

Clinical Summary Guide

September 2022



HEALTHY MALE

Generations of healthy Australian men

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HEALTHY MALE

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Step-by-Step Male Genital Examination

Step-by-Step Male Genital Examination



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Testicular volume

Testicular volume is assessed using an orchidometer; a sequential series of beads ranging in size from 1 mL to 35 mL (see Image 1).

Conduct the examination in a warm environment, with the patient lying on their back.

1. Gently isolate the testis and distinguish it from the epididymis. Then stretch the scrotal skin, without compressing the testis.
2. Use your orchidometer to make a manual side-by-side comparison between the testis and beads (see Image 2).
3. Identify the bead most similar in size to the testis, while making allowance not to include the scrotal skin.

Normal testicular volume ranges

Childhood	Puberty	Adulthood
< 3 mL	4-14 mL	15-35 mL

Clinical notes

- Asymmetry between testes is common (e.g. 15 mL versus 20 mL) and not medically significant.
- Asymmetry is sometimes more marked following unilateral testicular damage.
- Testes are roughly proportional to body size.
- Low testicular volume suggests impaired spermatogenesis¹.
- Small testes (< 4 mL) from mid puberty are a consistent feature of Klinefelter syndrome².

Examination of secondary sexual characteristics

Gynecomastia

- Gynecomastia is the excessive and persistent development of benign glandular tissue evenly distributed in a sub-areolar position of one or both breasts (see Image 3)³.
- Can cause soreness and considerable embarrassment.
- Common during puberty, usually resolves in later adolescence³.
- Causes include increased estrogen, low testosterone, various medications, marijuana, androgen abuse and abnormal liver function^{3, 4, 5}.
- Distinguish glandular tissue from sub-areolar fat in obese subjects.
- Rare secondary causes include hypothalamic/pituitary and adrenal/testis tumours (oestrogen excess)⁴.
- Rapidly developing gynecomastia may indicate testicular tumour⁵.
- In contrast to gynecomastia, breast cancer can be located anywhere within the breast tissue and feels firm or hard.³

Onset of puberty

- Average onset is 12-13 years.

Virilisation

- Facial and body hair development.
- Muscle development.
- Penile growth.

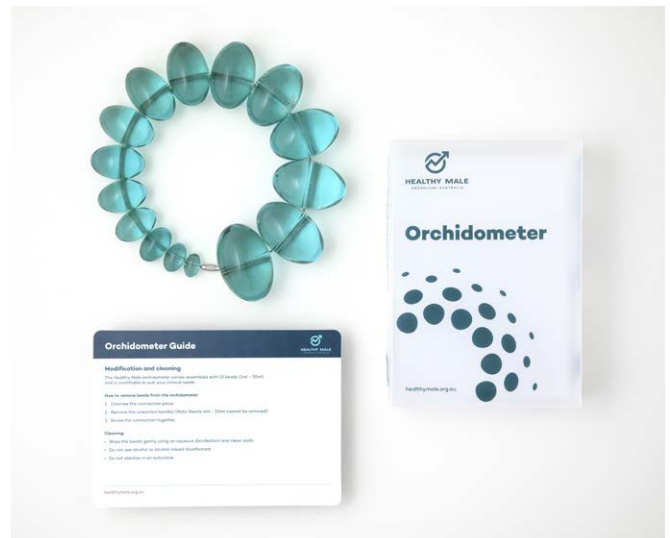


Image 1 – Orchidometer

Why use an orchidometer?

Testicular volume is important in the diagnosis of androgen deficiency, infertility and Klinefelter syndrome.

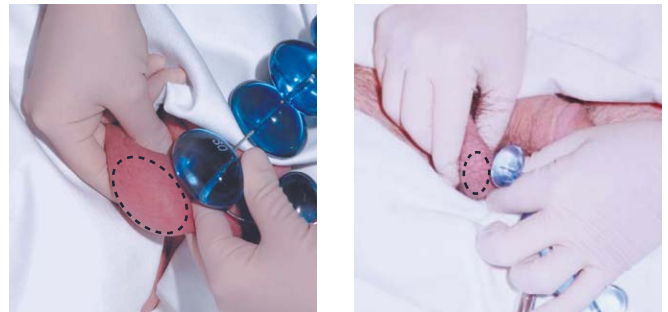


Image 2 – Example of 30 mL and 4 mL adult testis



Image 3 – Gynecomastia

(Photo courtesy of Mr G Southwick, Melbourne Institute of Plastic Surgery)

Examination of testis and scrotal contents

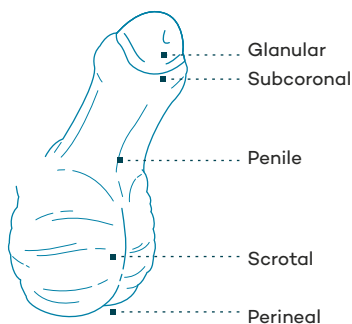
Testis	Gently palpate the testis between your thumb and first two fingers. Note: Atrophic testes are often more tender to palpation than normal testes.	If a testis cannot be felt, gently palpate the inguinal canal to see if testis can be 'milked' down. Note: Testis retraction can be caused by cold room temperature, anxiety and cremasteric reflex.	Examine the testis surface for irregularities. It should be smooth, with a firm, soft rubbery consistency. Note: A tumour may be indicated by deep or surface irregularity, or differences in consistency between testes.
Epididymis	Locate the epididymis, which lies along the posterior wall of the testis. It should be soft, slightly irregular and non-tender to touch.	Tenderness, enlargement or hardening can occur as a result of obstruction (vasectomy) or infection. This can be associated with obstructive infertility. Cysts in the epididymis are quite common. These are something mistaken for a testicular tumour.	
Vas deferens	Locate the vas deferens, a firm rubbery tube approximately 2-3 mm in diameter. The vas deferens should be distinguished from the blood vessels and nerves of the spermatic cord.	Nodules/thickening around the vas deferens ends may be apparent after vasectomy. Absence of the vas deferens is a congenital condition associated with low semen volume and azoospermia.	
Varicocele	Perform examination with the man standing. A Valsalva manoeuvre or coughing helps delineate smaller varicoceles.	Indicators include: <ul style="list-style-type: none"> • Palpable swelling of the spermatic veins above testis • Swelling is usually easy to feel and can be compressed without discomfort • Nearly always on left side • Associated with infertility. 	  <p>(Photo courtesy of Prof D de Kretser)</p>

Examination of penile abnormalities

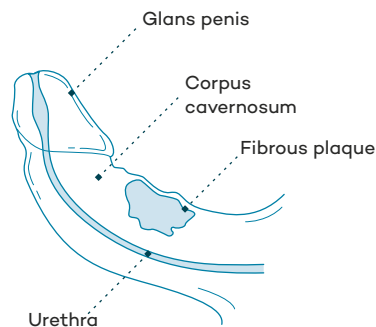
Hypospadias	Peyronie's disease	Micropenis	Phimosis	Urethral stricture
Abnormal position of meatus on the underside of the penile shaft. May be associated with a notched penile head.	Fibrous tissue, causing pain and curvature of the erect penis. Check for tenderness or thickening.	May indicate androgen deficiency prior to puberty.	The foreskin cannot be pulled back behind the glans penis. Can be normal in boys up to 5-6 years.	Abnormal urethral narrowing, which alters urination. Can be caused by scar tissue, disease or injury.

Hypospadias

Position of urethral opening



Peyronie's disease



(Photo courtesy of Dr M Lowy, Sydney Centre for Men's Health)

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2

**Child and
Adolescent
Male Genital
Examination**

Child and Adolescent Male Genital Examination



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When to perform an examination

A physical examination of male children and adolescents is vital for the detection of conditions such as testicular cancer, Klinefelter syndrome, and penile and hormonal abnormalities.

How to approach an examination with young patients

Good communication can assist the process of physical examinations with children and adolescents.

- Communicate with both the patient and their parents, using simple language and visual aids if available.
- Explain why you need to perform the examination and ask for permission to proceed.
- Allow the patient to ask questions and express any discomfort before/during the examination.
- When it seems appropriate, humour can be used (particularly with children) to reduce anxiety, foster rapport and improve cooperation before or during the examination.
- If you refer the patient to another specialist, take the time to explain why, and what may be involved.
- Never perform an examination of a child if they are restrained by a parent.
- Always wear gloves during an examination unless there is a specific indication for not doing so (e.g. neonatal examination, detection of a small scrotal mass).

Childhood history and examination

Presentation with acute testicular pain

- Testicular torsion.
- Refer immediately for evaluation for possible surgery.
- This is a medical emergency.
- Later follow up review (e.g. epididymo–orchitis).

History

- Undescended testes (increased risk of testicular cancer, and associated with inguinal hernia).
- Inguinal-scrotal surgery or hypospadias.

Testicular examination

- Undescended testes.
- Testicular volume: Normal childhood (pre-pubertal) range of testicular volume is ≤ 3 mL.

Penile examination

- Hypospadias.
- Micropenis.
- Phimosis (physiological or pathological).

Best time to perform an examination

1. Part of a standard health check-up with new or existing patients.
2. On presentation of relevant disorders or symptoms, including:

Risk factors	Associated disorders
Undescended testes as an infant	Testicular cancer
Delayed puberty	Androgen deficiency
Gynecomastia	Androgen deficiency Klinefelter syndrome Testicular cancer
Past history of testicular cancer	Testicular cancer
Acute testicular - groin pain	Testicular cancer
Testicular pain or lumps	Testicular torsion

Adolescent history and examination

Presentation with acute testicular pain

- Testicular torsion.
- Refer immediately for evaluation for possible surgery.
- This is a medical emergency.
- Later follow up review (e.g. epididymo–orchitis).

History

- Undescended testes.
- Pubertal development.
- Testicular trauma, lump and/or cancer.
- Gynecomastia.
- Prior inguinal-scrotal surgery or hypospadias.

Testicular examination

- Testicular volume.
 - Normal pubertal range is 4-14 mL.
 - < 4 mL by 14 years indicates delayed or incomplete puberty.
 - Small testes (< 4 mL) may suggest Klinefelter syndrome.
 - Adult testis size is established after completion of puberty.
- Scrotal and testicular contents.
 - Abnormalities in texture or hard lumps (tumour or cyst).

Penile examination

- Hypospadias.
- Micropenis.
- Infections (STI) or inflammation.
- Phimosis (physiological or pathological).
- Balanitis.

Examination of secondary sexual characteristics

- Gynecomastia: excessive and/or persistent breast development.
- Delayed puberty (average onset is 12-13 years). Indicators:
 - Short stature compared to family, with reduced growth velocity
 - Absent, slow or delayed genital and body hair development compared to peers
 - Anxiety, depression, school refusal, or behaviour change in school years 8-10 (age 14-16 years).

Puberty: delayed onset or poor progression

Presentation

- Short stature compared to family.
- Absent, slow or delayed genital development.
- Anxiety, depression, school refusal and/or behaviour change.

(±) Other features

- Headache/visual change (CNS lesions).
- Inability to smell (Kallmann's syndrome).
- Behavioural or learning difficulty (47,XXY).
- Unusual features (rare syndromes).

Primary investigations

- Growth chart in context of mid parental expectation (velocity, absolute height).
- Penile size (standard growth chart).
- Testicular volume (> 4 mL puberty imminent).
- Bone age.

Specific investigations

- LH/FSH (may be undetectable in early puberty but if raised can be useful).
- Total testosterone level (rises with onset of puberty).
- Karyotype (if suspicion of 47,XXY).

General investigations

- U&E, FBE & ESR, coeliac screen, TFT.

Treatment and specialist referral

- If all normal for prepubertal age, observe for 6 months.
- Refer to paediatric endocrinologist if patient is > 14.5 years without pubertal onset and/or a specific abnormality.

Klinefelter syndrome (47,XXY)

Presentation

- Small testes < 4 mL characteristic from mid puberty.
- Presentation varies with age and is often subtle.
- Behavioural and learning difficulties.
- Gynecomastia (adolescence).
- Poor pubertal progression (adolescence).

Investigations

- Total testosterone level (androgen deficiency).
- LH/FSH level (both elevated).
- Karyotype.

Treatment and specialist referral

- Refer to paediatric endocrinologist.
- Refer for educational and allied health assistance if needed.

Refer to Clinical Summary Guide 10: Klinefelter Syndrome

Testicular mass

Presentation

- Painless lump.
- Self report, incidental.
- Past history undescended testes (cancer risk).
- Consider possibility of epididymal cyst.

Primary investigations

- Testicular ultrasound.

Treatment and specialist referral

- Refer to uro-oncologist.
- Offer pre-treatment sperm cryostorage.

Refer to Clinical Summary Guide 6: Testicular Cancer

Penile abnormality

Presentation

- Hypospadias.
- Micropenis.
- Phimosi.

Treatment and specialist referral

- Refer to urologist for investigation and treatment plan.
- Refer to paediatric endocrinologist for investigation of micropenis.

Gynecomastia

Presentation in adolescence

- Excessive and/or persistent breast development.
- More prominent in obesity.
- Often normal, resolves over months.

Rare secondary causes

- Hypothalamic pituitary lesions.
- Adrenal/testis lesions (oestrogen excess).

Treatment and specialist referral

- If persistent or acute onset, refer to paediatric endocrinologist.

3

Adulthood Male Genital Examination



Adult Male Genital Examination

When to perform an examination

- As part of a standard health check-up with new or existing patients.
- 45-49 year old health assessment (MBS) (Note, Aboriginal and Torres Strait Islander men are eligible at younger ages).
- Prior to initiation of drug treatment (e.g. testosterone, PDE5 inhibitors) or investigation of conditions such as infertility or prostate disease.
- On presentation of relevant risk factors and symptoms (below).

Risk factors	Associated disorders
Undescended testes as an infant	Testicular cancer
Past history of delayed puberty	Androgen deficiency
Gynecomastia	Androgen deficiency, Klinefelter syndrome, testicular cancer
Infertility	Androgen deficiency, testicular cancer
Erectile dysfunction (ED)	Co-morbidities
Past history of testicular cancer	Testicular cancer
Pituitary disorders	Androgen deficiency, male infertility
Osteoporosis and atraumatic fractures	Androgen deficiency
Haemochromatosis	Androgen deficiency, male infertility

Symptoms	Associated disorders
Testicular pain or lumps	Tumour or cyst
Reduced libido, hot flushes, fatigue, gynecomastia, ED, mood changes, reduced beard or body hair, poor or reduced muscle development	Androgen deficiency

How to approach an examination with a patient

- Posters or pamphlets in your clinic can raise awareness about men's health examinations and convey that patients can discuss reproductive health concerns with you.
- Explain why you need to perform the examination and ask for permission to proceed.
- Allow the patient to ask questions and express any discomfort before/during the examination.
- Ask specific questions during history-taking, to assist those patients reluctant to raise sensitive problems.

Adulthood history and examination

Presentation with acute testicular pain

- This is a medical emergency.
- Testicular torsion.
- Refer immediately for evaluation for surgery.
- Later follow up review (e.g. epididymo-orchitis).

History

- Fertility in current and past relationships.
- Testicular trauma, cancer or STI.
- Inguinal-scrotal surgery (undescended testes, childhood hernia).
- Symptoms of androgen deficiency.
- Systemic treatment for malignancy, immunosuppression or organ transplant (for possible testicular damage).
- Gynecomastia.
- Occupational or toxin exposure.
- Past and present drug, alcohol or androgen use.
- Family history of haemochromatosis.

Testicular examination

Testicular volume

- Normal range for adult testicular volume is 15-35 mL.
- Small testes <4 mL suggests Klinefelter syndrome.

Scrotal and testicular contents

- Abnormalities in the texture or hard lumps suggests tumour or cyst.
- Enlargement, hardening or cysts of the epididymides.
- Varicocele.
- Nodules or absence of vas deferens.

Penile examination

- Hypospadias.
- Peyronie's disease.
- Micropenis.
- Urethral stricture.
- Evidence of infection (e.g. STI) or inflammation.
- Balanitis or phimosis.

Secondary sexual characteristics of androgen deficiency

- Reduced facial, body and pubic hair.
- Gynecomastia.
- Reduced or poor muscle development.

Prostate and other examinations

- In suspected prostate disease, digital rectal examination may be considered, or an initial referral to urologist.
- If prostate enlargement, tenderness or nodularity is found, refer to urologist.
- General medical review of erectile dysfunction.
- Focus on cardiovascular risk (BP, pulses) and diabetes (including neuropathy).

Androgen deficiency (AD)

Presentation

- Symptoms of AD in men of any age.
- Following testis surgery, torsion, trauma or cancer treatment.
- Incidental findings of small testes.
- In association with infertility.

Primary investigations

- Total testosterone level (two morning fasting samples, preferably using LC/MS) and LH/FSH level.

Investigations if low total testosterone with normal or low LH/FSH

- Serum prolactin (prolactinoma).
- MRI pituitary (various lesions).
- Olfactory testing (Kallmann's syndrome).
- Iron studies (haemochromatosis).
- Also commonly seen with co-morbidities (obesity, depression, chronic illness) — focus on underlying condition.

