FOP

Fibrodysplasia ossificans progressiva:

the essential facts

This leaflet is intended to give a brief overview of the rare genetic condition, fibrodysplasia ossifcans progressiva, or FOP.

FOP is a complex illness and anyone who suspects a diagnosis of FOP should contact a medical professional and not rely on the information given here for diagnostic or treatment purposes.

For trusted and detailed medical information, please contact FOP Friends to be directed to the latest agreed medical guidelines from qualified FOP doctors:

info@fopfriends.com

or visit the website for the International Clinical Council for FOP:
iccfop.org

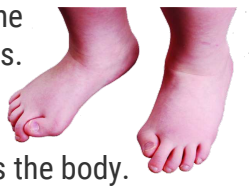
Fibrodysplasia ossificans progressiva: the essential facts

What is FOP?
Fibrodysplasia ossificans progressiva, or FOP, is an ultra-rare genetic condition that affects around 1 in 1.4 million people. It is one of the most disabling conditions known to medicine.

FOP causes the soft connective tissue of the body to turn into new bone. When that occurs over or near joints, or within a muscle, it restricts the person's movements. This new bone, or ossification, can mean that the sufferer is no longer able to move the joint. Once movement has been lost in a part of the body, it is not possible to remove the new bone as that can aggravate the FOP and trigger further bone growth.

What causes FOP?
Most cases of FOP are new. It is caused by a fault in the ACVR1 gene and this gene mutation usually happens at conception: it is an accident of nature. Where one parent has FOP, the likelihood of passing the condition on to a child is 50%.

Symptoms
People with FOP appear normal at birth, except for the tell-tale malformed, turned-in big toes. The big toes may be shortened and curl under. The person may also have shorter or turned in thumbs. The toes, combined with unexplained swellings across the body, can be an indicator of FOP.



The swellings are known as flare-ups. These are painful lumps that can appear anywhere across the body. They can be red and inflamed. They can last from a few weeks to a few months, causing inconvenience and discomfort, and be excruciatingly painful for the patient. A flare-up can appear spontaneously or after an incident. As the flare-up subsides, new bone growth may have occurred.

Progression
FOP is a variable and progressive illness. Whilst a fall, injury or virus can trigger an FOP flare-up, a flare-up can also appear spontaneously and without warning.

Some people with FOP suffer from flare-ups from very early on in life, even as young as birth, whilst other people may not have any symptoms or restrictions until their 20s or beyond. FOP can cause people to have long periods of flare-ups whilst at other times the FOP can be quiet for months or even years before something triggers it again. People can experience more than one flare-up at a time. They can also suffer one flare-up after another for a period of time; this is known as a flare cycle.

Assessment
If you are concerned that you or someone else may have FOP, you should see your GP and request a referral to the Centre for Metabolic Bone Disease at the Royal National Orthopaedic Hospital, Stanmore. They have an experienced FOP medical team who will be able to advise and assess your symptoms. They will carry out investigations and/or request a genetic blood test to confirm the diagnosis.

Due to the rarity of FOP, many medical practitioners will never have heard of the condition. Under no circumstances should the lumps be biopsied before confirmation, as this can aggravate the FOP and cause irreversible damage.

What can make FOP worse?
A knock or fall can trigger the FOP and cause a flare-up. Any impact into the muscle can trigger FOP activity. Precautions need to be taken to reduce the likelihood of a person with FOP encountering unnecessary risks.

Intramuscular injections (IM) can cause FOP activity so should be avoided if at all possible. Where medically necessary, *specialist advice should be sought prior to any administration of injections*. Injections should be administered by skilled and experienced practitioners and *not* medical students.

Vaccinations and childhood immunisations can aggravate FOP but the benefits may outweigh the risks. Advice should be sought from an experienced FOP medical specialist before carrying out any immunisations. The NHS childhood vaccination programme can be delayed until the diagnosis is confirmed and the precautionary steps have been advised and agreed. Most vaccines can be administered sub-cutaneously (under the skin) to reduce the risk of aggravating the FOP. Some vaccinations can be omitted, under advice from your GP.

Muscle fatigue can trigger a flare-up, as can viruses, with flu being of significant risk.

Surgery carries an extremely high risk and should be avoided unless absolutely necessary. Where surgery is required, expert advice should be sought and appropriate steps taken to minimise the risk of any FOP complications. ***In the event of an emergency, protection of life should be taken as if FOP is not a consideration.***

Dental treatment can be extremely invasive and carries the risk of triggering extra bone formation in the jaw, resulting in restrictions in the jaw movement. Seek advice from a Special Care Dentist.

Potential Complications
All injections and vaccinations should be administered sub-cutaneously wherever possible. IM injections should be avoided. No vaccinations should be given if a person is suspected to be in a state of active flare.

People with FOP need to take precautions against flu, as the illness can cause complications for them.

People with FOP can often develop scoliosis which causes further complications.

Invasive dental treatments carry a higher risk for people with FOP, so excellent preventative dental hygiene is vital.

Hearing is affected by FOP, with around 50% of people reporting some or complete loss.

