BARE YOUR RARE · PATIENT GUIDE

Fechtner Syndrome
& MYH9-Related Disease

A comprehensive reference for patients, families, and the clinicians who care for them — drawn from the world's leading MYH9-RD registries and written in plain language.

Version 1.0 · April 2026
Verified against GeneReviews (Pecci & Savoia, 2021),
Orphanet, MedlinePlus, and 2024–2026 peer-reviewed literature.

Produced by **Bare Your Rare** · bareyourrare.org
A patient-led rare disease platform · Founded by Thomas Cheesman, HCS patient

Contents

This guide is written for you. For the patient trying to understand a condition most doctors have never heard of, for the family member trying to help, and for the nurse or physician providing direct care. It is designed to be printed, marked up, and brought to appointments.

SECTION 01

What is Fechtner Syndrome?

Fechtner syndrome is a rare inherited condition caused by a change in a single gene called **MYH9**. It is passed down through families in what doctors call an "autosomal dominant" pattern — meaning you only need to inherit the changed gene from one parent to be affected.

> **WORTH KNOWING**
>
> Approximately **30–35% of people with this condition are the first in their family to have it.** The gene change happened new, just in them. A negative family history does not rule out MYH9-RD.

Today, Fechtner syndrome is understood to be one presentation of a broader condition called **MYH9-Related Disease (MYH9-RD)**. Think of it as a spectrum — like different shades of the same color — ranging from mild to more serious. Fechtner syndrome represents the end of the spectrum where the most organ systems are involved: blood, kidneys, hearing, eyes, and sometimes liver.

The estimated prevalence is approximately **1 in every 20,000 to 25,000 people** worldwide. Because it is so often mistaken for other conditions, the true number of people living with it is almost certainly higher than the records show.

SECTION 02

Symptoms — What to Watch For

Not every person gets every symptom. Some people live their whole lives with only the blood abnormality. Others develop kidney, hearing, or eye problems over time. The same gene change can affect people very differently, even within the same family.

Blood (always present from birth)

The body produces too few platelets, and the ones it makes are abnormally large — sometimes as big as a red blood cell. White blood cells also carry tiny spots inside them called *Döhle-like inclusion bodies*. This is actually a valuable clue for diagnosis.

What you might notice: Easy bruising, nosebleeds that take longer to stop than they should, heavier-than-usual periods in women, or longer bleeding after cuts or dental work. Life-threatening bleeding is rare in everyday life, but surgical procedures and deliveries require careful advance planning.

> **A RARE PRESENTATION**
>
> A 2025 case report confirmed that MYH9-RD can occasionally present with a **normal platelet count**. If inclusion bodies are seen on the smear or hearing loss develops, MYH9 testing should still be considered even with normal numbers.

Kidneys (affects roughly 30% of patients)

The kidney's filtering system is gradually damaged. **Proteinuric nephropathy** — protein leaking into the urine — is the first sign. Approximately 75% of those who develop kidney disease are diagnosed before age 35. Without monitoring and treatment, most of those affected progress to end-stage renal disease within a few years of the protein first appearing in the urine.

Hearing (near-universal, highly variable)

Sensorineural hearing loss is seen in almost all patients, with the age of onset ranging from the first to the sixth decade and severity varying greatly — from mild defects to profound deafness. Some people lose hearing in childhood; others only notice changes in their 50s or 60s.

Eyes (about 20% of patients)

Cataracts occur in about one in five patients, are often bilateral, and usually appear in early adulthood. Congenital cataracts have been reported. Regular eye exams can catch cataracts early, while they are most treatable.

Liver (often present but rarely symptomatic)

Elevated liver enzymes are a documented feature of MYH9-RD, though the condition rarely causes active liver disease. Testing once every few years is adequate for most patients.

SECTION 03

The Spectrum — Five Conditions, One Disease

For decades, these presentations were named and studied as separate conditions. Once the MYH9 gene was identified as the common cause, researchers realised they were all the same disease — just with different combinations of features. The label a doctor uses often depended on which symptoms

appeared first and how the patient was examined.

Condition	Blood features	Organ involvement	Notes
May-Hegglin anomaly	Large platelets and leukocyte inclusion bodies	None	The mildest presentation
Sebastian syndrome	Same as above	None	Inclusion-body appearance differs on electron microscopy
Epstein syndrome	Large platelets (inclusions not visible on standard staining)	Kidney disease + hearing loss	Inclusions may still be present but require immunofluorescence
Fechtner syndrome	Large platelets + visible inclusion bodies	Kidney + hearing + cataracts	The most complete presentation
DFNA17	May be normal on CBC	Hearing loss	Originally classified as isolated deafness; confirmed as MYH9-RD

Because the condition evolves over time, someone may start with what looks like May-Hegglin anomaly and later develop features of Fechtner syndrome. For this reason, regardless of the label originally assigned, the current recommendation is to monitor all MYH9-RD patients for kidney, hearing, and eye changes.