Other investigations

- SHBG/calculated free total testosterone (selected cases, e.g. obesity, liver disease).
- Bone density study (osteoporosis).
- Semen analysis (if fertility is an issue).
- Karyotype (if suspicion of 47,XXY).

Treatment and referral

- Testosterone replacement therapy (TRT).
 - *Contraindicated in prostate and breast cancer
 - *Withhold treatment until investigation complete
 - *Negatively impacts fertility
- In general, TRT is not justified in older men with borderline low testosterone levels and without underlying pituitary or testicular disease.
- Low-normal total testosterone is common in obesity or other illness and may not reflect AD. Address underlying disorders first.
- Consult a specialist to plan long term management:
 - Refer to endocrinologist
 - Refer to fertility specialist as needed.

Refer to Clinical Summary Guide 4: Androgen Deficiency

Klinefelter syndrome (47,XXY)

Presentation

- Small testes < 4 mL characteristic from mid puberty. Infertility (azoospermia) or androgen deficiency.
- Other features vary, and are often subtle. These include taller than average height, reduced facial and body hair, gynecomastia, behavioural and learning difficulties (variable), osteoporosis and feminine fat distribution.

Primary Investigations

- Total testosterone level (androgen deficiency).
- LH/FSH level (both elevated).
- Karyotype confirmation.

Other investigations

- Bone density study (osteoporosis).
- Semen analysis (usually azoospermic).
- TFT (hypothyroidism).
- Fasting blood glucose (diabetes).

Treatment and referral

- Develop a plan in consultation with an endocrinologist.
- Refer to endocrinologist, as TRT is almost always needed.
- Refer to fertility specialist as appropriate, for sperm recovery from testis (occasionally) or donor sperm.

Refer to Clinical Summary Guide 10: Klinefelter Syndrome

Penile abnormality

Presentation

- Hypospadias.
- Peyronie's disease.
- Micropenis.
- Urethral stricture.
- Phimosis.

Treatment and referral

- Refer to urologist for investigation and treatment plan.

Testicular mass

Presentation

- Painless lump.
- Self report, incidental.
- Past history undescended testes (cancer risk).
- Confirm lump is in testis rather than epididymal cyst.

Primary investigations

- Testicular ultrasound.

Treatment and referral

- Refer to uro-oncologist
- Offer pre-treatment sperm cryostorage.

Refer to Clinical Summary Guide 6: Testicular Cancer

Gynecomastia

Presentation in adulthood (common)

- Excessive and/or persistent breast development.
- Androgen deficiency.
- Chronic liver disease.
- Hyperprolactinaemia.
- Adrenal or testicular tumours.
- Drugs (e.g. spironolactone), marijuana or sex steroids.
- Distinguish from 'pseudogynecomastia' of obesity.

Primary investigations

- Total testosterone level, estradiol, FSH/LH.
- LFTs, iron studies (haemochromatosis).
- Serum prolactin (pituitary tumour).
- Karyotype (if suspicion of 47,XXY).
- β hCG, α FP, ultrasound (testicular cancer).

Treatment and referral

- Refer to endocrinologist.
- Refer to plastic surgeon (after evaluation) if desired.

Male infertility

Presentation

- Failure to conceive after 12 months of regular (at least twice weekly) unprotected intercourse.
- Consider early evaluation if patient is concerned and/or advancing female age an issue.

(±)Other features:

- Testis atrophy (androgen deficiency).
- Past history undescended testis (cancer risk).
- Psychosexual issues (primary/secondary).
- Past history STI (obstructive azoospermia).
- Androgen use (impaired gonadal function).

Primary investigations

- Semen analysis: twice at 6-week intervals. Analysis at specialised reproductive laboratory if abnormalities.
- FSH: increased level in spermatogenic failure.
- Testicular ultrasound (abnormal physical examination, past history of undescended testes).
- Total testosterone and LH (small testes < 12 mL or features of androgen level)

Treatment and referral

- Healthy lifestyle, cease smoking.
- Advice on natural fertility timing.
- Identification of treatable factors (often unexplained and no specific treatment).
- Refer to an endocrinologist as necessary.
- Refer to a fertility specialist (ART widely applicable).

Refer to Clinical Summary Guide 5: Male Infertility



4

Androgen Deficiency



Androgen Deficiency

The GP's role

GPs are generally the first point of contact for men with symptoms of androgen deficiency.

GPs are relied upon for clinical and laboratory examinations, appropriate referral, and ongoing patient management.

Patient referral to an endocrinologist, urologist or sexual health specialist is required for PBS-subsidised testosterone prescriptions.

Condition overview

Androgen deficiency is a syndrome caused by poor testicular function (hypogonadism), resulting from either primary (testicular) or secondary (hypothalamic-pituitary) disease, and is characterised by a low testosterone level accompanied by signs and symptoms^{1,2,3}.

It is estimated that approximately 5 in 1000 men have androgen deficiency warranting treatment with testosterone⁴.

A low testosterone level alone does not constitute androgen deficiency⁵, and neither does the normal age-related decline in testosterone (of approximately 1% annually⁶).

Androgen deficiency may have subtle effects on health and wellbeing, which can make diagnosis challenging.

Causes

Primary hypogonadism

- Chromosomal (e.g. Klinefelter syndrome (the most common cause of androgen deficiency)).
- Undescended testes.
- Trauma.
- Infection (e.g. mumps orchitis).
- Systemic disease (e.g. haemochromatosis, thalassaemia, myotonic dystrophy).
- Medical or surgical procedures (e.g. radiotherapy, chemotherapy, surgery (bilateral orchidectomy), medication (spironolactone, ketoconazole)).

See [Clinical Summary Guide 10: Klinefelter Syndrome](#)

Secondary hypogonadism

- Hypogonadotropic hypogonadism (e.g. Kallmann's syndrome).
- Pituitary micro- or macro-adenoma: typically macroprolactinoma.
- Pituitary trauma or disease.
- Medical or surgical procedures (e.g. pituitary radiotherapy or surgery).

Diagnosis

Medical history

- Undescended testes.
- Testicular surgery.
- Pubertal development or virilisation.
- Fertility.
- Genitourinary infection.
- Coexistent illness (e.g. pituitary disease, thalassaemia, haemochromatosis).
- Sexual function (all men presenting with erectile dysfunction should be assessed for androgen deficiency, even though it is an uncommon cause).
- Drug use (medical or recreational).

Clinical examination and assessment

Prepubertal onset

- Micropenis.
- Small testes.

Peripubertal onset

- Delayed or incomplete sexual and somatic maturation.
- Small testes.
- Attenuated penile enlargement.
- Attenuated pigmentation of scrotum.
- Attenuated laryngeal development.
- Attenuated growth of facial, body and pubic hair.
- Poor muscle development.
- Gynecomastia.

Postpubertal onset

- Regression of virilisation.
- Small testes.
- Mood changes (low mood and/or irritability).
- Poor concentration.
- Lethargy.
- Hot flushes and sweats.
- Low libido.
- Reduced growth of facial or body hair.
- Low semen volume.
- Gynecomastia.
- Reduced muscle mass and strength.
- Increased fat mass.
- Bone fracture (resulting from low bone mineral density).

Laboratory examinations and assessment

Serum total testosterone* (morning, fasting):

- Young men: (21-35 years) 10.4-30.1 nmol/l⁷; (19-22 years) 7.4-28.0 nmol/l⁸
- Healthy older men (71-87 years), 6.6-26.7 nmol/l⁹.

*Accurate serum testosterone measurements require mass spectrometry. Values from immunoassays are less reliable.

Serum FSH reference range

- Young adult: (21-35 years), 1.2-9.5 IU/ml⁷; (19-22 years), 1.3-12 IU/l⁸.
- Older adult (74-84), mean 10.11, 95% confidence intervals 9.27-11.02 IU/l¹⁰.

Serum LH reference ranges

- Young adult: (21-35 years), 1.5-8.1 IU/l⁷; (19-22 years), 5.1-18.7 IU/l⁸.
- Older adult (74-78 years), median 4.1, interquartile range 3.0-6.1¹¹.
- Older adult (84-87 years), median 6.8, interquartile range 4.3-10.4¹¹.

At least two measurements of serum testosterone, LH and FSH (from samples collected on separate days) are required for diagnosis of androgen deficiency.

PBS criteria require androgen deficiency to be confirmed by serum testosterone below 6 nmol/l, or 6-15 nmol/l with LH 1.5 times higher than reference range (or above 14 IU/l).

Subsequent investigations for treatable causes of androgen deficiency:

- Serum prolactin (for prolactinoma and macroadenoma)
- Iron studies and full blood count (for haemochromatosis and thalassaemia)
- Anterior pituitary function (for hypopituitarism and/or hyperfunctioning adenoma)
- Karyotyping (for suspected Klinefelter syndrome)
- Y chromosome microdeletion analysis
- Magnetic Resonance Imaging (for various hypothalamic or pituitary lesions).

Management

Testosterone replacement therapy (TRT)

TRT is aimed at relief of symptoms and signs of androgen deficiency, using convenient and effective (intramuscular or transdermal) testosterone preparations¹².

Product name	Usual (starting) dose	Dose range
Injectable (IM)		
Combined testosterone propionate, testosterone phenylpropionate, testosterone isocaproate, testosterone decanoate* Testosterone enantate*	250 mg fortnightly	250 mg at 10-21-day interval
Testosterone undecanoate	1000 mg twice at 6-week interval, followed by 12-weekly	1000 mg at 8-16-week interval

Product name	Usual (starting) dose	Dose range
Transdermal patch		
Testosterone	5 mg nightly	2.5-5 mg nightly
Transdermal gel		
Testosterone	50 mg daily	25-100 mg daily
Transdermal cream		
Testosterone	100 mg daily applied to upper body	Up to 200 mg daily (to torso)
	25 mg daily applied to scrotum	Up to 50 mg daily (to scrotum)
Oral		
Testosterone undecanoate	80 mg 2-3 times daily	80-240 mg daily

*Not PBS-subsidised

Contraindications and clinical considerations for TRT

TRT should be withheld until all investigations are complete.

Absolute contraindications for TRT:

- Known or suspected cancer of the prostate or breast
- Haematocrit > 55%.

Relative contraindications for TRT:

- Haematocrit > 52%
- Untreated sleep apnoea
- Severe urinary obstructive symptoms of benign prostatic hyperplasia (international prostate symptom score > 19)
- Advanced congestive heart failure.

Exogenous testosterone suppresses spermatogenesis in eugonadal men. Men with secondary hypogonadism who wish to preserve fertility should be managed using gonadotrophin therapy.

Monitoring TRT

Alleviation of a patient's leading symptom is the best clinical measure of effective management.

Blood sampling for serum testosterone, LH and FSH measurement should be timed to allow estimation of steady-state testosterone levels, which is feasible by sampling during the trough (immediately before next dose) for men using injectable and transdermal preparations. Timing of sampling for accurate measurement in men taking oral testosterone is more difficult.

Random sampling of blood for measurement of serum testosterone, without consideration of dosage timing is effectively useless.

Persistently elevated LH levels during TRT may indicate inadequate dosing.

Periodic monitoring (1-2 year intervals) of bone mineral density may assist in monitoring TRT.

Haematology profile should be assessed 3 months after initiating TRT and annually thereafter.

Monitoring for prostate disease in men using TRT should occur as for eugonadal men of the same age.

Referral

PBS-subsidised prescription of TRT requires treatment by, or in consultation with, a specialist endocrinologist, urologist or registered member of the Australian Chapter of Sexual Health Medicine.

Long-term management of androgen deficiency is best planned in consultation with a specialist endocrinologist.

Refer to a fertility specialist as needed.

Refer males aged > 14.5 years with delayed puberty to a paediatric endocrinologist.

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5

Male Infertility



Male Infertility

The GP's role

- Do not wait before beginning assessments.
- GPs can begin with simple, inexpensive and minimally invasive investigations.
- Infertility needs to be assessed and managed as a couple, and may require several different specialists.
- See Healthy Male's Male Fertility Assessment tool to accompany this guide on our website (healthymale.org.au).

Diagnosis

Brief assessment and pre-pregnancy advice

Age	What age is the couple?
Fertility history	How long have they been trying to conceive, and have they ever conceived previously (together/separately)? Do they have any idea why they have not been able to conceive?
Contraception	When it was ceased, and the likely speed of its reversibility
Fertile times	Whether the couple engage in regular intercourse during fertile times
Female risk factors	Aged 35+, irregular menstrual cycles, obesity, painful menses or concomitant medical conditions
Female health	Screening for rubella and chicken pox immunity, Cervical Screening Test (25 years or older)
Lifestyle: female	Diet, exercise, alcohol, smoking cessation and folate supplementation
Lifestyle: male	Diet, exercise, alcohol and smoking cessation

Reproductive history

Assess	Why?
Prior paternity	Previous fertility
Psychosexual issues (erectile, ejaculatory)	Interference with conception
Pubertal development	Poor progression suggests underlying reproductive issue
A history of undescended testes	Risk factor for infertility and testis cancer
Past genital infection (STI), mumps infection or trauma	Risk for testis damage or obstructive azoospermia
Symptoms of androgen deficiency	Indicative of hypogonadism
Previous inguinal, genital or pelvic surgery	Testicular vascular impairments, damage to vasa, ejaculatory ducts, ejaculation mechanisms
Medications, alcohol, tobacco, illicit drugs and androgens	Transient or permanent damage to spermatogenesis
General health (diet, exercise and smoking)	Epigenetic damage to sperm affecting offspring health

Physical examination

General examination	Acute/chronic illness, nutritional status
Genital examination	Refer to Clinical Summary Guide 1: Step-by-Step Male Genital Examination
Lack of virilisation	Androgen deficiency/Klinefelter syndrome
Prostate examination	If history suggests prostatitis/STI

Investigations

Semen analysis is the primary investigation for male infertility¹.

Key points

- Men should abstain from sexual activity for between 2-7 days before sample collection.
- Semen analysis provides guidance to fertility; it is not a direct test of fertility. Fertility remains possible even in those with severe deficits.

Reference limits for semen analysis²

Volume	≥ 1.5 mL
pH	≥ 7.2
Sperm concentration	≥ 15 million spermatozoa/mL
Motility	≥ 40% motile within 60 minutes of ejaculation
Vitality	≥ 58% live
White blood cells	< 1 million/mL
Sperm antibodies	< 50% bound motile sperm

Serum total testosterone

- Testosterone is often normal 8-27 nmol/L*, even in men with significant spermatogenic defects.
- Some men with severe testicular problems display a fall in testosterone levels and rise in serum LH, these men should undergo evaluation for AD.
- The finding of low serum testosterone and low LH suggests a hypothalamic-pituitary problem (e.g. prolactinoma) (serum prolactin levels required).

* Testosterone reference range may vary between laboratories.

Serum FSH levels

- Elevated levels are seen when spermatogenesis is poor (primary testicular failure).
- In normal men, the upper reference value is approximately 8IU/L.
- In an azoospermic man:
 - 14 IU/L suggests spermatogenic failure
 - 5 IU/L suggests obstructive azoospermia but a testis biopsy may be required to confirm that diagnosis.

Management

Treatment options

Protecting and preserving fertility

Mumps vaccination, sperm cryopreservation (prior to chemotherapy, vasectomy or androgen replacement), safe sex practices, and early surgical correction of undescended testes.

Options for improving natural fertility

It may be possible to improve fertility for a minority of infertile men, including those with pituitary hormonal deficiency or hyperprolactinemia, genitourinary infection, erectile and psychosexual problems, and through the withdrawal of drugs. Evidence for varicocele removal to improve fertility is limited but may have a place in selected cases — seek specialist input.

Assisted reproductive technology (ART)

ART options range in cost and invasiveness:

- Artificial insemination with men's sperm at midcycle
- Conventional IVF
- Intracytoplasmic sperm injection (ICSI) for severe male factor problems. Sperm can be readily obtained by testicular needle aspiration in the setting of obstructive azoospermia. Some azoospermic men with spermatogenic failure may have sperm recovered for ICSI by microdissection testicular sperm extraction (micro-TESE).

Donor insemination

For men with complete failure of sperm production.

Specialist referral and long-term management

Warning: Never institute testosterone replacement therapy in a newly recognised androgen deficient man who is seeking fertility. The fertility issue must be addressed first as testosterone therapy has a potent contraceptive action via suppression of pituitary gonadotrophins and sperm output.

When should I refer a patient?

GPs can refer couples immediately or after a few months during which baseline tests are performed.

Referral will depend on the associated problem

- Endocrinologist (endocrine associated problems).
- Urologist (undescended testes, surgery).
- Fertility specialist/ART clinic that offers full assessment, including examination of the male partner.

Long-term management

- Includes assessment for late-onset androgen deficiency, testis cancer.

Fertility clinics

A list of Australian ART Clinics, accredited by the Reproductive Technology Accreditation Committee are available via the Fertility Society of Australia website fertilitysociety.com.au.

