Hereditary Multiple Exostoses-Multiple Osteochondromas (HME-MO) Association The Netherlands

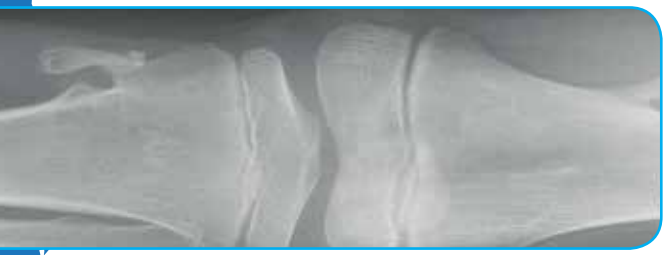
HME-MO VERENIGING NEDERLAND
Hereditaire Multiple Exostosen - Multiple Osteochondromen

What is the HME-MO?

Hereditary Multiple Exostoses-Multiple Osteochondromas (HME-MO) is a hereditary skeletal disorder which affects the long bones, pelvis, ribs and the shoulder girdle. Characteristic are many benign bone tumours (exostoses/osteochondromas) which are provided with a cartilage cap and sometimes a bursa. The osteochondromas often arise at a very young age and grow especially during growth age very fast. HME-MO is a hereditary, autosomal dominant disorder. This means that someone with HME-MO has a 50% chance of passing on the illness to his or her children. In about 20% of cases there is a so-called spontaneous mutation involved. This way a child can get HME-MO without any of the parents having it. HME-MO is caused by one of the two known genes, i.e. EXT1 or EXT2 on chromosome 8 and 11, respectively. HME-MO is a very rare condition and occurs at estimated 800 people (children and adults) in the Netherlands.

Diagnosis

The World Health Organization (WHO) changed the name of the syndrome from HME to Multiple Osteochondromas (MO) in 2002 because the term "exostosis", bone outgrowth,



was nonspecific and confusing. According to the WHO the diagnosis of MO applies when at least two osteochondromas are found in a patient. Likewise one or more relatives with MO or a mutation in one of the EXT- genes need to be known. In Dutch we use the name "Multiple Osteochondromen". For the time being within the HME-MO Association we use both names, Hereditary Multiple Exostoses and Multiple Osteochondromas (HME-MO).

The diagnosis of HME-MO is relatively easy to make for a practitioner on the basis of X-ray photographs. Sometimes an

additional investigation is needed by means of an MRI. In order to confirm the diagnosis, an orthopaedic surgeon can ask for a heredity investigation. In the Netherlands the practitioner may seek advice from the Commission for Bone Tumours about the diagnosis or treatment, free of charge.

Symptoms

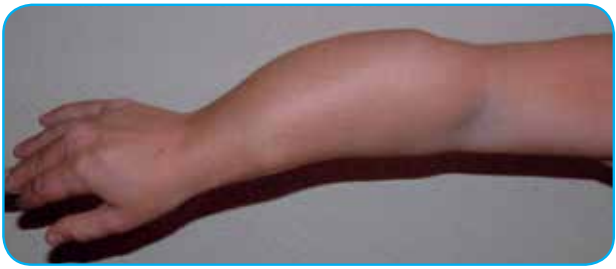
The osteochondromas (exostoses/bone overgrowths/lumps/bone outgrowths) are located on the long bones between the middle part and the end part on site of the so-called metaphysis. During childhood, the growth plate is also found here. Besides that, osteochondromas occur on the flat bones of the shoulder girdle and pelvis, but also on the vertebrae.

The shape of the osteochondromas is rather erratic. It varies from an osteochondroma sharply pointing off a joint to a wide, tumour-like distortion. Someone with HME-MO usually has dozens osteochondromas. The expression of HME-MO is very diverse. It may be so that someone with relatively few osteochondromas may have few to no complaints. On the other hand someone can develop a severe form with a lot of osteochondromas with deformities and also a lot of pain and/or difficulty in movement.

Complications

Deformities

Osteochondromas can cause growth disturbances of the bones, such as deformities of forearms, crooked fingers, feet and knees.



Malignancy

In adults with HME-MO there is a chance averaging 5% of

malignant degeneration. An osteochondroma then turns into a chondrosarcoma. This occurs mainly in the shoulder girdle and pelvis.

Other remarks

It can be said that the pressure, which the osteochondromas exert on neighbouring tissue, can cause various problems. Pressure on tendons and muscles can result in pain complaints, but also in problems with movement. Pressure on nerves can give pain as well, but also numbness and/or symptoms of systemic failure. People with HME-MO may have too steep pelvis, whereby women can experience problems at childbirth. Because of that a C-section may sometimes need to be performed.

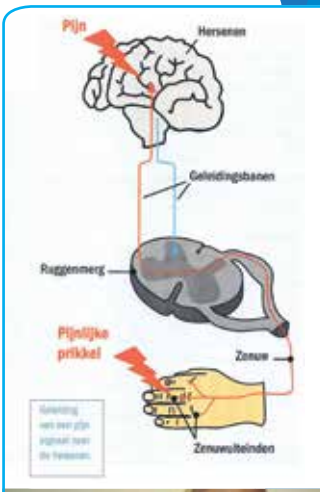
Treatment

Surgical

When someone has a lot of osteochodromas, it is not possible to remove them all. If necessary, on occurrence of movability problems, severe pain and entrapment of nerves, an osteochondroma may be removed surgically. This sometimes happens for cosmetic reasons as well. In case of young people, where the osteochondroma is situated still too close to the growth plate, considerations are sometimes made whether to postpone the surgery. There is a risk that the growth plate may be damaged which may cause growth impairments. The growth of the osteochondromas during later age may be an indication for removal as well in connection with possible malignancy. Corrective surgeries may be performed on deformities, such as correcting a tilt or lengthening e.g. a forearm.

