Did you know that some platelet disorders are passed down through families?

**RUNX1 Familial Platelet Disorder (RUNX1-FPD)** is one of these rare inherited disorders, and is often unknowingly overlooked when a diagnosis is made. Approximately 10-20% of those diagnosed with **RUNX1** were previously diagnosed with at least one of the following:

- Immune Thrombocytopenic Purpura (ITP)
- Von Willebrand Disease
- Platelet Storage Pool Disorder

Obtaining an early and accurate diagnosis of **RUNX1** can help you avoid extra or inappropriate treatment, as well as enable more precise medical management. However, it is important to discuss the risks and benefits of genetic testing with an experienced genetic counselor beforehand.

Here are some FAQs to review with your physician while discussing your potential need for **RUNX1** genetic testing.

### 1. What is **RUNX1-FPD**?

The **RUNX1** gene provides the blueprint for a protein that is essential to the formation of our blood system and critical for the proper function of blood cells, like platelets. **RUNX1-FPD** is a disorder caused by one of over 100 different mutations in this gene, which causes low and/or dysfunctional blood platelets, and predisposes an individual to developing blood cancer. The most common blood cancers **RUNX1-FPD** patients develop are acute myeloid leukemia (AML) and myelodysplastic syndromes (MDS).

### 2. Why aren't more people diagnosed with **RUNX1-FPD**?

The link between **RUNX1** mutations and blood cancer was discovered in 1999, so awareness of it in the medical field is still relatively low. Experts agree that the disorder is also largely undiagnosed due to the lack of genetic testing required to identify it. While over 200 families have been described in the scientific literature, estimates suggest as many as 18,000 people may be living with **RUNX1-FPD** in the United States alone.

### 3. Does this disorder present the same way for all affected family members?

Another factor making this disorder difficult to diagnose in some families is that **RUNX1-FPD** causes a variable decrease in platelet counts (from profoundly abnormal counts to those that are within the normal range) and variably disrupts platelet function. So the severity of bleeding varies across individuals, even within the same family. Also, while some families may have a history of blood cancers, others do not.

### 4. Does having **RUNX1-FPD** mean I will develop cancer?

Not necessarily. Individuals with **RUNX1-FPD** have a 35-50% risk of developing a blood cancer, and the median age of cancer onset is 29 years. This means the timing of blood cancer ranges significantly and can happen early in childhood or late in adulthood. Early diagnosis can help you and other affected family members develop a personalized cancer surveillance plan with your doctor.

### 5. How do you determine if you have **RUNX1-FPD**?

If you and your health care team decide to test for **RUNX1-FPD** or other inherited platelet disorders, a qualified genetic testing lab can test saliva samples. In some cases, blood, bone marrow, or skin samples are required for an accurate and complete diagnosis.