Supporting the couple

- Acknowledge both partners' experience of infertility, and encourage couple communication.
- Provide empathy and normalise feelings of grief and loss.
- Refer on to a psychologist or counsellor if the couple require further support.

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6

Testicular Cancer



Testicular Cancer

The GP's role

GPs are typically the first point of contact for men who have noticed a testicular lump, swelling or pain. The GP's primary role is assessment, referral and follow-up.

- All suspected cases must be thoroughly investigated and referred to a urologist.
- Treatment frequently requires multidisciplinary therapy that may include the GP.
- Most patients will survive, hence the importance of long-term regular follow-up.

Note on screening: There is little evidence to support routine screening. However, GPs may screen men at higher risk, including those with a history of previous testicular cancer, undescended testes, infertility or a family history of testicular cancer.

Overview

- Testicular cancer is the second most common cancer in Australian men aged 20-39 years¹. It accounts for about 20% of cancers in men aged 20-39 years and between 1% and 2% of cancers in men of all ages.
- The majority of tumours are derived from germ cells (seminoma and non-seminoma germ cell testicular cancer).
- More than 70% of patients are diagnosed with stage I disease (pT1)².
- Testicular tumours show excellent cure rates of > 95%, mainly due to their extreme chemo- and radio-sensitivity.
- A multidisciplinary approach offers acceptable survival rates for metastatic disease.

Benign cysts

Epididymal cysts, spermatocele, hydatid of Morgagni and hydrocele are all non-cancerous lumps that can be found in the scrotum. Diagnosis can be confirmed via an ultrasound.

Epididymal cysts	Common fluid-filled cysts which feel slightly separate from the testis and are often detected when pea-sized. Should be left alone when small, but can be surgically removed if they become symptomatic.
Spermatocele	Fluid-filled cysts containing sperm and sperm-like cells. These cysts are similar to epididymal cysts except they are typically connected to the testis.
Hydatid of Morgagni	Small common cysts located at the top of the testis. They are moveable and can cause pain if they twist. These cysts should be left alone unless causing pain.
Hydrocele	A hydrocele is a swelling in the scrotum caused by a buildup of fluid around the testes. Hydroceles are usually painless but gradually increase in size and can become very large. Hydroceles in younger men may be a warning of an underlying testis cancer, albeit rarely. In older men, hydroceles are almost always a benign condition, but a scrotal ultrasound will exclude testicular pathology.

Diagnosis and management

Medical history

- Scrotal lump.
- Genital trauma.
- Pain.
- History of subfertility or undescended testis.
- Sexual activity/history of urine or sexually transmitted infection.

Physical examination

- Perform a clinical examination of the testes and general examination to rule out enlarged nodes or abdominal masses.

Clinical notes

On clinical examination it can be difficult to distinguish between testicular and epididymal cysts. Lumps in the epididymis are rarely cancer. Lumps in the testis are nearly always cancer.

Refer to [Clinical Summary Guide 1: Step-by-Step Male Genital Examination](#)

Ultrasound

- Organise ultrasound of the scrotum to confirm testicular mass (urgent, organise within 1-2 days).
- Always perform in young men with retroperitoneal mass.

Investigation and referral

- Advice on next steps for investigation and treatment.
- Urgent referral to urologist (seen within 2 weeks).
- CT scan of chest, abdomen and pelvis.
- Serum tumour markers (AFP, hCG, LDH) before orchidectomy: may be ordered by GP prior to urologist consultation.
- Semen analysis and hormone profile (testosterone, FSH, LH).
- Discuss sperm banking with all men prior to treatment.
- Fine needle aspiration: scrotal biopsy or aspiration of testis tumour is not appropriate or advised.

Clinical notes

The urologist will form a diagnosis based on inguinal exploration, orchidectomy and en bloc removal of testis, tunica albuginea, and spermatic cord. Organ-sparing surgery can be attempted in specific cases (solitary testis or bilateral tumours) in specialist referral centres.

Follow-up

Patient follow-up (in consultation with treating specialist) for:

- Recurrence
- Monitoring the contralateral testis by physical examination
- Management of complications, including fertility.

American Joint Committee on cancer staging of testicular cancer^{3, 4}

pT – Primary Tumour*

pTX	Primary tumour cannot be assessed.
pT0	No evidence of tumour.
pTis	Germ cell neoplasia in situ.
pT1	Tumour limited to testis (including rete testis invasion) without vascular/lymphatic invasion (LVI).
T1a	Pure seminoma < 3 cm in size.
T1b	Pure seminoma [\geq] 3 cm in size.
pT2	Tumor limited to testis (including rete testis invasion) with LVI, or tumor invading hilar soft tissue or epididymis or penetrating visceral mesothelial layer covering the external surface of tunica albuginea with or without LVI.
pT3	Tumour invades spermatic cord with or without LVI.
pT4	Tumour invades scrotum with or without LVI.

Regional lymph nodes*

NX	Regional lymph nodes were not assessed.
NO	No positive regional nodes.
N1	Metastasis with a lymph node mass [\geq] 2 cm in greatest dimension, or multiple lymph nodes, none more than 2 cm in greatest dimension.
N2	Metastasis with a lymph node mass more than 2 cm but not more than 5 cm in greatest dimension or multiple lymph nodes, any one mass more than 2 cm but not more than 5 cm in greatest dimension.
N3	Metastasis with a lymph node mass > 5 cm in greatest dimension.

*Clinical (based on clinical examination and histological assessment) or Pathological (based on histological examination post-orchidectomy) lymph node classifications may be made, denoted by the prefix 'c' or 'p', respectively (e.g. pN1, cN2).

Distant metastasis

MX	Distant metastasis cannot be assessed.
MO	No distant metastasis.
M1	Distant metastasis.
M1a	Nonretroperitoneal nodal or pulmonary metastases.
M1b	Nonpulmonary visceral metastases.

Serum markers†

Sx	Serum markers not available or not performed.
S0	Serum marker study levels within normal limits.
S1	LDH < 1.5 x Normal* and hCG < 5000 mIU/mL and AFP < 1000 ng/mL.
S2	LDH 1.5-10 x Normal or hCG 5000-50,000 mIU/mL or AFP 1000-10,000 ng/mL.
S3	LDH > 10 x Normal or hCG > 50,000 mIU/mL or AFP > 10,000 ng/mL.

† LDH, lactate dehydrogenase; hCG, human chorionic gonadotrophin; AFP, alpha fetoprotein

* Upper limit of normal for LDH assay

Treatment options for localised testicular cancer

Orchidectomy cures almost 85% of stage I seminoma patients and 70-80% of stage I non-seminomatous germ cell tumour (NSGCT) patients. Adjuvant treatments may reduce the risk of metastases in those not cured by orchidectomy, but this comes at the cost of possible adverse effects. Surveillance is another management option. A risk-adapted approach is now used to determine subsequent management.

pT1 Seminoma

- Surveillance is recommended (if facilities are available and the patient willing and able to comply).
- Carboplatin-based chemotherapy decreases recurrence rates by 75% or 90%, for one or two courses, respectively⁵.
- Adjuvant treatment not recommended for patients at very low risk (< 4 cm size, absence of rete testis invasion).
- Radiotherapy is not recommended as adjuvant treatment, although it is a treatment option.

pT1 Non-Seminomatous Germ Cell Tumour (NSGCT)

Low risk

(No Lymphovascular invasion, Embryonal component < 50%, Proliferative index < 70%).

- If the patient is able and willing to comply with a surveillance policy, long-term (at least 5 years) close follow-up should be recommended.
- In patients not willing (or unsuitable) to undergo surveillance, adjuvant chemotherapy or nerve-sparing retroperitoneal lymph node dissection (RPLND) are options⁵.

High risk

(Lymphovascular invasion, pT2-pT4)

- Adjuvant chemotherapy with one or two courses of bleomycin, etoposide and cisplatin (BEP) is recommended.
- If the patient is not willing to undergo chemotherapy or if chemotherapy is not feasible, nerve-sparing RPLND or surveillance with treatment at relapse (in about 50% of patients) are options⁶.

Treatment of metastatic disease (pT2-pT4)

The treatment of metastatic germ cell tumours⁶ depends on:

- The histology of the primary tumour and
- Prognostic groups as defined by the International Germ Cell Cancer Collaborative Group (IGCCCG)⁷.

Seminoma

- Radiotherapy (30Gy), or chemotherapy (BEP) can be used with the same schedule as for the corresponding prognostic groups for NSGCT.
- Any pT, N3 seminoma is treated as "good prognosis" metastatic tumour with three cycles of BEP or four cycles of EP.
- PET scan plays a role in evaluation of post-chemotherapy masses larger than 3 cm.

NSGCT

- Low volume NSGCT with elevated markers (good or intermediate prognosis), three or four cycles of BEP; if no marker elevation, repeat staging at 6 weeks surveillance to make final decision on treatment.
- Metastatic NSGCT with a good prognosis, primary treatment three courses of BEP.
- Metastatic NSGCT with intermediate or poor prognosis, four courses of BEP and inclusion in clinical trial recommended.
- Surgical resection of residual masses after chemotherapy in NSGCT is indicated in case of visible residual mass and when tumour marker levels are normal or normalising.

IGCCCG Prognostic- based staging system for metastatic germ cell cancer⁷

Prognosis	Seminoma	Non-Seminoma
Good (If ALL criteria are met)	Any primary site. <ul style="list-style-type: none"> No non-pulmonary metastases. Normal AFP/normal LDH, low hCG. 	If all criteria are met: <ul style="list-style-type: none"> Testis/retroperitoneal primary No non-pulmonary metastases (e.g. liver and/or brain) Lower levels of tumour markers.
Intermediate (If ALL criteria are met)	If all criteria are met: <ul style="list-style-type: none"> Any primary site No non-pulmonary metastases Normal AFP/normal LDH, medium hCG. 	If all criteria are met: <ul style="list-style-type: none"> Testis/retroperitoneal primary No non-pulmonary metastases (e.g. liver and/or brain) Medium levels of tumour markers.
Poor (If ANY criteria are met)	No seminoma carries poor prognosis.	If any criteria are met: <ul style="list-style-type: none"> Non-pulmonary metastases (e.g. liver and/or brain) Higher level of tumour markers Mediastinal primary for NSGCT.

Additional Investigations

Serum tumour markers

Post-orchidectomy half-life kinetics of serum tumour markers.

- The persistence of elevated serum tumour markers 6 weeks after orchidectomy may indicate the presence of metastases, while its normalisation does not necessarily mean an absence of tumour.
- Tumour markers should be assessed until they are normal, as long as they follow their half-life kinetics and no metastases are revealed on scans.

Other examinations

Assessment of abdominal and mediastinal nodes and viscera (CT scan) and supraclavicular nodes (physical examination).

- Other examinations such as brain or spinal CT, bone scan or liver ultrasound should be performed if metastases are suspected.
- Patients diagnosed with testicular seminoma who have a positive abdominal CT scan are recommended to have a chest CT scan.
- A chest CT scan should be routinely performed in patients diagnosed with NSGCT because in 10% of cases small, subpleural nodes are present that are not visible radiologically.

Classification and risk factors

There are three categories of testicular epithelial cancer. Germ cell tumours account for 90-95% of cases of testicular cancer⁸.

1. Germ cell tumours

- | | |
|-------------|-------------------------|
| a. Seminoma | b. Non-seminoma (NSGCT) |
| | - Embryonal carcinoma. |
| | - Yolk sac tumour. |
| | - Choriocarcinoma. |
| | - Teratoma. |

2. Sex cord stromal tumours

3. Non-specific stromal tumours

Prognostic risk factors

Pathological (pT1-pT4)

- | | |
|--------------------------------|--|
| • Histopathological type | • For non-seminoma |
| • For seminoma | - Vascular/lymphatic invasion or peri-tumoural invasion. |
| - Tumour size (> 4 cm). | - Percentage embryonal carcinoma > 50%. |
| - Invasion of the rete testis. | - Proliferation rate (MIB-1) > 70%. |

Clinical (for metastatic disease)

- Primary location.
- Elevation of tumour marker levels (AFP, hCG, LDH).
- Presence of non-pulmonary visceral metastasis.
- Only clinical predictive factor for metastatic disease in seminoma.

Staging of testicular tumours

The Tumour, Node, Metastasis (TNM) system³ is recommended for classification and staging purposes. The IGCCCG staging system is recommended for metastatic disease.

Treatment

- The first stage of treatment is usually an orchidectomy: removal of the diseased testis via an incision in the groin, performed under general anaesthetic. Men can be offered a testicular prosthesis implant during or following orchidectomy.
- Further treatment depends on the pathological diagnosis (seminoma vs non-seminoma and the stage of disease) and may include surveillance, chemotherapy or radiotherapy⁹.
 - Men with early stage seminoma have treatment options of surveillance, chemotherapy or radiotherapy. The treatment is based on patient and tumour factors.
 - Men with early stage non-seminoma have treatment options of surveillance, chemotherapy or further surgery. The treatment is again based on patient and tumour factors.
 - Men with early stage disease who relapse and men with advanced disease are generally referred for chemotherapy. If chemotherapy leaves residual masses, these may contain cancer and usually will need surgical removal.
- If a man has a bilateral orchidectomy (rare) he will require ongoing testosterone replacement therapy.

Patient support

Diagnosis and treatment can be extremely traumatic for the patient and family. Regular GP consultations can offer patients a familiar and constant person with whom to discuss concerns (e.g. about treatment, cancer recurrence, and the effects of testis removal on sexual relationships and fertility). Referral to a psychologist may be required.

Patient follow-up

- Regular follow-up is vital, and patients with testicular cancer should be watched closely for several years. The aim is to detect relapse as early as possible, to avoid unnecessary treatment and to detect asynchronous tumour in the contralateral testis (incidence 5%).
- Plan follow-ups in conjunction with the urologist/oncologist. Follow-up schedules are tailored to initial staging and treatment, and can involve regular physical examination, tumour markers and scans to detect recurrence. The timing and type of follow-ups need to be determined for each patient in conjunction with the treating urologist/oncologist.

Semen storage

- Men with testicular cancer often have low or even absent sperm production even before treatment begins^{10,11}. Chemotherapy or radiotherapy can, but does not always, lower fertility further². All men should be offered pre-treatment semen analysis and storage as semen can be stored long-term for future use in fertility treatments. Men who have poor sperm counts may need to visit the sperm-banking unit on 2 or 3 occasions or, in severe cases, an Andrology referral may be required. Surgical removal of one testis does not affect the sperm-producing ability of the remaining testis.
- Provide prompt fertility advice to all men considering chemotherapy or radiotherapy, to avoid delaying treatment. It is highly recommended that men produce semen samples for sperm storage prior to treatment.
- Sperm storage for teenagers can be a difficult issue requiring careful and delicate handling. Coping with the diagnosis of cancer at a young age and the subsequent body image problems following surgery can be extremely difficult. Fatherhood is therefore not likely to be a priority concern. Producing a semen sample by masturbation can also be stressful for young men in these circumstances.
- Refer the patient to a fertility specialist or a local infertility clinic. These clinics usually offer long-term sperm storage facilities.

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7

Prostate Disease



Prostate Disease

Benign Prostatic Hyperplasia (BPH)

The GP's role

- GPs are typically the first point of contact for men with BPH.
- The GP's role in the management of BPH includes clinical assessment, treatment, referral and follow-up.

Overview

- BPH is the non-cancerous enlargement of the prostate gland¹.
- Whilst not normally life threatening, BPH can impact considerably on quality of life².

Diagnosis

Medical history

- Lower urinary tract symptoms (LUTS).

Urinary symptoms of BPH

Obstructive symptoms:

- Hesitancy
- Weak stream
- Post micturition dribble
- Sensation of incomplete bladder emptying.

Overactive symptoms:

- Frequency
- Urgency (if severe incontinence)
- Nocturia.

Other:

- Nocturnal incontinence
- Urinary retention.

Some men with BPH may not present with many or any symptoms of the disease.

Symptom score

- Evaluation of symptoms contributes to treatment allocation and response monitoring.
- The International Prostate Symptom Score (IPSS)³ questionnaire is recommended.

Physical examination

- Digital rectal examination (DRE) can estimate prostate size and identify other prostate pathologies.
- Basic neurological examination.
- Perianal sensation and sphincter tone.
- Bladder palpation.
- Calibre of the urethral meatus.
- Phimosis.

Investigations

- Urinalysis or midstream urine.
- If suspect urinary retention/large post void residual volume.
 - Ultrasound (Kidneys and bladder).
 - Renal function (Creatinine).
- PSA:
 - If suspect prostate cancer (e.g. based on prostate examination)
 - As part of screening of prostate cancer, after discussion of pros and cons
 - Routine PSA screening is not necessary for patients with BPH. Patients with LUTS are not at increased risk of having prostate cancer.

