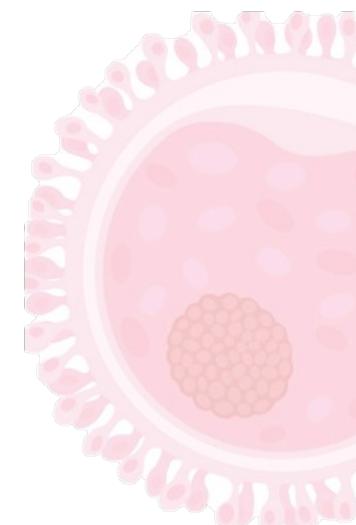
- El síndrome de Turner (ST) es la anomalía cromosómica más común en mujeres (1/2500 nacimientos).
- **Alta tasa de insuficiencia ovárica primaria e infertilidad.**
- **La reserva ovárica puede agotarse antes de la pubertad.**
- **Diagnóstico temprano** es clave para opciones de preservación de la fertilidad.
- Derivar precozmente a especialista en fertilidad.
- Evaluar **marcadores**: AMH, FSH, LH, inhibina-B, recuento de folículos antrales.
- **Identificar a las niñas con potencial de preservación.**



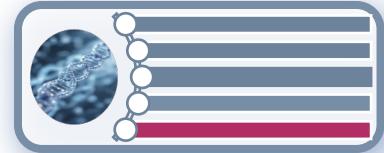
Conclusiones



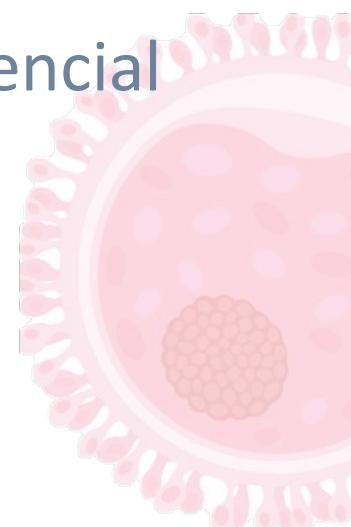
- Ofrecer **preservación fertilidad** siempre que haya **expectativas razonables**
- La **vitrificación de ovocitos** es la técnica de preservación de la fertilidad preferida, más utilizada y más exitosa hasta la fecha, y debe discutirse con las niñas con ST cuando su madurez psicológica lo permita.
- **Ovodonación, subrogación y adopción** amplían las posibilidades de maternidad.



Conclusiones



- Considerar **apoyo psicológico y ético**.
- **Abordaje multidisciplinar**, previo a preservación y previo a potencial gestación.





Comunidad
de Madrid

Dirección General de Humanización,
Atención y Seguridad del Paciente
CONSEJERÍA DE SANIDAD

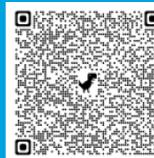
El Síndrome de Turner: Detección, Atención y Necesidades de las pacientes

Fecha: 27 de octubre 2025

Horario: 16:00 h a 20:00 h
Modalidad: Presencial

Lugar: Pº de la Castellana nº 280 (SERMAS)
Aula 0-Rs1

Cómo llegar



muchas gracias