SECTION 04

A Brief History of How We Got Here

1985

Dr. L.C. Peterson and colleagues publish the first description of the **Fechtner family** in the journal *Blood*. The syndrome is named after this family. Eight members across four generations are affected. It is initially classified as a variant of Alport syndrome because of the kidney and hearing similarities.

1988

A second family is documented, confirming the syndrome is real and recurring.

2000

Scientists from the May-Hegglin/Fechtner Syndrome Consortium identify mutations in the **MYH9 gene** as the cause, linking May-Hegglin anomaly, Fechtner syndrome, and Sebastian syndrome as the same disease.

2001–2003

Epstein syndrome is confirmed to share the same genetic cause. The full spectrum is unified and the term **MYH9-Related Disease (MYH9-RD)** is proposed.

2008

First study confirms that ACE inhibitors and ARBs reduce kidney damage in MYH9-RD patients (Pecci et al., *Nephrology Dialysis Transplantation*).

2010

Eltrombopag (a platelet-boosting medication) is shown to be effective in raising platelet counts in MYH9-RD patients (Pecci et al., *Blood*). Cochlear implants are confirmed safe and effective for hearing loss in these patients.

2014

A landmark **prognostic model** is published showing that where in the MYH9 gene the mutation sits predicts the likely course of the disease. Head / motor domain mutations carry higher risk of serious organ involvement; tail domain mutations tend to be milder.

2020

Research confirms that megakaryocyte migration defects underlie the thrombocytopenia — deepening understanding of the mechanism.

2024–2025

Multiple studies confirm the widespread misdiagnosis problem. **Avatrombopag** emerges as an alternative when eltrombopag fails. A nationwide Japanese registry study and a Chinese retrospective of 40 misdiagnosed patients highlight the global scale of the problem. A 2025 case report confirms MYH9-RD can present with a normal platelet count, complicating diagnosis further.

SECTION 05

The Gene — What MYH9 Does and Why It Matters

The MYH9 gene contains the instructions for making a protein called **non-muscle myosin heavy chain IIA**. Myosin is one of the motor proteins in your cells — the part that drives movement, shape change, and division. Non-muscle myosin IIA is specifically the motor used by cells that are not muscle cells, including platelets, white blood cells, kidney podocytes, inner ear hair cells, and eye lens cells.

When MYH9 is mutated, every cell that relies on this motor has to work with a defective engine. Platelets are the first to show the defect because they are produced in massive numbers from a cell called a megakaryocyte, and their shape, size, and release depend critically on this motor. That is why platelets in MYH9-RD are large and sometimes fewer.

Why location in the gene matters

The MYH9 gene has two major regions: the **head (motor) domain**, which does the mechanical work, and the **tail domain**, which keeps the protein stable. The 2014 prognostic model showed that mutations in the head domain carry a much higher risk of kidney disease and hearing loss, while tail domain mutations tend to cause milder presentations with blood abnormalities alone.

When a genetic report is returned, it will list the exact mutation and sometimes name the affected domain. Knowing which domain is involved helps your care team determine whether you need closer kidney and hearing monitoring from the start, or whether standard monitoring is sufficient.

> **ASK YOUR DOCTOR**
>
> If you have MYH9-RD, ask your hematologist or geneticist to explain which domain your mutation is in. This is clinically meaningful information that should be part of your own understanding of your condition.

SECTION 06

Diagnosis — What Should Be Tested and How

Diagnosis of MYH9-RD is not difficult once someone thinks to look for it. The challenge is that standard blood testing does not show what is wrong. A human looking through a microscope is essential.

Step 1 — Complete Blood Count (CBC)

The CBC will typically show a low platelet count, though the count can occasionally be normal. It may not capture the unusual size of the platelets accurately because most analyzers discard platelets above a certain size threshold as noise.

Step 2 — Peripheral Blood Smear

A slide of blood viewed under a microscope. The doctor looks for **giant platelets** and the faint pale-blue **Döhle-like inclusion bodies** in neutrophils. This is the single most important test and is where most missed diagnoses are eventually caught.

