GP REFERRAL GUIDE

hereditary connective tissue disorder *diagnostic pathway.*

A reference for general practitioners referring patients into *biio.*'s diagnostic pathway for hypermobile EDS, HSD, and the broader family of hereditary connective tissue disorders. This guide enables you to spare your patients *years of unnecessary suffering.*

AUDIENCE	CONDITIONS	ELIGIBLE AGES	ENDORSEMENT
general practitioners	**hEDS · HSD** · *hereditary CTD*	**paediatric from** *8* · **diagnostic** *16+*	**EDS Society** *Centre of Excellence*

why this *pathway exists.*

01 — the gap

For thirty years, hypermobile Ehlers-Danlos syndrome was cited at a prevalence of 1 in 5,000. The current best evidence — *twenty-seven years* of GP and hospital records — puts it at **1 in 500.** A tenfold revision. The pattern is structural: the conditions cluster — hypermobility, dysautonomia, mast cell activation, post-viral fatigue — but the system that assesses them does not.

15–22

years — published diagnostic delay for hEDS.

15.6

clinicians seen on average; 24% see 20 or more.

94%

of hEDS patients have been told their symptoms were psychological.

302

rheumatologist shortfall in Australia — public waits often exceed 5 years.

The result: referral rejection, medical trauma, unnecessary cost, and poor outcomes. **70 percent** of patients in a 2025 chart review received *inappropriate treatment* as a consequence of misdiagnosis.

biio. has built a structured diagnostic pathway that compresses the *15 to 22 year delay* to weeks — **achieving diagnostic clarity 99 percent faster than Australian averages.**

99%

FASTER *THAN AVERAGE*

GLOBAL ENDORSEMENT

biio. is formally endorsed by the **EDS Society** as one of only *37 CORE centres of excellence* in the world.

what *biio.* changes.

02 — usual vs. biio. pathway

The usual route to a hereditary connective tissue diagnosis is fragmented across disconnected clinicians, with patients carrying their own history between appointments. *biio.* compresses that into a **single coordinated pathway** — with diagnostic clarity in *weeks, not years.*

USUAL PATHWAY	• BIIO. PATHWAY
Patients move between multiple disconnected clinicians.	One **structured interdisciplinary pathway**.
Diagnosis often takes 15–22 years.	Pathway completed in *6–10 weeks*.
Physical signs may be assessed inconsistently.	**EDS-informed**, physio-led physical assessment.
Medical history is repeated across appointments.	A dedicated **care coordinator** holds the complete picture.
Genetics is often delayed or unclear.	Genetic testing **in-house**, used *selectively*.
Specialist access depends on location.	**Telehealth access** Australia-wide.
Remote assessment is limited.	Custom *digital tools* support structured telehealth assessment.

built for *national access.*

03 — telehealth tooling

Distance is one of the largest barriers to a hereditary CTD diagnosis in Australia. *biio.* uses **purpose-built digital tools** to make telehealth assessment as consistent and complete as an in-person workup — wherever your patient lives.

TOOL · 01

structured *pre-intake* screening.

Validated instruments and red-flag screens completed before the first consult.

TOOL · 02

digital *skin* assessment.

Guided capture of skin texture, elasticity, and scarring — reviewed by our clinical team.

TOOL · 03

video-supported *hypermobility* measurement.

Standardised Beighton-equivalent capture, reviewed asynchronously by our physiotherapists.

TOOL · 04

standardised *symptom* capture.

A consistent record of multi-system symptoms — comparable across clinicians and over time.

who to *refer.*

04 — eligibility

REFERRAL · DIAGNOSTIC

patients with hypermobility or a *suspected hereditary CTD.*

Features such as **recurrent joint instability**, chronic pain, dysautonomia or POTS, MCAS, post-viral fatigue, or atypical presentations that have not resolved through standard pathways.

REFERRAL · MANAGEMENT

already-diagnosed patients seeking *connective-tissue-informed care.*

If your patient has a confirmed diagnosis of **hEDS, HSD, or another HCTD**, refer directly. The investigations described in this guide are not required — *the diagnostic workup is already done.*

age *criteria.*

05 — eligible ages

ADULT PATHWAY

16 years and older

Full diagnostic pathway and management capacity.