PSA levels for different age groups of Western men⁴

Age range years	Serum PSA (ng/mL) median	Serum PSA (ng/mL) upper limit of normal
40-49	0.7	2.5
50-59	1.0	3.5
60-69	1.4	4.5
70-79	2.0	6.5

Other PSA tests

- PSA velocity or doubling time: if the PSA level doubles in 12-months it may indicate prostate cancer or prostatitis. An elevated PSA and a stable velocity suggest BPH.
 - Free-to-total PSA ratio: high ratio (> 25%) suggests BPH; low ratio (< 10%) suggests prostate cancer.
 - Prostate Health Index (PHI): not covered by the MBS, PHI thought to be more specific for diagnosing prostate cancer than PSA level alone; good quality evidence lacking & not recommended in Australian prostate cancer testing guidelines.
- Creatinine levels.
- Post-void residual urine (ultrasound).

Investigations by the urologist

- As per GP investigations as indicated +/-.
- Uroflowmetry and post void residual assessment.
- Voiding diary.
- Cystoscopy.
- Urodynamic assessment.

Management

Observation and review: for mild or low impact symptoms

- Optimise through reassurance, education, periodic monitoring and lifestyle modifications.
- Consider adjustment of medication (e.g. timing of diuretic).

Medical therapy: for moderate to severe symptoms

Alpha blockers (once daily)

- These are generally 1st line.
 - Amsulosin.
 - Silodosin.
 - Alfuzosin.
- Side effect profiles may favour tamsulosin.

5 α -reductase-inhibitors (5ARIs)

- Dutasteride.
- Finasteride.

Very rarely used as monotherapy.

Combination therapy

- Dutasteride and tamsulosin.
- Better for patients with large prostates (> 30 ml).
- 5ARI can affect sexual function so consider carefully in sexually active men.
- Controversy regarding 5ARI association with prostate cancer risk so recommend PSA surveillance. If PSA increased whilst on 5ARI, refer to urologist to exclude prostate cancer.

Other drugs

Bladder directed medications are most commonly used for overactive bladder symptoms.

- Beta-3 adrenergic agonist – Mirabegron.
 - Requires blood pressure monitoring within first week.
- Anticholinergics.
 - Oxybutynin.
 - Solifenacin.
 - Darifenacin.
- Side effects include dry mouth, dry eyes and/or constipation.

Urologist referral

Treatment

- Urinary retention history.
- Urinary tract infection.
- Haematuria.
- Failed medical therapy.
- Incontinence (of any type).
- Post void residual of > 100 ml.
- Severe symptoms (especially if poorly responsive to medications).
- Renal impairment.
- Bladder stones.
- Cancer suspected — prostate or bladder.
- Associated Neurological condition (e.g. Parkinson's disease, Multiple sclerosis).

Surgery

Indications for surgery are similar to the indications for referral to a urologist. Surgery can be considered when medications are no longer suitable for whatever reason. Cessation of medication therapy usually results in recurrence of symptoms.

There are multiple operations available. The gold standard operation is a transurethral resection of the prostate (TURP). There are however numerous operations that are available. Each has their pros and cons. When considering the operation the patient is undertake there are different factors that are considered such as:

- Prostate size/configuration
- Anti-coagulation status
- Side effects (e.g. preference to preserve antegrade ejaculation)
- Day stay vs overnight stay
- Catheter duration
- Co-morbidities (long term SPC)
- Durability of operation.

The operations available include:

- Transurethral Resection of the Prostate (TURP)
- Transurethral Incision of the Prostate (TUIP)
- Green light laser resection of the prostate
- Minimally invasive insertion of small retractors into prostate
- Plasma Vaporisation
- Water Vapour therapy
- Holmium Laser Enucleation of the Prostate (HoLEP).

Long term catheterisation:

- Last resort for patients unfit/unwilling for surgery but with complications (e.g. urinary retention)
- Supra-pubic catheter is preferred to indwelling urethral catheter
- Intermittent Self catheterisation should also be considered in such patients.

Follow-up

- It is appropriate for the GP to monitor and follow-up a patient with respect to all the treatment modalities. However, if the patient is not responding to medical treatment, refer to the urologist.
- Men who have had TURP remain at risk for prostate cancer and need routine prostate cancer checks, as per guidelines.

Recommended follow-up timeline after BPH treatment

Treatment modality	First year after treatment			Annually thereafter
	6 weeks	12 weeks	6 months	
Observation and review	X	X	✓	✓
5 α -reductase inhibitors	X	✓	✓	✓
α -blockers	✓	X	✓	✓
Surgery or minimal invasive treatment	✓	✓	✓	✓

Prostatitis

The GP's role

- GPs are typically the first point of contact for men with prostatitis.
- The GP's role in the management of prostatitis includes clinical assessment, treatment, referral and follow-up.

Overview

- Prostatitis is inflammation of the prostate gland.
- It can be a result of bacterial or non-bacterial infection.
- Acute bacterial prostatitis, the least common form, can be serious if the infection is left untreated.
- Whilst not normally life threatening, prostatitis can impact considerably on a man's quality of life.

Diagnosis

Medical history

- Urinary symptoms.
- Pain.

Symptoms of prostatitis

- Dysuria — painful urination.
- Urgent need to urinate.
- Frequent urination.
- Painful ejaculation.
- Lower back pain.
- Perineal pain.
- Chills and/or fever.
- Muscular pain.
- General lack of energy.

Investigations

- Digital rectal examination (DRE):
 - Should not be performed if you suspect acute severe prostatitis because it can be very painful
 - Some tenderness and swelling may accompany sub-acute prostatitis.
- Prostate specific antigen (PSA) levels:
 - Levels may be dramatically high
 - PSA velocity: if the PSA level doubles in 12-months it may indicate prostate cancer or prostatitis.
- Urine analysis:
 - First pass urine: Chlamydia urine PCR test
 - Midstream urine: MC&S
 - Urine PCR for STIs should be done if Chlamydia or other STI a likely cause.

Management

Treatment

- There are several therapeutic options available. Evidence for benefits of these treatment options is limited; however, they may be trialled with the patient.
- Urologists' use of the following forms of treatment will vary according to the individual, their condition and the stage of their treatment.
- Most patients will have antibiotic therapy at some stage.

Bacterial prostatitis (acute and chronic) can be treated using antibiotics. Once diagnosed, rapid treatment is essential to avoid further complications.

Chronic nonbacterial prostatitis (chronic prostate pain syndrome); treatment is difficult and cure is often not possible. Treatment focus is on symptom management, to improve quality of life. Non-medical therapy is recommended as the initial treatment.

Medication options⁵

- α -blockers.
 - Suited to patients with moderate/severe LUTS.
 - Tamsulosin.
 - Silodosin.
 - Alfuzosin.
 - Side effect profiles may favour tamsulosin.
- Antibiotics (not all antibiotics penetrate the prostate gland).
 - Recommend: Norfloxacin, Ciprofloxacin, Trimethoprim, Sulphamethoxazole/Trimethoprim, Erythromycin, Gentamicin.
 - Young men with confirmed Chlamydia prostatitis: Doxycycline.
- Analgesics.
- Non-steroidal anti-inflammatory drugs.

Surgical options

- Transurethral incision of the bladder neck.
- Transurethral resection of the prostate.

Surgery has a very limited role and requires an additional, specific indication (e.g. prostate obstruction, prostate calcification).

Other options

- Lifestyle changes: avoid activity that involves vibration or trauma to the perineum (e.g. bike riding, tractor driving, long-distance driving, cut out caffeine, spicy foods, alcohol, avoid constipation).
- Some patients may benefit from treatment by a specialised pelvic floor physiotherapist, which may include pelvic floor relaxation techniques and trigger point massage.
- Prostate massage.
- Supportive therapy: biofeedback, relaxation exercises, acupuncture, massage therapy, chiropractic therapy and meditation.
- Heat therapy.

Referral

Indicators for referral to a urologist

- When the GP is not confident in managing the condition.
- If the GP is concerned there are other potential diagnoses, particularly prostate or bladder cancer.
- Those who do not respond to initial first-line therapy such as antibiotics and/or α -blockers. For these patients, more invasive investigations, such as cystoscopy and transrectal prostate ultrasound scan, are commonly done.

Follow-up

- The need for urologist follow-up depends on the patient's progress.
- Most urologists will refer back to the GP to monitor the progress of the patient.
- The urologist will seek re-referral if the patient's progress is not satisfactory.
- A GP can re-refer if they do not feel comfortable in managing a relapse.

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8

Ejaculatory Disorders



Ejaculatory Disorders

The GP's role

- GPs are typically the first point of contact for men with a disorder of ejaculation.
- The GP's role in management of premature ejaculation (PE) includes diagnosis, treatment and referral.
- Offer brief counselling and education as part of routine management.

How to approach the topic with patients

- "Many men experience sexual difficulties. If you have any difficulties, I am happy to discuss them."

Overview

- Ejaculatory disorders include premature ejaculation, delayed ejaculation, anorgasmia, retrograde ejaculation, anejaculation and painful ejaculation.
- Ejaculatory disorders result from a disrupted mechanism of ejaculation (emission, ejaculation and orgasm).
- Disorders of ejaculation are uncommon, but are important to manage when fertility is an issue.
- Etiologies of ejaculatory dysfunction are numerous and multifactorial, and include psychogenic, congenital, anatomic causes, neurogenic causes, infectious, endocrinological and secondary to medications (antihypertensive, psychiatric (SSRIs), α -blocker).

Premature ejaculation

- The most common ejaculatory disorder¹.
- Ejaculation that occurs sooner than desired.
- Primary (lifelong) PE.
 - Patient has never had control of ejaculation.
 - Disorder of lower set point for ejaculatory control.
 - Unlikely to diagnose an underlying disease.
- Secondary (acquired) PE.
 - Patient was previously able to control ejaculation.
 - Most commonly associated with erectile dysfunction (ED).
- Definition²:
 - An intravaginal ejaculatory latency time (IELT) of less than about 1 minute (lifelong) or about 3 minutes (acquired)
 - An inability to delay ejaculation on nearly all occasions
 - Negative personal consequences such as distress.
- Primary (lifelong) PE is present from the first sexual experience; secondary (acquired) PE represents a significant reduction in latency time occurring later in life, after initially having an acceptable IELT.

Clinical notes

PE is a self reported diagnosis, and can be based on sexual history alone.

Diagnosis

Medical history

Sexual history

- Establish presenting complaint (i.e. linked with ED).
- Intravaginal ejaculatory latency time.
- Onset and duration of PE.
- Previous sexual function.
- History of sexual relationships.
- Perceived degree of ejaculatory control.
- Degree of patient/partner distress.
- Determine if fertility is an issue.

Medical

- General medical history.
- Medications (prescription and non prescription).
- Trauma (urogenital, neurological, surgical).
- Prostatitis or hyperthyroidism (uncommonly associated).

Psychological

- Depression.
- Anxiety.
- Stressors.
- Taboos or beliefs about sex (religious, cultural).

Physical examination

- General examination.
- Genito-urinary: penile and testicular.
 - Rectal examination (if PE occurs with painful ejaculation).
- Neurological assessment of genital area and lower limb.

Refer to [Clinical Summary Guide 1: Step-by-Step Male Genital Examination](#)

Management

Treatment

Treatment decision-making should consider:

- Aetiology
- Patient needs and preferences
- The impact of the disorder on the patient and his partner
- Whether fertility is an issue.

Management of PE is guided by the underlying cause.

Primary PE

- 1st line: SSRI, reducing penile sensation, e.g. using topical penile anaesthetic sprays (only use with a condom).
- 2nd line: Behavioural techniques and/or counselling.
- Most men require ongoing treatment to maintain normal function.

Secondary PE

- Secondary to ED: Manage the primary cause.
- 1st line: Behavioural techniques and/or counselling.
- 2nd line: SSRI, reducing penile sensation and/or PDE5 inhibitors.
- Many men return to normal function following treatment.

Treatment options

Erectile dysfunction (ED) treatment

- If PE is associated with ED, treat the primary cause (e.g. PDE5 inhibitors).

Behavioural techniques

- 'Stop-start' and 'squeeze' techniques, extended foreplay, pre-intercourse masturbation, cognitive distractions, alternate sexual positions, interval sex and increased frequency of sex.
- Techniques are difficult to maintain long-term.

Psychosexual counselling

- Address the issue that has created the anxiety or psychogenic cause.
- Address methods to improve ejaculatory control. Therapy options include meditation/relaxation, hypnotherapy and neuro-biofeedback.

Oral pharmacotherapy

A common side-effect of some selective serotonin reuptake inhibitors (SSRI) and tricyclic antidepressants is delayed ejaculation. SSRIs are commonly prescribed for PE; except for dapoxetine hydrochloride, all other SSRIs are used off-label for treating PE. Common dosing regimens are:

- **Dapoxetine hydrochloride:** A short-acting on-demand SSRI, the only SSRI approved for treatment of PE in Australia; 30 mg taken 1-3 hours before intercourse
- **Fluoxetine hydrochloride:** 20 mg/day
- **Paroxetine hydrochloride:** 20 mg/day. Some patients find 10 mg effective; 40 mg is rarely required. Pre-intercourse dosing regime is generally not effective
- **Sertraline hydrochloride:** 50 mg/day or 100 mg/day is usually effective. 200 mg/day is rarely required. Pre-intercourse dosing regime is generally not effective
- **Clomipramine hydrochloride*:** 25-50 mg/day or 25 mg 4-24 hrs pre-intercourse.

*Suggest 25 mg on a Friday night for a weekend of benefit (long acting).

PDE-5 Inhibitors: e.g. Sildenafil (50-100 mg), 30-60 minutes pre-intercourse if PE is related to ED.

'Start low and titrate slow'. Trial for 3-6 months and then slowly titrate down to cessation. If PE reoccurs, trial drug again. If one drug is not effective, trial another.

Reducing penile sensation

- **Topical applications:** Local anesthetic gels/creams can diminish sensitivity and delay ejaculation. Excess use can be associated with a loss of pleasure, orgasm and erection. Apply 30 minutes prior to intercourse to prevent trans-vaginal absorption. Use a condom if intercourse occurs sooner.
 - **Lignocaine spray:** this should be used with a condom to prevent numbing of partner's genitalia).
 - **Condoms:** Using condoms can diminish sensitivity and delay ejaculation, especially condoms containing anaesthetic.
- Combination treatment can be used.

Referral

For general assessment refer to a specialist (GP, endocrinologist or urologist) who has an interest in sexual medicine.

- Refer to a urologist: If suspicion of lower urinary tract disease.
- Refer to an endocrinologist: If a hormonal problem is diagnosed.
- Refer to counsellor, psychologist, psychiatrist or sexual therapist: For issues of a psychosexual nature.
- Refer to fertility specialist: If fertility is an issue.

Delayed ejaculation

- Delayed ejaculation occurs when an 'abnormal' or 'excessive' amount of stimulation is required to achieve orgasm with ejaculation.
- Often occurs with concomitant illness.
- Associated with ageing.
- Can be associated with idiosyncratic masturbatory style (psychosexual).

Treatment³

- **Aetiological treatment:** Management of underlying condition or concomitant illness (e.g. androgen deficiency).
- **Medication modification:** Consider alternative agent or 'drug holiday' from causal agent.
- Psychosexual counselling.
- Testosterone levels.

Anorgasmia (no orgasm)

- Anorgasmia is the inability to reach orgasm
- Some men experience nocturnal or spontaneous ejaculation
- Aetiology is usually psychological

Treatment⁴

- Psychosexual counselling
- **Medication modification:** Consider alternative agent or 'drug holiday' from causal agent.
- **Pharmacotherapy:** Pheniramine maleate, pseudoephedrine or cyproheptadine may help but have a low success rate.
- Testosterone levels.

Retrograde "dry" ejaculation (orgasm with no ejaculation)

- Retrograde ejaculation occurs when semen passes backwards through the bladder neck into the bladder. Little or no semen is discharged from the penis during ejaculation.
- Causes include prostate surgery and diabetes.
- Patients experience a normal or decreased orgasmic sensation.
- The first urination after sex looks cloudy as semen mixes into urine.

Treatment⁵

- **Counselling:** To normalise the condition.
- **Pharmacotherapy:** Possible restoration of antegrade ejaculation and natural conception; note that pharmacotherapy may not be successful.
 - Imipramine hydrochloride (10 mg, 25 mg tablets) 25-75 mg three times daily.
 - Pheniramine maleate (50 mg tablet) 50 mg every second day.
 - Decongestant medication such as pseudoephedrine; antihistamines such as cyproheptadine.
- **Medication modification:** Consider alternative agent or 'drug holiday' from causal agent.
- **Behavioural techniques:** The patient may also be encouraged to ejaculate when his bladder is full, to increase bladder neck closure.
- **Vibrostimulation, electroejaculation, or sperm recovery from post-ejaculatory urine:** Can be used when other treatments are not effective, to retrieve sperm for assisted reproductive techniques (ART).
- **Post-ejaculatory urinalysis** — presence of sperm and fructose.

Anejaculation

- Anejaculation is the complete absence of ejaculation, due to a failure of semen emission from the prostate and seminal ducts into the urethra.
- Anejaculation is usually associated with normal orgasmic sensation.