Step 3 — Immunofluorescence Staining

A more sensitive test that stains specifically for the MYH9 protein in neutrophils. The pattern of staining reveals the diagnosis even when standard stains miss the inclusions. Considered the most reliable diagnostic sign short of genetic testing.

Step 4 — MYH9 Genetic Testing

Confirms the specific mutation and its location, which helps predict the course of the disease. Typically performed by a clinical genetics laboratory. In most healthcare systems this is available on request once the smear findings are consistent with MYH9-RD.

> **FOR NURSES AND EMERGENCY STAFF**
>
> If a patient with known MYH9-RD or Fechtner syndrome presents for any procedure, bleeding episode, or surgery, **the platelet count alone is not a reliable guide to bleeding risk.** These patients need individualized assessment. Platelet transfusion can be used for active bleeding that cannot be controlled locally but is not a routine prophylactic measure.

Why it is so often missed

The low platelet count looks identical to immune thrombocytopenia (ITP) on a standard CBC. Doctors often start treating for ITP with steroids or immunosuppressants, which do nothing for MYH9-RD. Some patients go years or even decades before someone looks at a smear and notices the giant platelets and inclusion bodies.

SECTION 07

Monitoring — What Should Be Checked and When

Once diagnosed, patients need a team of specialists working together. The following monitoring schedule is recommended based on current guidelines.

For all MYH9-RD patients

Test	Frequency
Complete blood count & platelet assessment	Annually
Urine test for protein (proteinuria screening)	Annually
Kidney function (creatinine, eGFR) — standard risk	Annually
Kidney function — high-risk gene mutations	Every 6 months
Hearing test (audiogram)	Every 3 years
Eye exam for cataracts	Every 3 years
Liver enzyme check	Every 3 years

Things to avoid

- **Drugs that inhibit platelet function** (aspirin, ibuprofen, and similar NSAIDs) should only be used after a careful risk-benefit conversation with your doctor.
- **Drugs that are toxic to the kidneys** (nephrotoxic) **or ears** (ototoxic) — always tell every prescribing doctor and dentist about your diagnosis.
- High-risk activities with significant injury risk.
- Hazardous noise exposure (use hearing protection).

BEFORE ANY PROCEDURE

Before any surgery, dental procedure, or delivery: tell your care team well in advance. Platelet counts may need to be temporarily raised using medications (see Treatment section). **Never have elective surgery without your hematologist involved in planning.**

SECTION 08

Treatment — What Helps Today

There is currently no cure for MYH9-RD. Treatment focuses on protecting the organs that are at risk and managing symptoms when they arise.

Kidney protection

ACE inhibitors (such as enalapril, lisinopril, ramipril) and **ARBs** (losartan, valsartan) reduce protein leakage and slow kidney damage. Treatment is typically started when urine protein first appears, before kidney function declines. Pecci et al. confirmed their effectiveness in MYH9-RD in 2008.

Raising platelet counts for procedures

Eltrombopag and **avatrombopag** are oral medications that stimulate platelet production. They are particularly useful to raise platelet counts in advance of surgery, dental extraction, or delivery. Romiplostim (an injectable alternative) is also used in some settings.

Avatrombopag has emerged in recent years as a useful alternative when eltrombopag is not tolerated or is not available, and has shown good responses in MYH9-RD in 2024–2025 studies.

Hearing

Hearing aids help in early-to-moderate hearing loss. **Cochlear implants** are confirmed safe and effective in MYH9-RD patients when hearing loss becomes severe enough that hearing aids are no longer adequate. Referral to a specialist audiologist or otolaryngologist familiar with genetic hearing loss is recommended.

Cataracts

Surgical removal with intraocular lens replacement is the standard treatment, and the surgery itself is considered low-risk in MYH9-RD with careful perioperative planning. Regular eye exams catch cataracts when they are still small.

End-stage renal disease

When kidney function declines to the point of needing renal replacement therapy, **dialysis** (both hemodialysis and peritoneal dialysis) and **kidney transplantation** have been performed successfully in MYH9-RD patients. A November 2025 Springer case report documented successful transplantation following hemodialysis as a bridge therapy.

Pregnancy and delivery

Women with MYH9-RD can have successful pregnancies, but advance planning with both a hematologist and an obstetrician familiar with platelet disorders is essential. Platelet counts may need to be raised before delivery, and specific anaesthesia considerations apply. Discuss options with your care team well before the third trimester.

SECTION 09

Current Research — What's Emerging

The last three years have seen meaningful progress on several fronts.

