If you see a patient with a family history of chronic kidney disease
It could be **FABRY DISEASE**

In addition to a family history of chronic kidney disease, patients with Fabry disease may present at an early age with:

- Progressive and/or unexplained chronic kidney disease
- Proteinuria/isosthenuria
- Tubular dysfunction (polyuria, polydipsia)
- Symptoms suggesting Fanconi’s syndrome
- Elevated serum creatinine

Other manifestations include:

- Premature cardiac disease
- Gastrointestinal problems
- Premature stroke
- “Burning” pain in the hands and feet
- Heat/cold and exercise intolerance
- Impaired sweating
- Angiokeratomas (reddish-purple skin lesions)

Information for Nephrologists

Nephrologists have the opportunity to identify patients with this progressive, often life-threatening genetic disease.

While Fabry disease is rare, it may be more common within a Fabry family.

**FABRY DISEASE**
FABRY DISEASE PROFILE
Fabry disease is an inherited disorder that affects men, women, and children of all ethnicities. It is a multisystemic disorder that ultimately results in irreversible, potentially life-threatening disease of the kidney, heart, and brain. The disease is characterized by the progressive and unrelenting cellular accumulation of a lipid substrate called globotriaosylceramide (or GL-3). Ongoing build-up of this substance is caused by deficiency of the lysosomal enzyme alpha galactosidase A (or α-GAL), which usually metabolizes GL-3 and keeps it from accumulating. Without enough of this essential enzyme, GL-3 accumulates in the lysosomes of most cell types over the course of a lifetime, often causing debilitating symptoms in childhood and adolescence and potentially irreversible tissue damage by adulthood.

DISEASE RISK IN FAMILIES
• Unlike many other X-linked disorders, females with the defective gene are affected to varying degrees due to random X inactivation.
• Males with the disease pass the defective gene on to all of their daughters and none of their sons.
• Females have a 50% chance with each pregnancy of passing the defective gene to both their sons and daughters.
• If you identify a patient with Fabry disease, family testing should be considered.

DIAGNOSIS
• Although Fabry disease usually presents in childhood, the disease often goes unrecognized by physicians until adulthood, when the underlying pathology is advanced.
• Delayed diagnosis may be the result of disease under-recognition and/or symptoms being mistaken for those of other disorders, such as rheumatoid or juvenile arthritis, rheumatic fever, erythromelalgia, multiple sclerosis, or lupus.
• Diagnosis is confirmed in males by enzyme assay (blood test) detecting low or absent levels of alpha-galactosidase A (α-GAL), or in females through genetic testing to detect a mutation.

Early diagnosis and intervention are key.
Nephrologists can play a role.