Treatment⁵

- Counselling: To normalise the condition.
- Medication modification: Consider alternative agent or 'drug holiday' from causal agent.
- Vibrostimulation or electroejaculation: Used when other treatments are not effective, to retrieve sperm for ART.
- Pharmacotherapy: Dopamine receptor agonists, serotonin antagonists, oxytocin and drugs that increase noradrenaline.
- Testosterone levels.
- Post-ejaculatory urinalysis — absence of sperm and fructose.
- MRI or ultrasound of seminal vesicles and post ejaculatory ducts (usually via the rectum).

Painful ejaculation

- Painful ejaculation is an acquired condition where painful sensations are felt in the perineum or urethra and urethral meatus.
- Multiple causes (e.g. ejaculatory duct obstruction, post-prostatitis, urethritis and autonomic nerve dysfunction).

Investigation

Treatment

- Aetiological treatment (e.g. infections-prostatitis, urethritis): Implement disease specific treatment.
- Behavioural techniques: If no physiological process identified. Use of relaxation techniques (e.g. ejaculation in conditions when muscles can be relaxed), use of fantasy for distraction.
- Psychosexual counselling.
- Transurethral resection of the ejaculatory duct.
- Urine analysis (first pass urine – chlamydia & gonorrhoea urine PCR test; midstream urine MC&S).
- Cultures of semen (MC&S).
- Cystoscopy.
- Consider imaging (MRI and transrectal ultrasound) to assess for ejaculatory duct obstruction.

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9

Erectile Dysfunction



Erectile Dysfunction

The GP's role

- GPs are typically the first point of contact for men with erectile dysfunction.
- The GP's role in the management of erectile dysfunction (ED) includes clinical assessment, treatment including counselling, medication, referral and follow-up as needed.

Overview

- ED is a persistent or recurrent inability to attain and/or maintain a penile erection sufficient for satisfactory sexual activity and intercourse.
- ED is a separate (but occasionally related) clinical condition to premature ejaculation, which has its own distinct management (See Clinical Summary Guide 8: Ejaculatory Disorders).
- ED is a common condition affecting 6-64% of males aged 45-79¹.
- ED is associated with chronic disease including cardiovascular disease and diabetes. Furthermore, ED may be an early warning sign of these chronic diseases².
- ED is a treatable condition that can impact strongly on the well-being of men and their partners.
- The sexual health of older patients is often overlooked.
- Understanding female partners' sexual needs as part of management should be considered.

How to approach the topic with patients

- "Many men (of your age/with your condition) experience sexual difficulties. If you have any difficulties, I am happy to discuss them".
- "It is common for men with diabetes/heart disease/high blood pressure to have erectile problems. Also, erectile problems can indicate you are at higher risk for future health problems such as heart disease. So it's an important issue for us to discuss if it is a problem for you".

Diagnosis

History

Medical	Sexual	Psychosocial
Lifestyle	Define the nature of the sexual dysfunction	Depression
General health	ED onset	Anxiety
Chronic disease	Spontaneous morning erections	Relationship difficulties
Genital disease	Penetration possible	Sexual abuse
Medications	Maintenance of erection	
Pelvic surgery/radiation		

Physical examination

- Genito-urinary: penis (plaques), testes (size).
- Cardiovascular: BP, HR, waist circumference, cardiac examination, carotid bruits.
- Neurological: focused neurological examination (e.g. perheral neuropathy).

Refer to Clinical Summary Guide 1: Step-by-Step Male Genital Examination

Investigations

- Diabetes mellitus.
- Hyperlipidemia.
- Hypogonadism.
- Cardiovascular disease.
- Others as indicated.

Management

Treatment decision-making

- Cause: organic, psychosocial or combined.
- Determine patient and partner goals.
- Benefits, risks and costs of treatment options.

Treatment summary

1st line

- Alter modifiable risk factors and causes.
- Counselling and education.

2nd line

- Oral agents (PDE5 inhibitors).
- Vacuum devices/rings.

3rd line

- Consider specialist referral.
- Intracavernous vasoactive drug injection.

4th line

- Specialist referral.
- Surgical treatment (penile implants).

For full details of treatment, refer over page.

Referral

Indicators for referral

- Level of GP training/experience.
- Patient request.

Refer to endocrinologist

- Complex endocrine disorders.

Refer to urologist

- Pelvic or perineal trauma.
- Penile deformities.
- Patients for penile implants.
- Treatment failure (e.g. poor or non-responders to medication).

Refer to ED specialist (either endocrinologist or urologist)

- Complex problems including vascular, neurological and treatment failures.

Follow-up

Follow-up is essential to ensure the best patient outcomes.

Assess:

- Effectiveness of treatment and patient/partner satisfaction
- Any adverse effects of treatment
- Overall physical and mental health
- Partner's sexual function (e.g. libido) and couple's adaptation to changes to sex life.

Treatment of erectile dysfunction

1st line treatment

Alter modifiable risk factors and causes

- Modify medication regime: change current medications linked to ED (e.g. antidepressants, antihypertensives) when possible.
- Manage androgen deficiency: when diagnosed and a cause is established, androgen replacement therapy.
- Address psychosocial issues: includes relationship difficulties, anxiety, lifestyle changes or stress.

Facilitating sexual health

- Lifestyle changes: Smoking cessation, reduced alcohol, improved diet and exercise, weight loss, stress reduction, illicit drug cessation, compliance with diabetes and cardiovascular medications.

2nd line treatment

Oral agents: PDE5 inhibitors²

- Adapt dose as necessary, according to the response and side-effects.
- Treatment is not considered a failure until full dose is trialled 7-8 times.
- Ensure patient knows that sexual stimulation is required for drug to work.
- Common side-effects: headaches, flushing, dyspepsia, nasal congestion, backache and myalgia.
- Contraindicated in patients who take long and short-acting nitrates, nitrate-containing medications, or recreational nitrates (amyl nitrate or methamphetamines).
- Exercise caution when considering PDE5 inhibitors for patients with active coronary ischaemia, congestive heart failure and borderline low blood pressure, borderline low cardiac volume status, a complicated multi-drug antihypertensive program, and drug therapy that can prolong the half-life of PDE5 inhibitors (consult patient's cardiologist if in doubt).

On demand dosing

- Sildenafil: 25, 50 and 100 mg; recommended starting dose 50 mg (usually need 100 mg); should be taken on an empty stomach.
- Tadalafil: 10 and 20 mg; recommended starting dose 20 mg.
- Vardenafil: 5, 10 and 20 mg; recommended starting dose 10 mg (usually need 20 mg).

Daily dosing

- Tadalafil: 5 mg at the same time every day. The dose may be decreased to 2.5 mg but not exceed 5 mg daily.

As of September 2020 Sildenafil and Tadalafil are off-patent and have generics available, making these more affordable.

Counselling and education

- Offer brief counselling and education to address psychological issues linked with ED, such as relationship difficulties, sexual performance concerns, anxiety and depression.
- Discuss sexual misinformation: Includes importance of sufficient arousal and lubrication, and realistic expectations, such as normal age-related changes.
- Consider concurrent patient/couple counselling with a psychologist, to address more complex issues, and/or to provide support during other treatment trials.

Vacuum devices and rings

- Suitable for men who are not interested in, or have contraindications for oral or injectable pharmacologic therapies.
- Typically suitable for patients in long-term relationships.
- Adverse effects include penile discomfort, numbness and painful ejaculation.

3rd line treatment

- Consider referral or specialist training/supervision.

Intracavernous vasoactive drug injection

- Alprostadil: 10 and 20 µg is the first choice for its high rate of effectiveness and low risk of priapism and cavernosal fibrosis. If erection is not adequate with alprostadil alone, it may be combined with other vasoactive drugs (bimix/trimix) to increase efficacy or reduce side-effects.
- Commence with minimum effective dose and titrate upwards if necessary.
- Initial trial dose should be administered under supervision of an experienced GP or specialist.
- Erection usually appears after 5 to 15 minutes and lasts according to dose injected. Aim for hard erection not to last longer than 30 minutes.
- Recommended maximum usage is 3 times a week, with at least 24 hours between uses.
- Contraindicated in men with history of hypersensitivity to drug or risk of priapism.
- Patient comfort and education are essential. Inform patient of side-effects (priapism, pain, fibrosis and bruising, particularly if on blood-thinning agents). Provide a plan for urgent treatment of priapism if necessary.

4th line treatment

- Refer to urologist (surgical treatments).
- Penile prosthesis (penile implant): A highly successful option⁴ for patients who prefer a permanent solution or have not had success with pharmacologic therapy. Surgery is irreversible and eliminates the normal function of the corpus cavernosa. Cost may be a limiting factor for some patients.
- Vascular surgery: Microvascular arterial bypass and venous ligation surgery to increase arterial inflow and decrease venous outflow may be appropriate for some men⁵ but is not considered a standard approach².

Possible emerging treatments

- Low dose shock wave therapy, topical nitrates and new oral agents are being evaluated and may play a role in treatment of ED in the future.

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10

Klinefelter Syndrome



HEALTHY MALE

Generations of healthy Australian men

Klinefelter Syndrome

The GP's role

- General practitioners should consider Klinefelter syndrome as a possible cause in male patients presenting with any relevant symptoms or signs.
- Most diagnoses of Klinefelter syndrome occur either prenatally, around the expected time of puberty or in association with infertility.
- Accurate measurement of testicular volume (using an orchidometer; Image 1) during routine physical examination would likely improve detection of Klinefelter syndrome, thereby facilitating simple, effective treatment with life-long benefit.
- GPs are ideally placed to perform clinical assessments, order laboratory investigations, refer for specialist assessment and initiation of testosterone replacement therapy, and coordinate ongoing management of Klinefelter syndrome.

Clinical notes

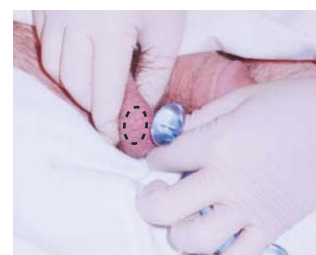
Some features of Klinefelter syndrome are specific to the syndrome (e.g. behavioural and cognitive) and some features relate to the androgen deficiency (e.g. osteoporosis).

Condition overview

- Klinefelter syndrome refers to a collection of characteristics in males caused by the presence of two or more X chromosomes.
- The most common (80–90%) karyotype of males with Klinefelter syndrome is 47,XXY. Some males with Klinefelter syndrome have more than two chromosomes, or chromosomal mosaicism¹.
- Klinefelter syndrome prevalence is estimated to be 1-2 per 1000 men, with only around one quarter ever diagnosed².
- Klinefelter syndrome is characterised by impaired testosterone production and spermatogenesis.
- Klinefelter syndrome is the most common cause of androgen deficiency in men.
- Men with Klinefelter syndrome benefit from testosterone replacement therapy³.
- Infertility is common in men with Klinefelter syndrome, due to oligi- or azoospermia, but paternity may be possible with assisted reproductive technologies using sperm collected by testicular biopsy.
- Classical features of Klinefelter syndrome (Image 2) are present to differing degrees in individuals.
- The only consistent feature of Klinefelter syndrome is small testes volume (< 4 ml)⁴.
- The subtle effects of Klinefelter syndrome in many men accounts for low rates of diagnosis and illustrates the importance of genital examination as part of routine clinical exams.

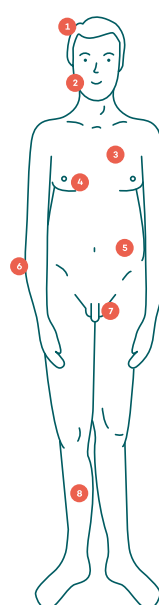


30 mL normal



4 mL Klinefelter syndrome

Image 1 – Example of 30 ml and 4 ml adult testis



- 1 Taller than average height
- 2 Reduced facial hair
- 3 Reduced body hair
- 4 Breast development (Gynecomastia)
- 5 Feminine fat distribution
- 6 Osteoporosis
- 7 Small testes (testicular atrophy)
- 8 Varicose veins

Image 2 – Clinical features of Klinefelter syndrome. Features present may be few, some or all.

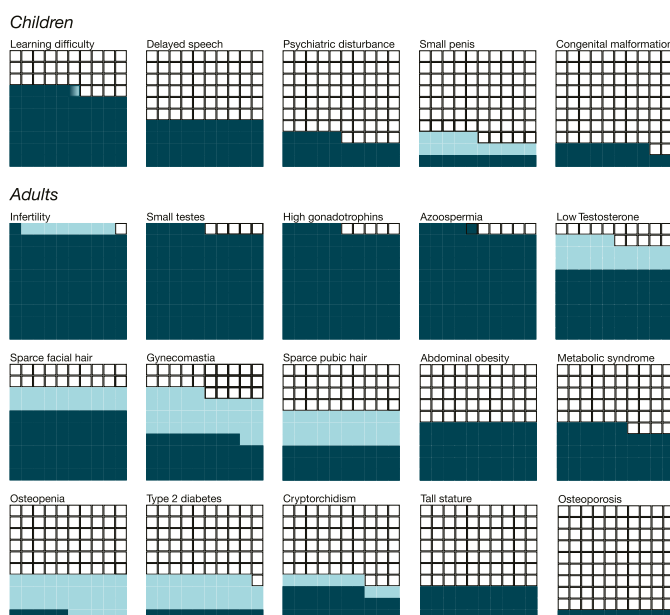


Image 3 – Prevalence of Klinefelter syndrome characteristics

Diagnosis

Medical history

- Pubertal development (poor progression).
- Sexual function (low libido).
- Degree of virilisation.
- Psychosocial (learning, schooling and behaviour).
- Infertility.

Examination

Infancy

- No hormonal features prior to puberty.
- Undescended testes.
- Rarely ambiguous genitalia.

Adolescence

- Small testes (< 4 mL) characteristic from mid puberty.
- Poor pubertal progression and facial, body and pubic hair relative to age.
- Gynecomastia.
- Feminine fat distribution.
- Taller than average height.
- Poor muscle development.

Adult

- Small testes (< 4 mL).
- Reduced facial, body and pubic hair.
- Gynecomastia.
- Feminine fat distribution and weight gain.
- Taller than average height.
- Poor muscle development.

Refer to Clinical Summary Guides 1-3

Testicular Volume

Assessment of testicular volume is essential

- Testicular volume is assessed using an orchidometer.
- Normal testicular volume range:
 - Childhood 3 mL or smaller
 - Puberty 4–14 mL
 - Adulthood 15–35 mL
- Small testes < 4 mL is the only consistent feature of Klinefelter syndrome (image 2).
- The testes may start to develop in early puberty, but soon regress to < 4 mL by mid puberty.

Investigations

- Two morning fasting samples of serum total testosterone, taken on different mornings.
- Total serum testosterone, low or low normal from mid puberty (normal range 8–27 nmol/L).
- Serum LH, elevated from mid puberty (normal range 1–8 IU/L).
- Serum FSH, elevated from mid puberty (normal range 1–8 IU/L).
- Karyotype (47,XXY)
 - 10% mosaic 46,XY/47,XXY.

Other investigations

- Bone density study, DEXA (osteoporosis).
- Semen analysis if fertility is an issue (usually azoospermic).
- TFT (hypothyroidism).
- Fasting blood glucose (diabetes).

T formulation	Usual (starting) dosage	Dosage range
Injections (IM)		
Combined testosterone propionate Testosterone phenylpropionate Testosterone isocaproate Testosterone decanoate* Testosterone enantate*	250 mg every 2 weeks	10 to 21-day intervals
Testosterone undecanoate	1000 mg every 12 weeks following loading dose at 6 weeks (i.e., 0, 6, 18, 30 weeks)	Longer term: 8 to 16-week intervals
Transdermal patch		
Testosterone	5 mg applied nightly	2.5 to 5 mg daily
Transdermal gel		
Testosterone (1% 50 mg in 5 g sachet or pump pack dispenser; applied daily)	50 mg daily	25–100 mg daily
Transdermal cream		
Testosterone	100 mg daily applied to upper body 25 mg daily applied to scrotum	Up to 200 mg daily (to torso) Up to 50 mg daily (to scrotum)
Oral undecanoate		
Testosterone undecanoate	40 mg capsule 160 to 240 mg in 2 to 3 doses daily	80 to 240 mg daily

*Not available on Australian Pharmaceutical Benefits Scheme (PBS).

Management

Testosterone replacement therapy (TRT)

- TRT is life-long and may be started from mid puberty although many boys initially virilise normally.
- Gynecomastia is an indication to start TRT.
- Teenage boys usually start on a low dose and build to full adult dose as puberty progresses.
- Even if measured T levels are normal, there is evidence that bone density is reduced in the presence of chronically raised LH levels, suggesting that TRT is indicated.
- In adults, consult with a fertility specialist (if appropriate) to develop a plan for fertility prior to TRT, as TRT will suppress spermatogenesis.

Other treatments

- Gynecomastia may be transient, lasting one to three years.
- Adequate testosterone replacement often results in complete resolution over 12 months.
- Surgical removal, mastectomy (do not refer for early surgery as it may resolve naturally or following TRT).