PAEDIATRIC PATHWAY

children from age *8*

Medical and allied health management, and family education. Formal diagnosis deferred until fully grown.

the *pathway.*

06 — five phases

Five phases, each with explicit gate criteria. Your patient keeps their relationship with you — we hold the diagnostic workup and return a structured report of findings when the pathway concludes.

PHASE 01.

pre-intake *screening.*

Validated instruments, structured red-flag screening, and personal and family history.

WAIT TIME
on referral

PHASE 02.

physical *assessment.*

A **90 to 120 minute EDS-informed physiotherapy assessment**, running in parallel with medical investigations — coordinated by you, or by our nurse practitioners where more convenient.

CURRENT WAIT
2 weeks

PHASE 03.

genetic counselling *and testing.*

Applied selectively, with explicit triggers. Conducted internally by genetic counsellors focused entirely on hereditary CTD.

CURRENT WAIT
2 weeks appt · *4–6 weeks* testing

PHASE 04.

diagnostic *consultation.*

Rheumatologists review a **complete dataset**, exclude differentials, and provide formal diagnosis. Wait time below assumes all pre-screening and investigations are completed — fast-track instructions follow on the next page.

CURRENT WAIT
4-8 weeks

PHASE
05.

documentation *and care planning.*

A **standardised report** designed to hold across NDIS, insurance, and disability systems *without rewriting.*

DELIVERABLE
to referring GP

how to *fast-track* your patient.

07 — expedited pathway

A diagnostic referral on its own enters the pathway. A referral with the **three items below** is *expedited* — your patient walks straight into the assessment phase, and wait time is minimal.

STEP · 01

referral *letter.*

Generate the referral from your usual EHR, addressed explicitly to **'Rheumatologist, biio.'** stating rheumatology. This starts the process.

STEP · 02

pathology — *differential exclusion.*

Order the panel below. This clears the differentials we screen against, so the diagnostic consult is *not held waiting on labs.* Please ask the lab to **copy results to biio.** — your patient may also upload them directly to the patient portal.

STANDARD PANEL

- FBC *with differential*
- Liver function tests
- ESR and CRP
- TSH *(reflex to T4 / T3)*
- Iron studies
- B12, folate, homocysteine
- ANA — *if positive, dsDNA and ENA*
- RF, anti-CCP, HLA-B27

FURTHER — IF INDICATED

- Von Willebrand panel, general coags, vitamin C — *if bleeding history*
- Baseline serum tryptase — *if MCAS suspected*
- Coeliac antibodies — *if GI symptoms or comorbid autoimmunity*

STEP · 03

echocardiogram — *safety and context.*

A standard **transthoracic echocardiogram** serves two purposes. Send the echo report as soon as it's complete — *with or after the referral.*

PURPOSE · 01

aortic root *assessment.*

Aortic dilatation is a recognised feature of hEDS and **an important data point** at the diagnostic consult.

PURPOSE · 02

rare subtype *safety screen.*

Vascular EDS, Marfan syndrome, and Loeys-Dietz syndrome each carry **cardiovascular risks** that materially change management. Early recognition is a safety step.

what happens *after you refer.*

08 — timeline

AFTER YOUR REFERRAL	four checkpoints — *from intake to ongoing care*
WITHIN TWO WORKING DAYS	We'll **contact your patient** and schedule their initial assessment.
OVER THE FOLLOWING WEEKS	Your patient moves through the **five phases** of the pathway.
AT THE *end*	You receive a **formal report of findings** — diagnosis, criteria met, clinical rationale, and onward care recommendations. The report is designed to hold across healthcare, NDIS, insurance, and disability systems *without rewriting.*
THEN — *ongoing*	The patient **transitions back to you** for primary care. Our allied health and medical team remains available for ongoing management — physiotherapy, exercise physiology, psychology, occupational therapy, counselling, orofacial medicine, and cardiac nurse practitioners for POTS management.

to refer a *patient.*

09 — submission

Complete the **referral form** on the following page, *or* create a referral letter in your practice management software. Attach investigations where applicable.

SEND TO

attention *Rheumatologist, biio.*

referrals@biio.com.au