Improved diagnostic approaches

A 2024–2025 series of papers in the *Journal of Thrombosis and Haemostasis*, *British Journal of Hospital Medicine*, and *Frontiers in Pediatrics* have pushed for clearer screening algorithms to catch MYH9-RD earlier, particularly in patients misdiagnosed as ITP who do not respond to standard treatment. Automated smear analysis and AI-assisted platelet size measurement are in early development.

Treatment options

Avatrombopag has established itself as a viable alternative to eltrombopag. Longer-term safety data on both medications in MYH9-RD specifically has accumulated, with a 2025 Blood (ASH abstract) update confirming sustained efficacy over multi-year follow-up.

Transplantation outcomes

A November 2025 Springer case report documented successful kidney transplantation following hemodialysis as a bridge therapy, adding to the growing evidence base for renal replacement approaches.

Misdiagnosis research

A 2025 Chinese retrospective study of 40 patients misdiagnosed as ITP detailed the diagnostic journey and proposed clearer screening algorithms to prevent the problem. The scope of misdiagnosis is now being treated as a systemic issue rather than individual missed cases.

SECTION 10

Leading Researchers and Centers

The following researchers and institutions are the primary producers of MYH9-RD research worldwide. They are listed here because patients with rare conditions often benefit from knowing who the experts are — either for referral, for second opinions, or for contributing to research registries.

The world's leading expert on MYH9-RD

Dr. Alessandro Pecci, MD, PhD

Department of Internal Medicine

IRCCS Policlinico San Matteo Foundation & University of Pavia · Pavia, Italy

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Dr. Pecci has authored or co-authored the majority of landmark papers on MYH9-RD over the past two decades, including the definitive GeneReviews entry, the 2014 prognostic model, the eltrombopag clinical trial, and the 2021 comprehensive inherited thrombocytopenia guide. He is the primary author of record for anyone seeking expert consultation or collaboration.

Additional leading researchers

Dr. Anna Savoia, PhD

Institute for Maternal and Child Health — IRCCS Burlo Garofolo
University of Trieste · Trieste, Italy

Co-author of the GeneReviews MYH9-RD entry and a leading genetics researcher on the molecular basis of the disease.

Dr. Andreas Greinacher, MD

University Medicine Greifswald · Greifswald, Germany

One of the earliest researchers to characterize MYH9-related platelet disorders. His group contributed foundational work on the clinical recognition and perioperative management of MYH9-RD.

Dr. Shinji Kunishima, MD

Department of Medical Technology
Gifu University of Medical Science · Japan

Leading researcher in Asian populations with MYH9-RD and pediatric presentations.

Centers with specialist expertise

University of Pavia / IRCCS Policlinico San Matteo

Pavia, Italy

The single most productive center globally for MYH9-RD research and the home of the **Italian Registry of MYH9-RD**. Patients from outside Italy have contacted this center for expert consultation. For referral or registry inquiries, contact Dr. Pecci directly (see above).

SECTION 11

Patient Resources and Organizations

Authoritative medical resources

GeneReviews — MYH9-Related Disease (NCBI)

The most comprehensive clinical resource, authored by Pecci and Savoia. Updated 2021. Free and openly available.
https://www.ncbi.nlm.nih.gov/books/NBK2689/

MedlinePlus Genetics — MYH9-Related Disorder

Plain-language overview maintained by the U.S. National Institutes of Health.
https://medlineplus.gov/genetics/condition/myh9-related-disorder/

Orphanet — MYH9-Related Disease entry

The authoritative rare disease database. Includes expert reviewer information, diagnostic criteria, and clinical management summaries.
https://www.orpha.net/en/disease/detail/182050

Patient support organizations

Platelet Disorder Support Association (PDSA)

The primary patient-facing organization for platelet disorders in North America. Focus is primarily ITP, but PDSA maintains resources and provides community for inherited platelet disorders including MYH9-RD.
https://pdsa.org

NORD — National Organization for Rare Disorders

Provides advocacy resources, rare disease center directory, and support network tools.
https://rarediseases.org

Bare Your Rare

A patient-led knowledge node for rare diseases. If you have been diagnosed with MYH9-RD or suspect it, your story and data can help others.
https://bareyourrare.org

SECTION 12

Glossary

Plain-language definitions of medical terms used in this guide.

Autosomal dominant
A genetic inheritance pattern where you only need one copy of the changed gene to have the condition. Each child of an affected parent has a 50% chance of inheriting it.

Avatrombopag
An oral medication that stimulates platelet production. Used as an alternative to eltrombopag.

Cochlear implant
A surgically implanted device that can restore functional hearing when hearing aids are no longer sufficient. Confirmed safe in MYH9-RD patients.