Follow-up

Monitoring TRT is essential

Prostate

- Men with Klinefelter syndrome are less likely to die from prostate cancer, and restoring testosterone levels to the normal range is likely only to return their risks to those of their eugonadal peers.
- Subject to the same advice about testing for prostate cancer as their peers (PSA).
- Exclusion of significant prostate pathology is essential for those aged > 40 years at the commencement of therapy.

Raising clinical awareness

Aside from cognitive and behavioural features, it is important to note that despite the following recognised disease associations with Klinefelter syndrome the absolute risk is low.

- Tumours: leukaemia, mediastinal germ cell tumours, lymphoma, teratoma and breast cancer.
- Endocrine: hypothyroidism and diabetes mellitus (Type 1 and 2, rare).
- Cardiovascular: venous ulcers and venous thromboembolic disease.
- Auto-immune: systemic lupus erythematosus (SLE) and coeliac disease.

Referral

Children and adolescents

- Refer to a paediatric endocrinologist.
- Refer for educational and allied health assistance if needed.

Adults

- Develop a plan in consultation with an endocrinologist for:
 - Hormone deficiency
 - Infertility
 - Osteoporosis.
- Refer to a fertility specialist, as appropriate, for sperm recovery from testis (occasionally) or donor sperm.

Infertility

Infertility is a major implication of Klinefelter syndrome¹.

- Most men are azoospermic.
- Sperm are rarely found in the ejaculate but in 30-50% of cases sperm can be found in testicular biopsy tissue.
- Treatment options:
 - Intracytoplasmic Sperm Injection (ICSI) — the risk of 47,XXY offspring is low
 - Donor insemination.
- Counselling may be necessary.

Refer to Clinical Summary Guide 5: Male Infertility

Learning and behaviour difficulties

The general intellectual ability of boys with Klinefelter syndrome is within the normal range. However, boys with Klinefelter syndrome may have:

- Difficulties with speech and reading
- Delayed motor development
- Reduced attention span
- Behavioural problems (particularly in adolescence)
- Educational and allied health assistance may be required.

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11

Engaging Men



HEALTHY MALE

Generations of healthy Australian men

Engaging Men

Men's use of healthcare services

In general, men visit the doctor less often than women have shorter consultations and tend to present later in the course of illness.

Men's health-seeking behaviour should not be misinterpreted as reflecting a lack of men's interest in their own health, or avoidance of healthcare. Most Australian men (95%) consider their health important, but not as many of them engage often or regularly with primary healthcare services¹.

Men monitor their own health, and they pay attention to the onset and progression of symptoms of disease. Their decisions to seek help depend on the nature of their illness, its impact on their lives, the duration of their symptoms and their previous experiences with health care services².

If a man has an illness that he assesses as being serious, which is painful or disfiguring, that prevents him from his usual activities, or for which he thinks treatment will be effective, he is more likely to seek help than for illnesses without any these characteristics.

Where men get health information

The most common sources of health information used by men are family members, the internet or a healthcare professional¹. Younger men prefer to use the internet, and older men more often choose a doctor or nurse as their first source of health information¹.

Men are critical users of health information, preferring reputable sources that are known or recommended to them. They may use online sources of health information to prepare for a visit to the doctor, but generally they do not see the internet as a replacement for a visit to a doctor³.

Community-based organisations, such as Men's Sheds, are a common and effective means of providing health information for many men⁴.

What men value from healthcare professionals

"Men prefer collaborative interventions involving action-oriented problem solving"⁵.

When men seek care, they want a prompt resolution of their problem by cooperating with a clearly competent and empathetic health practitioner, who takes a direct approach to the problem and might use humour to lighten the mood⁶.

Men who have a regular GP are more likely than men without one, to attend routine check-ups and disclose health information to their doctor.

A good relationship with their GP can motivate men to make an appointment to see them, to attend for regular check-ups, and to follow through with treatment and screening tests.

Men appreciate having a doctor who knows their medical history, so that they don't need to revisit it each time they visit³.

What men need from health service providers⁷

Barriers to men's access to healthcare services may be structural (e.g. lack of services) or systemic (e.g. poor communication between men and healthcare professionals).

Structural changes to health services, that create male-friendly settings, cater for the time constraints of many men, and integrate telehealth and other new technologies can improve men's access to healthcare.

Systemic changes that improve practitioners' ability to relate to male patients, and their knowledge of men's health, will likely increase men's use of healthcare services.

Men's willingness to seek healthcare is influenced by past experiences, so enabling positive interactions will facilitate greater use of healthcare services.

The health literacy and information needs of men are varied, so tailored approaches to provision of health information are required to ensure men are adequately informed about what is required of them for disease prevention and resolution.

Men may be reluctant to raise sexual or mental health concerns with their doctor but they are generally welcoming of enquiries about these topics by their doctor, and are forthcoming in providing relevant information⁸.

Characteristics of primary care practices that cater well for men

- Located near to public transport.
- Parking available on-site.
- Gender neutral décor.
- Prominent provision of male-oriented reading material and patient information in waiting areas.
- Display of posters/photographs featuring men in non-stereotypical settings.
- Clear display of available services and practitioners' expertise.
- Clear display of cost of services, including cancellation fees (ideally with an option of bulk billing for patients in financial stress).
- Clear information about processes to make appointments.
- Promotion of the option to choose longer consultation times when making appointments.
- Encouragement of patients to phone prior to appointments, to check on possible delays in consultation times, or provision of a service to notify patients if such a delay occurs.
- Flexibility in appointment times (e.g. provision of early morning, evening or weekend appointments or 'drop-in' clinics).
- Efficient clinic operation (e.g. simple procedures, clear communication, avoidance of redundant processes).
- Provision of male-only clinics.
- Visibility of male staff, especially in reception and other roles with patient contact.
- Use of questionnaires and forms completed by patients for collection of 'sensitive' information.
- Provision of privacy and use of discretion.
- Support and encourage continuing education of staff about men's health and communicating with men.
- Provision of information about local community resources and support services.

Characteristics of practice staff that cater well for men

- Respectfully welcome patients.
- Display empathy and understanding.
- Demonstrate expertise, knowledge and interest in men's health and social issues.
- Have expertise in men's health.
- Use patient-centred communication.
- Use non-deficit approaches to discussing health and wellbeing.
- Thoughtfully use humour.
- Make eye contact.
- Use direct communication.
- Avoid use of jargon and unnecessarily complicated explanations.
- Provide precise and clear instruction, information about next steps and prognosis.
- Encourage health seeking behaviour.
- Use time efficiently but do not rush during patient interactions.
- Ask questions about sexual and mental health, and other potentially sensitive issues.
- Are aware of local community services and facilities to support men's health and wellbeing.

Men's adherence to stereotypical masculine traits, such as stoicism, self-reliance, strength and control can stigmatise, and thereby discourage, healthcare seeking⁹.

There is widespread recognition in Australia that traditional masculine stereotypes are both inaccurate and harmful. Freeing men from these restrictive stereotypes will likely be good for their health and wellbeing, and that of society more generally¹⁰.

Health services should avoid blaming men and making assumptions about their behaviour, and focus on solutions rather than problems.

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12

**Engaging
Aboriginal and
Torres Strait
Islander Men**

Engaging Aboriginal and Torres Strait Islander Men



HEALTHY MALE
Generations of healthy Australian men

Aboriginal and Torres Strait Islander men, as a group, are among the most disadvantaged when it comes to health in our country¹.

Barriers to accessing health services for Aboriginal and Torres Strait Islander men

Some significant issues affect the way Aboriginal and Torres Strait Islander men engage with the health system and access health services. In particular, when seeking health care for multiple issues the pathway from one service to another can be difficult due to any or all of the following factors.

Societal	Illness related stigma. Gender differences in health.
Cultural	Traditional gender-related lores. Masculinity and gender roles. Language and gender barriers. Beliefs around causation.
Logistical	Lack of transport. Appointment times conflict with other family and community priorities (e.g. ceremonies).
Health system	Limited access to specialist service and/or treatment. Complicated referral process. Too few (male) health professionals (leading to patients seeing many different doctors). Medical terminology/jargon.
Financial	Difficulties in meeting health service costs.
Individual	Knowledge/perception of the nature of the illness. Previous illness experience. Low prioritisation of preventative healthcare. Lack of understanding and embarrassment. Self-esteem and confidence.

Strategies for health services to engage men from remote communities

Cultural diversity of Aboriginal and Torres Strait Islander communities means that what may work in one setting may not work in another. The need in remote communities for a male-specific place may not be as strong in urban settings as there may be greater integration and interaction with non-Indigenous people compared to a remote area. Seeking feedback from the community is important.

Where English is often the second language and men may not be familiar with a clinic environment

- Identify, acknowledge and consult with “cultural bosses” and community male elders.
- Gain local knowledge from men’s groups through consultation and documentation of their needs and issues.
- Plan services with input from local men: for men who are employed and cannot regularly attend the clinic during normal (daytime) operating hours, consider having an after-hours clinic or mobile/outreach service to visit men in their workplaces and/or at men’s places.
- Check patient register for the proportion of adult and young males attending.

- Involve male Indigenous Health Practitioners (IHPs) and traditional healers in outreach services in communities, such as 715 Health Checks with the footy team, providing confidentiality is not compromised.

Where remote clinics are available

- Get male IHP/IHW to help organise outreach services in the community as needed, or in appropriate spaces such as men’s areas.
- Have male-orientated clinics to help men feel comfortable. If the environment feels ‘foreign’ then men may be less likely to engage with doctors or be upfront in providing information.
- Where appropriate, plan and involve family members in the consultations so that the family understands the medical treatment and can provide support if needed.
- Provide flexible delivery, such as telemedicine, home visits or after hours services.
- Engage the local community by holding a clinic meet-and-greet or open day so that men can meet the current/new staff. If feasible, arrange a tour of the clinic and its functions, led by the IHP/IHW.
- Develop men’s health promotion resources (such as posters/ brochures) that include artwork or imagery of local men; if specific to men’s business, posters can be placed in public/ council male restrooms.
- Respect Aboriginal and Torres Strait Islander men and their cultural values. For example, it is important for men (and women) to be able to enter and leave a clinic without passing through a shared reception, particularly when they present for more sensitive health concerns.

To ensure health services are engaging men appropriately by identifying and valuing culture

- Undertake an informal community consultation process to better understand the health care service needs of local men, if not already documented by local men/service.
- Learn local protocols from local health workers on how to access/engage men or men’s groups.
- Set up a male-specific clinic in a dedicated men’s space.
- Where possible, have more male health staff (doctors, nurses and Indigenous Health Workers (IHWs)) and men’s counselling programs to help support men, and go out into the community to encourage access to health services.
- Encourage doctors to stay for the long haul to help men feel comfortable and develop trust.
- Minimise waiting times where possible and encourage men to bring a male IHW, a family member or friend to an appointment to help translate.
- Advise the patient in advance of additional costs when being referred onto other health services/clinics.
- Integrate Follow-up Care Plans as part of ongoing care to ensure the patient has followed treatment and understands when they need to come back for results/re-testing.
- Promote Aboriginal and Torres Strait Islander men and women who work in the health service to build empowerment and encourage community members to return to the health service.

Strategies for GPs and other healthcare professionals to engage Aboriginal and Torres Strait Islander men

Most health professionals intuitively communicate well with patients but when speaking with men from different cultural backgrounds, additional strategies may be helpful. For many men, attending a health service can be a negative experience, for example when blood/urine collection is needed. Trying to make the visit a pleasant experience with positive interaction will help ensure that men return and feel sufficiently comfortable to open up about health concerns, particularly those more sensitive and personal issues. Men will often talk to their family and friends when they have had a positive health experience — word-of-mouth is one of the best ways to encourage Aboriginal and Torres Strait Islander men to attend health services.

- Work on developing trust in the relationship — lifestyle behaviour change may only come after a long, trusting relationship has developed between the patient and doctor.
- Involve a male IHW that may help to identify issues before the appointment.
- Check when the man had his last annual health assessment and take the opportunity while he is at the clinic to repeat it if it has been longer than 12-months.
- Reinforce confidentiality and ensure all health discussions are private and not in open spaces such as reception areas.
- Provide simple, clear and accurate explanations of common medical terms and procedures to help reduce a patient's fears and anxieties about his health care: this may include locally developed material using imagery that men can relate to.

Supporting cultural respect with regard to men's health

Adopting a holistic approach to Aboriginal and Torres Strait Islander health is important. This includes not just the physical well-being of an individual but also the social, emotional and cultural well-being of the whole community.

Working in the Aboriginal and Torres Strait Islander health sector can be challenging for doctors/healthcare professionals who have been educated in a Western approach to health service provision.

It is important that non-Indigenous health professionals delivering services to Aboriginal and Torres Strait Islander people undergo cultural competency training.

This provides the basic tools to avoid cultural pitfalls while providing valuable insight into Aboriginal and Torres Strait Islander perceptions. Cultural respect is shown and real progress can be made.

To support cultural competency training, health professionals can also:

- Practice in a service that allows longer consultation times (e.g. half hour consultations) to build relationships and provide useful knowledge
- Stay in the community for the long-haul to develop a cumulative knowledge of people and backgrounds
- Take opportunities when in remote settings to visit the community and learn some of the local language
- Seek advice and learn from the experience of other health professionals and local IHWs.

Issues of cultural respect are particularly important for older Aboriginal men so a considered approach to some subjects (such as sexual health) is needed.

Strategies to talk about sexual health issues with Aboriginal and Torres Strait Islander men

Aboriginal and Torres Strait Islander men can find it hard to open up and discuss personal and sensitive health issues, particularly if they see someone other than their usual doctor. If a man is seen regularly and feels comfortable with the doctor he is more likely to initiate discussion.

For sexual health matters, it is particularly important for health professionals to be aware of cultural protocols around service providers engaging with Aboriginal and Torres Strait Islander men.

- Provide a safe, private, and comfortable environment that supports open and free dialogue.
- Confidentiality is a major concern of Aboriginal and Torres Strait Islander men, particularly when family and community members are working for the health service they may present to. Hence, seek approval from the client facilitated by other allied health service providers including traditional healers and IHPs/IHWs.
- Men may not open up in the first consultation—it may take time to build trust—but balance is also needed to take advantage of opportunistic discussions.
- For the older man, more care should be taken in approaching sensitive issues. However, often when the conversation has started, men are interested in their sexual health.
- Ask about erectile function for men with cardiovascular risk factors. Use simple analogy and resources (such as brochures, flip-charts, DVDs, visual aids) to help explain the links between erectile dysfunction and chronic disease.
- Incorporate questioning into annual health checks such as: “Have you got any sexual difficulties?” or “About half of men with diabetes will have difficulty getting an erection—is that a problem for you?”.
- Sometimes men have erectile problems when taking prescribed medication for other health issues: it is important to explain to men why this may happen and think about other treatment options if erectile problems are a concern.
- Raise the awareness about lifestyles that may impact on erectile dysfunction like smoking and heavy drinking.
- Think about making the consulting room more inclusive for talking about sensitive issues, for example a model or pictures of the male pelvis might help initiate discussion.
- Consider the sensitivity of physical examinations, such as DRE, for men with a past history of being sexually abused.

For the purposes of this guide, IHPs/IHWs provide clinical and primary health care for individuals, families, and community groups.

Healthy Male would like to thank the Aboriginal and Torres Strait Islander Male Health Reference Group for guidance and input into the development of this guide.

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13

Transgender Healthcare



Transgender Healthcare

Background

Most people are assigned a gender — either male or female — at birth (or even before), based on the appearance of their external genitalia. This classification of gender, although conventional, is inconsistent with the lived reality of some people.

An unknown number of people experience conflict between their assigned gender and their experienced gender. The consequences of this conflict may result in specific healthcare needs of these individuals.

Prevalence

It is difficult to estimate the number of transgender people (whose gender identity or expression is different to the gender they were assigned at birth) in our communities because they likely underreport their gender disparity due to concerns about stigma and privacy, and data collection methods are often inadequate (e.g. sex categorised as either male, female or 'other').

Gender dysphoria

Not all people whose gender experience differs from their assigned gender experience gender dysphoria (a diagnostic term used in the DSM-5). Feelings ranging from discomfort to considerable distress are reasonable responses to the conflict between a person's assigned gender and their gender identity.

Gender dysphoria itself can be the cause of psychological problems. The discrimination and abuse faced by transgender people may contribute to the higher rates of mental illness in transgender than cisgender people¹.

Discrimination and abuse of transgender people

Transgender people experience social marginalisation and health inequities².

Discrimination against transgender people, in many forms, can occur when they access healthcare services³, and is a cause of delay or avoidance of them seeking care. Healthcare providers therefore need to ensure an environment and procedures that are inclusive of transgender people.

The health of transgender people

Transgender people have higher rates of risk-taking behaviours (e.g. substance use, unprotected sex) than cisgender individuals, with attendant higher rates of the negative health consequences⁴. Transgender people appear to have higher rates of a variety of chronic diseases than cisgender people⁵ but the cause for this is unknown.

Inclusion of transgender people

Transgender people may seek healthcare for various reasons, ranging from issues that are unrelated to their gender identity, through to a desire to access gender affirming healthcare.