People with FOP can suffer from mental health issues as they come to terms with living with the condition.

Is there a treatment?
At present there is no known treatment or cure for FOP. There are a number of medications and therapies people use that may have some efficacy. Seek qualified medical guidance before making any decisions.

Prednisolone: it is generally accepted that there may be benefits to a person with FOP taking a short course of the steroid prednisolone to reduce inflammation. It is not suitable for regular use, nor for all flare-ups.

Ibuprofen: there is some anecdotal evidence that ibuprofen can help to reduce inflammation after a minor impact.

Imatantib: whilst there is no definitive evidence that imatinib prevents flare-ups, there have been some reports that it can help to break flare-cycles in children.

Montelukast (Singulair): this may help control or reduce unwanted inflammation in the lungs. Some people take this daily in the hope it might have a beneficial/potential effect on future flare-ups. There have been no formal studies to assess the drug's effectiveness for FOP.

Research
There is much research being carried out around the world into a treatment and a cure for FOP.

There is real hope for people who are suffering from this disease.

There are research centres at the University of Oxford and the University of Pennsylvania, USA, as well as other centres across Europe, America and beyond.

FOP Friends fundraises to support the FOP research team at the University of Oxford and works closely with its researchers.

Clinical trials
The Royal Nation Orthopaedic Hospital (RNOH), Stanmore, London, is the UK's clinical trial site for potential treatments for FOP. There are a number of active trials for treatment for FOP underway,

For up to date details of current trials, visit www.clinicaltrials.gov or contact FOP Friends.

Clinical guidelines
In July 2019, the International Clinic Council for FOP (a consortium of 26 medical professionals and consultants) released updated medical guidelines for the management of the condition, and treatment considerations for patients with FOP. www.iccfop.org All patients, their carers, their primary medical physician/s, and their local hospital should have a copy. These guidelines can be obtained, free of charge, by contacting FOP Friends or the IFOPA.

They are also available on the FOP Friends' website or scan this code.

Is there support?
Yes. Whilst FOP is a rare condition, only affecting around 70 people in the UK, there is a strong and supportive community for those who want it.

FOP Friends is the UK's registered charity. It exists to support people who are living with FOP and their families, offering advice and friendship. It is a run by parents of a child with FOP, so they understand the struggles and challenges faced by people living with FOP.

FOP Friends hold a biennial conference in Manchester where people with FOP, their families, researchers and medical professionals get together for research updates as well as friendship and support. There is also a family weekend for children with FOP and their families every two years.

The International FOP Association - IFOPA - is the international charity for people with FOP and those who care for them. They too can offer a wealth of advice, information and support.

There are a number of FOP support groups on Facebook, where patients and their families can connect with people across the UK and around the world, with people who truly understand what living with FOP means. These groups are private and secret. Contact FOP Friends if you would like to find out more, or be added to these groups.

How do I contact FOP Friends?

Tel: 0161 282 0441 www.fopfriends.com Email: info@fopfriends.com

Further information
This leaflet has been produced by FOP Friends. It is intended to be an overview of the condition, not a comprehensive guide.

FOP is variable in nature, and as such affects different people, in different ways, and at different stages in their lives. Speak to an FOP specialist if you have any concerns about your health or the health of someone else.

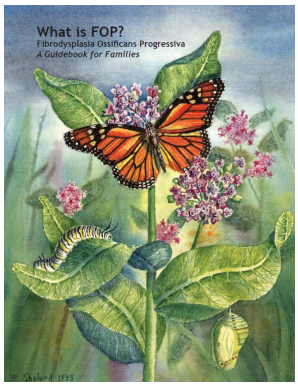
FOP Friends can signpost people who live with FOP, or their family, to other relevant services that will be able to help them to live with, and manage, the condition.

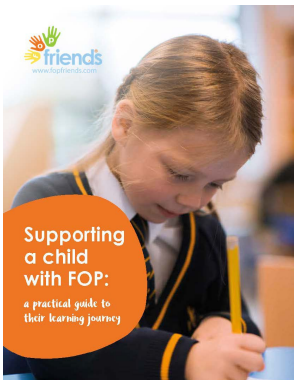
FOP Friends works closely with the medical team at the RNOH. They collaborate with the IFOPA and other FOP patient organisations around the world. FOP Friend's Chairman serves on the Board of the IFOPA and is also Chair of the International President's Committee.

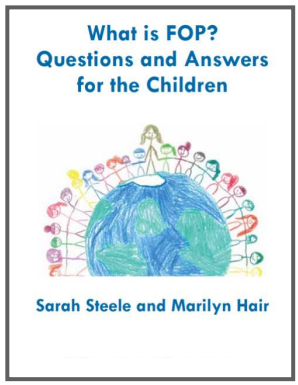
FOP Friends also works with the research team at the University of Oxford.

FOP Friends produces a quarterly newsletter as well as sending out monthly updates. Get in touch to be added to the mailing lists.

Further reading
The following publications are available, free of charge, from FOP Friends.







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Notes:

www.fopfriends.com

FOP Friends is a charity that exists to support people and families affected by fibrodysplasia ossificans progressiva