De novo mutation
A new genetic change that occurs in the patient and was not inherited from either parent. About 30–35% of MYH9-RD cases are de novo.

Döhle-like inclusion bodies

Small pale-blue spots seen inside white blood cells on a blood smear. A key diagnostic clue in MYH9-RD.

Eltrombopag

An oral medication that stimulates platelet production. The first platelet-boosting drug confirmed effective in MYH9-RD (Pecci et al., 2010).

Immunofluorescence staining

A laboratory technique that tags specific proteins with fluorescent markers so they can be seen under a microscope. The most reliable diagnostic test for MYH9-RD short of genetic sequencing.

Macrothrombocytopenia

The combination of low platelet count and unusually large platelets. The hallmark blood finding of MYH9-RD.

Megakaryocyte

The bone marrow cell that produces platelets. In MYH9-RD, these cells have difficulty moving and releasing platelets normally.

MYH9

The gene that carries the instructions for non-muscle myosin heavy chain IIA. Mutations in this gene cause the full spectrum of MYH9-RD.

Peripheral blood smear

A slide of blood examined under a microscope by a trained pathologist or hematologist. The single most important diagnostic test for MYH9-RD.

Proteinuria

Excess protein in the urine. The earliest sign of kidney involvement in MYH9-RD.

Sensorineural hearing loss

Hearing loss caused by damage to the inner ear or auditory nerve. The typical pattern of hearing loss in MYH9-RD.

Spectrum disorder

A range of presentations of one underlying condition, from mild to severe.

TPO receptor agonist

The class of drugs that eltrombopag, avatrombopag, and romiplostim belong to. They mimic thrombopoietin, the hormone that signals the body to make more platelets.

Variable expressivity

The same gene change can cause very different symptoms, even in members of the same family.

SECTION 13

Your Personal Action Checklist

If you think you might have this condition

- Ask your doctor for a **peripheral blood smear** — not just a standard blood count.
- Ask specifically about **MYH9 gene testing**.
- Request referrals to a hematologist and a nephrologist familiar with rare diseases.
- Ask whether any family members should also be tested.

If you already have a diagnosis

- Make sure every doctor, dentist, and emergency room you visit knows about your platelet disorder before any procedure.
- Wear a medical alert bracelet or carry a wallet card.
- Know your specific mutation — ask your doctor which part of the gene is affected, as this predicts your risk for kidney and hearing involvement.
- Follow the monitoring schedule (annual kidney tests, hearing tests every 3 years, eye exams every 3 years).
- Before any surgery, dental extraction, or delivery: contact your hematologist **at least 2–3 weeks in advance** so platelet-boosting medications can be arranged if needed.
- Avoid aspirin and ibuprofen unless your doctor has specifically said they are safe for you.
- If you are planning a pregnancy, speak with both your hematologist and your OB beforehand.

For family members of someone diagnosed

- First-degree relatives (parents, siblings, children) all have a **50% chance of carrying the gene change**.
- Ask your family member's specialist about getting tested.
- A negative family history does not rule out the condition — testing is the only way to know.

A CLOSING NOTE

You are not alone.

MYH9-RD is one of those conditions that teaches you how much a careful look through a microscope can change a life. Somewhere, right now, someone is being treated for ITP with medications that will never help them. They don't know yet that the answer exists. If this guide shortens the path for even one person — or arms one patient with the information they need to push back in a consult — it has done its job.

If you have been diagnosed with MYH9-RD or Fechtner syndrome, or if you are the family member of someone who has, please consider sharing your experience at bareyourrare.org. Rare diseases do not suffer from a lack of intelligence. They suffer from a lack of aggregation. Every story helps the next patient find their answer faster.

Thomas Cheesman

Founder, Bare Your Rare · HCS Patient · Grande Prairie, Alberta

Verification note: All content in this guide was verified against peer-reviewed medical literature including Pecci & Savoia, *GeneReviews MYH9-Related Disease* (2021); Orphanet 2014 (MYH9-RD); *Clinical Kidney Journal* 2019; PMC 2024 misdiagnosis case series; *Springer Renal Replacement Therapy* November 2025; *Journal of Thrombosis and Haemostasis* 2025; *Blood* (ASH) 2025; *Frontiers in Pediatrics* 2024; *British Journal of Hospital Medicine* January 2026.

This guide is educational and does not replace medical advice. Decisions about diagnosis, treatment, and monitoring should be made with a qualified healthcare provider who knows your individual history. Version 1.0, April 2026.