Fear of discrimination is a barrier to transgender people seeking medical care. Healthcare facilities that are welcoming, inclusive and safe for transgender individuals are essential to facilitate their presentation for care and return for follow-up. There are various practice design elements and procedures^{6,7,8} that contribute to establishing a practice that is inclusive of transgender people.

Aboriginal and Torres Strait Islander Gender Diversity

Sistergirl is a term used by Aboriginal and Torres Strait Islander people who have a female spirit and take on female roles within the community, including looking after children and family. Many Sistergirls live a traditional lifestyle and have strong cultural backgrounds.

Brotherboy is a term used by Aboriginal and Torres Strait Islander people to describe gender diverse people who have male spirit and take on male roles within the community. Brotherboys have a strong sense of their cultural identity.

The terms Sistergirl and Brotherboy can differ between locations, countries and nations. The terms may not specifically define who someone is, but instead identify a fluid affiliation that complements their identity.

In Aboriginal and Torres Strait Islander communities the term Sistagirl and Brothaboy are used as terms of endearment, for women and men respectively, with no reference to gender identity.

The healthcare needs of transgender people

Most healthcare required by transgender people, including most gender affirming treatments, does not require specialist medical knowledge. In some cases, the complex healthcare needs of transgender people require multidisciplinary care from general practitioners, mental health professionals, endocrinologists, sexual health physicians, surgeons, speech pathologists and social services, depending on individual circumstances. General practitioners are well placed to manage the healthcare needs of transgender people⁹.

Initiation of gender affirming treatment for patients of inexperienced general practitioners is usually performed by, or in close collaboration with, endocrinologists and mental health professionals.

Affirmation of gender identity

Healthcare providers should not make assumptions about someone's gender identity. Patient information paperwork should include an option for patients to mark their gender as something other than just male or female¹⁰. Simply asking a person's preferred name, pronoun (e.g. he/she/they) and gender identity (on a form or in conversation) is better than guessing.

An important aspect of gender affirmation for some people is consistency with official documents. In Australia, reissue of official federal documents (such as passports) with a person's affirmed gender is possible with the support of a medical practitioner. Different Australian states have their own requirements and procedures for changing the gender on birth certificates and other documents issued under their jurisdiction.

During the initial consultation, it is important to take a complete history, assess risks and identify available social support, and perform any necessary examinations.

Gender incongruence and gender dysphoria are not pathological conditions, but they may be accompanied by mental health issues that require attention. Screening by a psychologist or psychiatrist may be necessary to rule out gender dysphoria as a manifestation of mental health issues (e.g. body dysmorphic disorder) or other conditions (e.g. Asperger syndrome) to ensure appropriate care.

Transgender people have higher rates of suicidal ideation and self-harm than cisgender people¹¹, so appropriate screening, surveillance and referral (if necessary) are important elements of their ongoing healthcare.

Not all transgender people will seek gender affirming medical or surgical intervention but may socially affirm their gender by using behavioural changes such as altering their speech and clothing. Chest binding or genital tucking to hide secondary sexual characteristics¹² can result in bruising, skin irritation and pain, particularly if not performed correctly¹³.

Gender affirmation treatments

Many transgender people seek medical intervention to achieve physical affirmation of their gender identity¹⁴, the most common form being hormonal treatment.

Children

Children who experience gender conflict may benefit from psychological support (for the child and their family), particularly if their family, school or social environments are not supportive. Generally, the mental health of children who socially transition does not seem to be adversely affected¹⁵. For children with complicated circumstances, help from an experienced expert in child cognitive and emotional development will likely be necessary.

Medical treatment for gender affirmation is not recommended until the onset of puberty¹⁵.

Adolescents

Most adolescents with gender dysphoria are likely to need psychological support¹¹. For some people, the development of secondary sexual characteristics during puberty can cause considerable distress¹⁶, and many of these individuals desire interventions to affirm their gender identity¹⁷.

Puberty suppression

In general, gender-affirming hormonal treatments are not used until after 16 years of age.

Gonadotrophin-releasing hormone (GnRH) agonists can suppress androgen and oestrogen levels, hence slowing or preventing the development of secondary sexual characteristics. Such treatment may have some irreversible effects (e.g. consequences for testis development and pubertal increases in bone mineral density are not well understood¹⁸) but can relieve distress and prevent the permanent development of secondary sex characteristics of the natal sex¹⁹. Thus, pubertal suppression is considered an ethically responsible option for transgender adolescents²⁰.

The effects of GnRH agonists for puberty suppression on testis development and function are not known but use of these drugs has irreversible effects in older men. Puberty suppression may result in reduced bone mineralisation, so monitoring of bone mineral density is recommended¹⁵. Otherwise, growth (in terms of height and weight) continues whilst the person decides whether to use hormone therapy and/or surgery for permanent gender affirmation.

The Australian Pharmaceutical Benefits Scheme does not subsidise the use of gonadotrophin-releasing hormone agonists for puberty suppression.

Reproductive counselling

The fertility consequences of gender affirming hormone treatments are not thoroughly understood. Anecdotally, some people cease gender affirming treatments and have no trouble conceiving but others appear infertile. Some gender affirming surgical procedures may result in sterilisation (although these are among the least common interventions accessed by transgender people).

Discussion of fertility with transgender people prior to commencing gender affirming treatment is important for informed care. Information about options for fertility preservation, such as sperm and egg collection and cryopreservation, and assistance with accessing these services is desired by most transgender people seeking gender affirming hormone treatment²¹. However, starting a family after gender-affirming medical and surgical treatments is possible without this. Discussions and procedures relating to fertility preservation may exacerbate gender dysphoria, so these are best undertaken with sensitivity, ideally in a healthcare service with experience in this area²¹.

Hormone treatment for gender affirmation

Many transgender people want hormone treatment to affirm their gender identity²³. Testosterone and oestradiol to achieve masculinisation or feminisation of a person's appearance, respectively, are generally safe, effective and accepted by transgender people.

Pharmaceutical Benefit Scheme subsidy for testosterone requires treatment by, or in consultation with, a specialist in endocrinology, urology or sexual health. This influences the cost, and therefore availability, of testosterone to transgender people.

Testosterone for transgender males is generally administered according to protocols used for hormone replacement therapy in cisgender people. Oestradiol for transgender females is generally used in higher doses than for menopausal hormone therapy. Anti-androgen treatment to suppress testosterone is commonly used for transgender women and progestins might be required for cessation of menses in transgender males. Other medications may be necessary to augment or counteract side-effects of gender affirming hormone therapy (e.g. to treat acne arising from testosterone treatment).

Hormonal treatment for gender affirmation is best initiated in close consultation with an experienced endocrinologist or sexual health specialist, for general practitioners with limited experience.

Guidelines for prescribing hormones for gender affirmation, based on those of the Endocrine Society¹⁶, have been adapted for an Australian context¹³ (see Table 1).

Table 1

Masculinising hormone therapy
<p>Testosterone therapy for gender affirmation may be supported by the Pharmaceutical Benefits Scheme (PBS) for patients treated by, or in consultation with, a paediatrician, endocrinologist, urologist or sexual health specialist.</p> <p>Testosterone formulations available under the PBS, and appropriate doses, are:</p> <ul style="list-style-type: none"> • Testosterone undecanoate 1000 mg, intramuscularly administered 12-weekly (the first two doses 6 weeks apart) • Testosterone 1% (50 mg/5 g) gel sachets, applied transdermally, one sachet daily • Testosterone 1% (12.5 mg/actuation) gel in pump pack, applied transdermally, four actuations daily • Testosterone 5% (50 mg/mL) cream 2 mL, applied transdermally, daily. <p>Other testosterone preparations (testosterone enanthate, mixed testosterone esters) are available on private, non-PBS prescription.</p>
Feminising hormone therapy
<p>Oestradiol and anti-androgen medications are supported by the PBS. A gradual increase in oestradiol, to mimic puberty onset in adolescents, is possible by beginning with low doses that increase every 2-3 months over 2 years.</p> <p>Recommended preparations, and full doses, are:</p> <ul style="list-style-type: none"> • oestradiol or oestradiol valerate 2-6 mg, orally, daily • oestradiol patches 100-150 mg/24 h, transdermally, changed twice weekly • spironolactone 100-200 mg, orally, daily • cyproterone acetate 12.5-50 mg, orally, daily.

The feminising and masculinising effects of hormone treatment for gender affirmation are summarised in Table 2. Detailed information about necessary pre-treatment investigations, side effects, the medical risks, and monitoring of people using gender affirming hormone treatment is available^{13, 22}.

Table 2

The timing of masculinising and feminising effects of hormone therapy^{16, 25}

Testosterone therapy			
Physical effect	Onset	Maximum effect	Reversibility
Skin oiliness; acne	1-6 months	1-2 years	reversible
Cessation of menses	2-6 months		reversible
Vaginal atrophy	3-6 months	1-2 years	reversible
Clitoral enlargement	3-6 months	1-2 years	irreversible
Body fat redistribution	3-6 months	2 years and onwards	variable reversibility
Facial and bodily hair growth	3-6 months	3 years and onwards	irreversible
Deepened voice	3-12 months	1-2 years	irreversible
Increased muscle mass	6-12 months	2 years and onwards	reversible
Male pattern baldness	variable	variable	irreversible
Infertility	variable	variable	variable

Estrogen and anti-androgen therapy			
Physical effect	Onset	Maximum effect	Reversibility
Decreased spontaneous erections	1-3 months	3-6 months	variable
Decreased libido	1-3 months	1-2 years	variable
Cessation of male pattern baldness	1-3 months	1-2 years	reversible
Decreased muscle mass	3-6 months	1-2 years	reversible
Skin softness; decreased oiliness	3-6 months		reversible
Decreased testicular size	3-6 months	2-3 years	variable
Breast growth	3-6 months	2-3 years	irreversible
Body fat redistribution	3-6 months	2 years and onwards	variable reversibility
Reduced facial and bodily hair growth	6-12 months	3 years and onwards	reversible
Decreased sperm production	variable		variable
Erectile dysfunction	variable		variable

Surgical procedures for gender affirmation

About 25% of transgender people in the United States undergo some form of gender affirming surgery, which is more common in transgender males than transgender females²³. Chest surgery is more common than genital surgery for gender affirmation.

At present, there is no public funding for gender affirming surgery in Australia, so the cost is prohibitive for many people.

Gender affirming surgical procedures for transgender males includes chest reconstruction, hysterectomy, salpingo-oophorectomy, vaginectomy, metoidioplasty, phalloplasty, urethoplasty, scrotoplasty, and testicular and erectile prostheses.

Gender affirming surgical procedures for transgender women include breast augmentation, facial feminisation, tracheal chondroplasty, penectomy, orchidectomy, vaginoplasty, labiaplasty and cliteroplasty. Laryngeal surgery for voice alteration may also be performed.

Ongoing healthcare for transgender people

Preventive health approaches for transgender people need to consider their sex characteristics, conventional disease risk factors and gender identity.

Tailoring of disease screening and preventive measures is necessary for transgender patients (Table 3).

Table 3

Alterations to screening guidelines for transgender people		
Screening program	Alteration for transgender men	Alteration for transgender women
Cardiovascular disease	Initiate screening every 5 years from the beginning of hormone treatment	
Osteoporosis	Follow guidelines for birth sex	Use fracture risk assessment to identify age to begin screening
Breast cancer	Follow guidelines for birth sex	Over 50 years and after 5 years of hormone treatment, screen every 2 years
Cervical cancer	Follow guidelines for birth sex	Individualised, based on gender affirming surgical history
Prostate cancer		Follow guidelines for birth sex
Bowel cancer	Initiate screening at 50 years of age	
Gonorrhoea and chlamydia	Follow guidelines for birth sex	Follow guidelines for females if the person has vaginoplasty

Some transgender people may experience distress during discussions about their anatomy. Physical examinations and screening procedures should be performed by healthcare providers who are aware and considerate of each individual's gender identity. Often, writing to other healthcare providers can alleviate patients' concerns and help them to receive appropriate care.

Healthcare providers must bear in mind that, regardless of surgery, a transgender person's individual sex characteristics will dictate some aspects of health and risk of disease, while gender characteristics may influence others²⁴.

Resources

TransHub has a variety of useful resources including a language guide (transhub.org.au/language) and information for clinicians (transhub.org.au/clinicians).

The Australian Professional Association for Trans Health lists healthcare providers who care for transgender, gender-diverse and non-binary people (auspath.org/providers/).

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Clinical and stakeholder review by Prof Robert McLachlan (Healthy Male), Dr Gideon Blecher (Alfred Health), Dr Andrew Smith (GP), Dr Jeff Wilcox (Northside Clinic), Dr Glenn Duns (Men's Health Melbourne), Dr Anna Watts (Ballarat Health Service), Mish Pony (National LGBTI Health Alliance), Teddy Cook (ACON), Lisa Taylor (Sistergirls & Brotherboys Community Group)

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HEALTHY MALE
Generations of healthy Australian men

14

**Androgen
Use, Misuse
and Abuse**

Androgen Use, Misuse and Abuse



HEALTHY MALE
Generations of healthy Australian men

The general practitioner's role

General practitioners have a role in identification of androgen abuse and the management of its cessation, usually under the guidance of an experienced endocrinologist.

Overview

The extent of androgen abuse is unknown, however, it is estimated that lifetime use in the general population is probably 1-5% globally, with prevalence at least 50 times higher in men than women¹. Two percent of Australian secondary school students report using performance and image enhancing drugs (including androgens²).

Androgen abuse often involves massive doses and multiple drugs taken simultaneously that provide a vastly higher level of androgen action than required for physiological replacement in legitimate androgen deficiency¹.

Commonly abused androgenic substances^{1,3}

Substance	Common or brand name	Route
Boldenone undecylenate	Equipose, Parenabol	IM
Clostebol acetate	Steranabol	IM
Chlorodihydrodehydro- methyl-testosterone	CDMT	Oral*
Dihydrotestosterone		Oral*, transdermal, IM
Drostanelone propionate	Dromostanalone, Masteron	IM
Fluoxymesterone	Halotestin, Ultandren	Oral*
Formebolone	Esiclone	Oral*
Mesterolone	Proviron	Oral*
Metandienone	Methandrosteneolone, Dianabol	Oral*
Methenolone acetate	Primabolon	IM
Methyltestosterone	Android, Metandren, Testred	Oral*
19-Nortestosterone	Nandrolone, Deca- Durabolin	IM
Oxandrolone	Anavar	Oral*
Oxymetholone	Anadrol	Oral*
Stanozolol	Winstrol	Oral*
Testosterone	Sustanon, Testo depot, Nebido	IM
Tetrahydrogestinone	THG, The Clear	Oral*
Trenbolone ethanate	Trenabol, Parabolan	IM

* All orally taken androgenic compounds are alkylated and therefore hepatotoxic.

Definitions⁴

Legitimate androgen uses

Testosterone replacement for androgen deficiency due to pathological hypogonadism

- Primary testicular failure (hypergonadotrophic hypogonadism)
 - Klinefelter syndrome and other genetic disorders
 - Testicular trauma, torsion, infection or excision (orchidectomy)
 - Testis atrophy
- Secondary testicular failure (hypogonadotropic hypogonadism)
 - Congenital cause (e.g., Kallman's syndrome and variants)
 - Acquired (prolactinoma, pituitary disease, surgery, radiotherapy)
- Gender affirming treatment of transgender and gender diverse people⁵
- Delayed puberty

Pharmacological use for non-androgen deficiency

(Typically using synthetic androgen receptor modulators rather than testosterone).

- Osteoporosis (including corticosteroid-induced bone loss)
- Anaemia (due to bone marrow or renal failure)
- Advanced breast cancer
- Cachexia or wasting
- Burns
- Hereditary angioedema

Research

- Functional low testosterone, chronic disease and ill health
- Male contraception

Androgen misuse

Use without valid medical indication or despite contraindication

- Male infertility
- Sexual dysfunction (in absence of androgen deficiency due to pathological hypogonadism)
- "Low T (testosterone)", "late-onset hypogonadism", "male menopause", "andropause", functional hypogonadism
- Non-specific symptoms (e.g., lethargy, tiredness)
- Aging

Androgen abuse

Use without medical indication

- Physical performance (e.g., increased strength, endurance, recovery)
- Physical appearance (e.g., increased muscle mass)
- Occupational (e.g., security staff, police, armed forces)

Compounds commonly used in combination with androgens³

Androgen abuse often involves administration of multiple substances ("stacking") and various dosing regimens. Cycles of use, lasting for several weeks or months (interspersed with periods of abstinence, for "recovery") are usual, with an entrenched but incorrect belief that it is possible to maximise anabolic effect while minimising androgenic impact³. This is biologically implausible given the action of androgens through a single type of androgen receptor.

The below list of medications, commonly used in combination with androgens, is for information only, not as an endorsement of clinical use. At least one of the following applies to these drugs:

- There is no evidence to support use in the context of androgen misuse or abuse or for performance or image enhancement
- Usage is 'off label'
- Safety is uncertain
- Rationale is based on poor understanding of complex endocrinological mechanisms.

