The Platelet Charity
Helping people with bleeding disorders

Quick Guide to Platelet Function Disorders

What are they?

The term "Platelet function disorder" (PFD) could be applied to any condition where platelets do not work properly. Platelets are small particles in the blood that stick together at sites on injury (for example after a cut) to form a "plug" and stop the bleeding. If your platelets don't work properly you bleed more than normal, and this can vary from minor bruising to life threatening bleeding after surgery or an accident.

PFDs can be inherited (patients are born with them and will have them throughout life), or acquired (they appear later in life and may disappear). Acquired PFDs are more common in the population, with ITP (immune thrombocytopenia purpura) being the commonest one. Acquired PFDs can also be due to kidney problems or medications. The Platelet Charity focuses on inherited PFDs which are much rarer. PFDs are sometimes associated with the platelet count in blood being lower than normal (the medical term for this is "thrombocytopenia"), but it's also possible to have them with a normal platelet count.

Who gets them?

Anyone can have an inherited PFD, and getting them is not caused by any factors in the diet or in the environment such as smoking or drinking alcohol. Many patients with PFD are diagnosed when they are children as they bruise a lot when they are toddling or playing, but some patients are not diagnosed until much later in life, even into their seventies. In families where other members of the family are known to have PFD parents will often be alert to the possibility and have children tested at a very young age.

How do they affect day to day life?

This varies a lot according to the type of PFD. Some patients only get very minor bruising which can be painful and not look very nice, but is not life threatening. Some patients can have really severe bleeding such as nosebleeds that won't stop and lead to anaemia (a low red blood cell count), or very heavy periods in young girls and women. Having surgery (or having children in women) is a time of particular worry and many patients need treatment prior to these events. Patients with PFDs will be registered with the doctors and nurses at a haemophilia centre and will see them intermittently or if they need treatment. The haemophilia centre can often be a long way from their home, and attending can make patients miss time from work or school. Some patients carry a

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treatment pack with them at all times so they can give treatment quickly if they do develop a bleed. Most patients with PFD in the UK carry a "green card" to alert medical staff to the fact that they have a bleeding disorder. Simple things like going on holiday can be problematic, as patients with PFDs will need to find out where they should go if they develop a problem whilst they are away, and they may need to carry treatment with them. Some patients with PFDs are advised not to participate in sports with a lot of physical contact or risk of head injury, such as rugby or boxing. The label of having a PFD and the symptoms associated with it can cause embarrassment and stigma to some patients who just want to be "normal".

What treatments are available?

Current treatments include tranexamic acid liquid, tablets or injections, which strengthen blood clots and make them resistant to being broken down. Desmopressin or "DDAVP" is also given either as a nasal spray or an injection under the skin to increase the platelets' stickiness. In serious bleeding platelet transfusion may be given, although only if absolutely necessary. Injections of other clotting factors such as factor VII (seven) are sometimes used, and new agents which are based on the platelet stimulating hormone thrombopoietin may become more widely used over the next few years.

Why is it important to know more about them?

Diagnosing a PFD is complicated, so at present some patients who bleed more than normal don't know if they have a PFD or not. Research into improving the diagnosis will hopefully help these patients and also their families by giving them a more specific diagnosis. At the moment, PFDs are all treated in a similar way, but knowing more about the different types of PFD may improve treatment options and mean treatment can be tailored to the specific PFD that a patient has. Lastly, finding out more about their inheritance will help future generations and will give young patients with PFD very important information with regard to planning their families.

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