Substance	Details
Clenbuterol	A veterinary β 2-adrenergic receptor agonist, used to increase muscle mass and reduce body fat
Ephedrine, Synepherine	Used to reduce body fat
Growth hormone	Used for its anabolic and lipolytic properties, and its potential to reduce muscle and tendon rupture
Growth hormone releasing hormones/peptides (e.g. Modified GRF(1–29), Sermorelin, Ipamorelin, GHRP-2), HGH analogues (e.g. HGH Fragment 176–191)	Falsely claimed to increase muscle mass and do not increase GH levels
Human chorionic gonadotropin (hCG)	Used to promote testicular testosterone production
Recombinant human luteinising hormone (rLH)	May be promoted for stimulation of the testes but it is difficult to manufacture and exceedingly expensive, so marketed preparations are likely to be fake
Insulin-like Growth Factor-1 (IGF-1)	Used to increase muscle mass
Clomiphene citrate and aromatase inhibitors	Used to prevent gynaecomastia, promote testes growth and/or hasten recovery of the suppressed HPT axis
Insulin	Used (often in combination with growth hormone) to increase glucose uptake by muscle and adipose tissue, and inhibit protein breakdown
Prohormones (e.g., androstenedione, dihydroepiandrosterone)	Used as substrate for androgen production
Thyroid hormone (synthetic T3, T4 or natural extracts)	Used to increase metabolism, thereby reducing body fat
Prescription drugs (e.g., sedatives, analgesics, anti-inflammatories, PDE5 inhibitors, antidepressants, diuretics)	Used to treat unwanted side effects of androgen abuse
Illicit drugs (amphetamine, cocaine, cannabis, heroin)	May be used to lose or maintain weight, relax, or relieve pain.

Antiandrogens (clomiphene, tamoxifen) and aromatase inhibitors (letrozole, anastrozole, exemestane) are used in an attempt to reduce the risk or extent of androgen-induced gynaecomastia. This is often unsuccessful, and these drugs have inherent adverse effects such as bone loss and increased thromboembolic risk.

These drugs, together with injectable hCG, are also used as "post-cycle therapy" in an attempt to reverse the androgen-induced suppression of sperm and testosterone production. However, there is no evidence these regimens restart the reproductive system.

Sources of androgens and other substances

The Internet is a common source of androgens and other drugs used in combination, but there is concern about the actual content, purity and safety of any agents obtained in this way⁶.

People who misuse or abuse androgens may attempt to obtain prescriptions from doctors⁷, whose compliance would constitute professional misconduct.

Dietary supplements

Contamination of protein supplements with biologically active androgens^{8,9,10} can result in an identical clinical and biochemical picture to androgen abuse. Close examination of products acquired through gyms or the Internet is required.

The Australian Institute of Sport classifies dietary supplements according to scientific and practical considerations related to safety, efficacy and permissibility, and provides guidance for choosing safe products¹¹.

Patients should be advised to eat healthy food rather than use supplements. Supplements do not contain all the nutrients contained in whole foods and cannot compensate for a poor diet¹².

Diagnosis of androgen abuse

An effective means of knowing if a patient is abusing androgens is to ask them¹, after establishing a trusting, non-judgemental relationship. For patients suspected of androgen abuse, or who seek treatment for androgen abuse or its side effects, an appropriate medical history, physical examination and laboratory tests should be undertaken.

Typically, serum LH, FSH and SHBG will be suppressed. Serum testosterone levels will vary. High levels will be obtained if testosterone administration is recent. Low levels will occur if synthetic androgens (which are not detected by testosterone immunoassays) are used, or during the withdrawal phase when hypogonadotropic hypogonadism is induced.

Medical history

Social factors

- Education level
- Profession or occupation
- Sports and recreational activities (gym or other athletic activities, professional training)
- Marital and fertility status
- Support network
- Alcohol and other drug use
- Other types of abuse

Medical and psychological factors

- Puberty
- Prior reproductive pathology
- Presenting complaint(s)
- Psychiatric health
- Sleep
- Sexual function and reproductive health
- Current diseases
- Current medications
- Current dietary supplement use
- Allergies/hypersensitivity
- Family history (especially prostate and cardiovascular disease)

Physical examination

- General appearance (height, weight, body mass index, muscularity)
- Testicular size and consistency, or cliteromegaly
- Skin (acne, stretch marks)
- Chest (gynaecomastia)
- Hair (premature baldness, body hair)
- Abdomen (hepatic enlargement)
- Heart and lungs (pulse, blood pressure, signs of heart failure, cardiac murmurs)
- Neurological
- Musculoskeletal

Investigations

Laboratory tests

- Reproductive hormones (testosterone, sex hormone binding globulin, luteinising hormone, follicle stimulating hormone)
- Other hormones, when considering hypothalamo-pituitary-adrenal function (insulin-like growth factor-1, thyroid stimulating hormone, T3, T4, prolactin)
- Liver function (alanine aminotransferase, aspartate aminotransferase, alkaline phosphatase, gamma-glutamyl transferase, bilirubin)
- Routine biochemistry and haematology (renal function, glucose, full blood count, lipids)

Imaging

- Liver scan, MRI or CT scan – rarely required unless suspicion of hepatic dysfunction (fatty liver, peliosis or tumor)

Physical signs of androgen abuse

- Rapid and significant weight gain (approx. 10 kg in 2-3 months)
- Muscular physique (disproportionate muscle growth around chest, neck and shoulders)
- Preoccupation with physical fitness, diet or gym attendance dominating lifestyle
- Severe acne (mainly on back, shoulders and chest)
- Stretch marks (usually between biceps and pectoral muscles, possibly back and thighs)
- Excess body hair and/or accelerated baldness
- Hypertension
- Injection site swelling, tenderness, redness
- Tendon and muscle tears
- Abnormal blood lipids
- Abnormal liver function

Male-specific physical signs of androgen abuse

- Gynaecomastia
- Erectile dysfunction
- Subfertility
- Testicular atrophy

Female-specific physical signs of androgen abuse

- Deeper voice
- Increased growth of facial and body hair
- Abnormal menstruation
- Clitoral enlargement

Psychological signs of androgen abuse

- Aggression
- Impulsivity
- Altered libido
- Anxiety
- Dysphoria
- Depression
- Empathy disorder
- Jealousy
- Muscle dysmorphia
- Mood instability
- Panic attack
- Psychosis
- Reduced mentalising capacity
- Sleep disorders
- Suicidal ideation
- Suicide attempt
- Violence
- Worry

Adverse psychological effects of androgen abuse

- Irritability
- Nervousness
- Aggressiveness
- Recklessness
- Self-aggressiveness
- Dependence
- Withdrawal syndrome
- Depression
- Suicidal ideation

Adverse physical effects of androgen abuse

Haematological system

- Polycythaemia

Cardiovascular system

- Dyslipidaemia
- Coronary heart disease
- Myocardial infarction
- Hypertension
- Abnormal ECG
- Arrhythmia
- Left ventricular hypertrophy
- Hypertrophic cardiomyopathy
- Dilated cardiomyopathy
- Heart failure
- Sudden cardiac failure

Hepatic system

- Cholestasis
- Hyperbilirubinaemia
- Steatosis
- Peliosis
- Adenomas
- Hepatocellular carcinoma
- Liver coma

Integumentary system

- Acne
- Striae distensae
- Profuse sweating
- Alopecia
- Hirsutism

Renal system

- High creatinine
- High cystatin C
- Glomerulosclerosis
- Cholemic nephrosis (bile cast nephropathy)
- Renal failure

Reproductive system

Male

- Decreased testis volume
- Impaired spermatogenesis
- Infertility
- Low libido
- Erectile dysfunction
- Gynaecomastia
- Hypogonadism

Female

- Anovulation
- Amenorrhoea
- Dysmenorrhoea
- Infertility

Management of androgen abuse

There are no clinical trials of managing patients from the public who abuse androgens¹³, so information to guide practice¹⁴ is limited to that from observational studies^{15, 16, 17}.

Treatment usually requires a multidisciplinary approach involving the combined efforts of the general practitioner, endocrinologist and psychologist. A formal care plan could facilitate this approach and would also facilitate patient education for long-term behaviour change. The focus of care should be for the expeditious and permanent cessation of this dangerous lifestyle choice.

A supportive approach that is analogous to that used for other licit and illicit social drugs (e.g., alcohol, caffeine, cocaine) and behavioural disorders (e.g. anorexia nervosa, muscle dysmorphia) is the best option. Encouragement and advice that reproductive function will recover with time, and that patience is necessary, may be helpful.

Referral for psychological assessment and possible therapy may benefit patients who abuse androgens because of body image issues¹⁸.

Patients who are unwilling to cease androgen abuse should be advised of the adverse effects of continued use, focussing on the potential consequences for their fertility and long-term cardiovascular health. They should be counselled about the health risks associated with the use of potentially unknown substances and self-injecting.

Medical practitioners should not allow themselves to become resources for ongoing drug supply, including medications used in this subculture (e.g., antiestrogens, hCG), or to facilitate continuation of androgen abuse. The development of a trusting clinician-patient relationship may facilitate discontinuation of androgen abuse. A care plan should be established, aimed at cessation, ideally performed by a supportive team with endocrine and psychological expertise.

Discontinuing androgen abuse

People who abuse androgens develop psychological and physical dependence. Withdrawal symptoms from discontinuation of androgen abuse include decreased sexual drive and a flu-like syndrome consisting of fatigue, headache, musculoskeletal pain and insomnia, followed by depression¹⁹. These features are like those from withdrawal from caffeine or benzodiazepines, less severe than those from withdrawal from nicotine, and without the potential fatality of withdrawal from alcohol, amphetamines or opiates.

There is no legal pathway for the prescription of testosterone or synthetic androgens without a legitimate medical indication.

Fertility recovery

Many men who abuse androgens seek assistance with discontinuation because of infertility. Cessation of androgen abuse often allows recovery of fertility within 6-18 months^{20, 21}. Recovery of endogenous gonadotrophins, testosterone and spermatogenesis, may take many months with the duration of recovery depending mainly on the time since cessation. The outlook for recovery of fertility and testosterone production is generally very good but prolonged.

Ad hoc treatment with anti-estrogens, aromatase inhibitors or hCG lacks any sound evidence basis for safety and efficacy. If there is claimed urgency for restoration of spermatogenesis (e.g. restoration of fertility for a man with a female partner of advanced age), consultation with an experienced endocrinologist is essential because potential treatment may be subject to restrictions based on 'off label' uses and/or uncertain safety considerations.

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This resource was produced in response to requests from Australian General Practitioners, for information to help them respond to androgen abuse. Healthy Male supports GPs in their goal of helping patients to cease this damaging behaviour.

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Clinical and stakeholder review by Prof David Handelsman (Director, ANZAC Research Institute NSW); Prof Gary Wittert (Director, SA division, Freemasons Centre for Male Health & Wellbeing, NSW); Dr Rowan Flanagan; Healthy Male General Practitioners and Primary Health Care Nurse Reference Groups.

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15

**Body
Dysmorphic
Disorder**

Body Dysmorphic Disorder



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The GP's role

GPs have a role in identifying body dysmorphic disorder (BDD) in patients, organising referrals to psychologists or other medical specialists and assisting patients to understand and manage the disorder.

Overview

- BDD is characterised by a debilitating preoccupation with a perceived defect in one's own physical appearance. The physical attribute of concern may be non-existent or so minor as to be unnoticeable by others. Behavioural characteristics of BDD — such as constantly checking one's appearance, repeated attempts at correcting the perceived defect, or excessive exercising — can limit daily function.
- Muscle dysmorphia is a form of BDD characterised by a perceived lack of muscularity. Excessive exercise and specific dietary patterns are common behavioural consequences of muscle dysmorphia¹. Misuse or abuse of androgenic steroids appears commonly in males with muscle dysmorphia².
- Males are more likely than females to have genital manifestations of BDD³.
- BDD is distinct from gender dysphoria, although both may occur in individuals.

Prevalence

- Approximately 1 in 50 (2%) adults have BDD. The disorder most commonly manifests in adolescence, with subclinical symptoms occurring for years before diagnosis⁴. There is no gender difference in the prevalence of BDD⁵ but muscle dysmorphia occurs much more often in males⁶.

Etiology

- BDD is likely due to genetic, psychosocial and cultural factors⁷.
- BDD is more likely than usual to occur in people who:
 - Have a first-degree relative with BDD
 - Experience childhood trauma
 - Are from sexual or racial minority groups.

Use of social media, especially image sharing services (e.g. Snapchat, Instagram), is associated with concern about body image⁸ but there is no high-quality evidence linking social media use and formal diagnosis of BDD.

Comorbidities

- BDD is a chronic condition. It can persist throughout adulthood and its influence on adolescent social and emotional development may have long-term functional consequences⁵. However, treatment can lessen the symptom severity and the negative functional impact of BDD³.
- In males, BDD often accompanies depression, social and generalised anxiety, emotional and behavioural difficulties, problems with peer relationships, hyperactivity, drive for masculinity and a reduced quality of life⁵. Occurrence of these common comorbidities during adolescence can have lasting harmful effects on social functioning, romantic relationships and educational and vocational achievements⁵.

- People with BDD are more likely to have suicidal thoughts or behaviours than people without the disorder, with increasing severity and presence of comorbidities related to increasing risk⁹.

Screening and diagnosis

- People with BDD may lack insight into their disorder, making them unlikely to seek direct help for the condition⁴. Some behaviours that accompany muscle dysmorphia, such as adherence to exercise routines and avoidance of unhealthy foods, can be misinterpreted as beneficial and positively reinforced.
- Only a minority of people with BDD are diagnosed¹⁰.

Common repetitive behaviours of people with BDD

- Grooming.
- Picking at skin.
- Checking appearance in mirror.
- Mirror avoidance.
- Seeking reassurance.
- Camouflaging (covering perceived defect with makeup, hair and/or clothing).
- Touching perceived defect.
- Excessive exercise.
- Comparing appearance with others.
- Seeking correction of perceived defect (e.g. cosmetic surgery).

Useful screening questions for BDD

- Are you concerned about some aspect of your appearance?
- Do you spend a lot of time worrying about some aspect of your appearance?
- Do you spend a lot of time trying to hide physical defects?
- Do you think you are malformed, misshapen or disfigured in some way?
- Do you think your body functions in an offensive way (e.g. bad body odour, flatulence, sweating)?
- Has anyone told you that you look normal, even though you know something is wrong about your appearance?
- Have you felt like you need to see a medical specialist (e.g. cosmetic surgeon, dermatologist) to correct a problem with your appearance?
- Do you feel the need to change your appearance in photos by using apps (like Facetune) or filters (like in Snapchat)?

Diagnostic criteria for BDD³

- Preoccupation with one or more perceived defects or flaws in physical appearance that are not observable or appear slight to others.
- Performance of repetitive physical or mental behaviours (e.g. skin picking, comparing one's physical appearance with others) in response to appearance concerns.
- Significant distress or impairment in important functions (e.g. social, occupational).
- Preoccupation with appearance is not better explained by concerns about body fat or weight in the presence of an eating disorder.
- Body dysmorphic disorder occurs with muscle dysmorphia if there is a belief of insufficient muscularity or small build (with or without preoccupations about other body regions)³.

- Insight into BDD³ is considered absent for people who are convinced their body dysmorphia beliefs are true, or poor for people who think the beliefs might be true. Good or fair insight into BDD is attributed to people who consider their body dysmorphic beliefs to be definitely or probably false, or may or may not be true.

Treatment

- Psychological and psychopharmacological treatment of BDD can moderate symptoms and improve functionality, but only a minority of people with the disorder receive therapy¹⁰.
- Barriers to treatment include shame and stigma, a perception that psychological and psychiatric treatments are ineffective and denial of the disorder (and, hence, failure to seek treatment)¹⁰.
- Many people with BDD seek cosmetic treatments to fix the perceived physical defect, but such procedures generally have poor outcomes¹¹ and should be discouraged⁴.
- People with BDD should be counselled about the likely futility of pursuing cosmetic outcomes and the associated distress and cost that can arise⁴. People with BDD who seek referral for cosmetic procedures would likely be better served by discussion aimed at providing an understanding of the underlying psychological problem and highlighting the benefit of appropriate treatment.
- Cognitive behavioural therapy for BDD commonly consists of exposure with response prevention, over a period of 3–6 months and seems effective at reducing symptom severity for some time; however, longer-term monitoring is recommended to detect symptom severity and relapse¹². Telehealth and internet-based therapy show promise in treating BDD¹³.
- Selective serotonin reuptake inhibitors (SSRIs) may take weeks to months to be effective for BDD and are usually needed in a higher dose than for treatment of depression⁴. In cases where SSRIs are ineffective, the tricyclic antidepressant clomipramine may be used, or alternatively, off-label use of some antipsychotics may be considered⁴.

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Clinical review by Dr Scott Griffiths (University of Melbourne); Healthy Male Nursing and Allied Health Advisory Group; Healthy Male General Practice Advisory Group.

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