Pain relief

If, despite removal of osteochondromas, people with HME-MO continue to suffer from pain, it is important that they learn how to deal with it. Adequate pain relief with medication may provide a solution for a shorter or longer period of time. Advice from and treatment by pain-relief clinics or pain-relief centres can be of help as well. When the pain becomes of a chronic sort, it may be sensible to take control of it through a special programme. This can be arranged at a number of establishments, in particular at rehabilitation centres.



Therapies

Sometimes physiotherapy may be a solution. Where movability is affected through pain or other causes, massage, exercise therapy or sometimes manual therapy may reduce symptoms or even make them disappear. Other sorts of therapy may also be applied, such as occupational therapy, exercise therapies, rehabilitation treatment and more alternative forms of treatment. In the case of malignant development the patient will be treated in one of the four centres which have been designated for that purpose. It may also be important to seek advice from the Commission for Bone Tumours.

HME-MO Association

The HME-MO Association was founded in December 2009. It continues the work of the HME/MO Fellow Patient Contact Group which was set up in 1997. The HME-MO Association is dedicated to the people with HME-MO and others directly involved, such as parents or partners. In addition, other interested parties may turn to the HME-MO Association for information.

The goal of the HME-MO Association is:

- Representing the interests of current and future HME-MO patients and their relatives;
- Bringing HME-MO patients in the Netherlands in contact with each other, through peer support;
- Actively collecting new information and informing those involved about it.

Office and information desk:

Geert Lammertslaan 8, 8421 RT Oldeberkoop
tel. 0516 – 451760 or 0499 - 479293
e-mail: info@hme-mo.nl, website: www.hme-mo.nl
Bank account: NL85RABO0155774123
attn. of: HME-MO Vereniging, Oldeberkoop
swift code: RABONL2U
Chamber of Commerce (Kamer van Koophandel) Leeuwarden 01168710

Board of Directors:

- Mr J. de Lange, MSc., chairman
- Ms M. Post-Bloemhoff, secretary
- Mr W. van Dijke, treasurer
- Ms A. Posthuma, 2nd secretary

Activities

The following activities are being organised:

- Annual nationwide contact day with lectures and workshops;
- Annual regional meetings for the fellow-patient contact;
- Annual children's day;

- Annual day for Teenagers;
- Stimulation of and participation in scientific research in the field of HME-MO;
- Medical Advisory Council;
- An e-mail group;
- A private Facebook group for adults;
- A private Facebook group for Teenagers;
- A private Facebook group for children;
- Twice a year publishing of the HME-MO Newsflash, incl. Kids-Flash;
- Maintaining an informative website;
- An info desk which is accessible through e-mail or telephone;
- Publication of an information leaflet;
- Publication of a comprehensive information bulletin;
- Publication of an information DVD about HME-MO;
- Participation in working groups and umbrella organisations that deal with rare diseases and the interests of the patients' organisations;
- Maintaining contacts with other foreign patients' associations in the field of HME-MO.

Inscriptions

Inscriptions can be lodged with our office. Membership costs EUR 25 a year. It is eligible for reimbursement by many health insurance providers.
It is also possible to become a donor for at least EUR 20 a year. Again, it is possible to arrange at our office.

Donations and gifts

We are very pleased with any donation or a gift. This allows us to organise additional activities and support research activities. Donations or gifts can be deposited in our bank account (see above). Please indicate the goal that you wish to support as well. Our association is recognised as an official charity (ANBI).

Foundation Research Fund HME-MO

Bank account: NL13 RABO 0140649530 attn. of Stichting Onderzoeksfonds HME-MO.

Chamber of Commerce: 53118316
This foundation, like the HME-MO Association, has a status of a recognised charity (ANBI).

Medical Advisory Council:

- Dr. S.J. Ham (chair), orthopaedic surgeon, Onze Lieve Vrouwe Gasthuis in Amsterdam
- Prof. dr. J.M.V.G. Bovée, pathologist, professor at the Department of Pathology, Leids Universitair Medisch Centrum in Leiden
- Prof. dr. R.J.E.M. Smeets, rehabilitation consultant, Professor of Rehabilitation Medical Sciences at the UMC Maastricht and Know-How Centre Adelante Hoensbroek in Heerlen
- Dr. H.J. van de Woude, radiologist, Onze Lieve Vrouwe Gasthuis in Amsterdam
- Prof. dr. W. Wuyts, Head of the Laboratory, Centrum Medische Genetica, Universiteit Antwerpen / Universitair Ziekenhuis Antwerpen, Antwerp, Belgium

Commission for Bone Tumours:

c/o department of Radiology, LUMC,
PP Box 9600, C2-5, 2300 RC Leiden

The National Treatment Centre:

Since 2009 the OLVG in Amsterdam has been formally designated as a nationwide centre for the treatment of HME-MO. A lot of experience and expertise in the area of HME-MO is present there.
For the treatment of malignant-turned bone tumours (osteochondromas) there are four designated centres, namely AMC, LUMC, UMCG and Radboud UMC.

A modified version of this brochure for the visually impaired can be downloaded from our website.

Zeldzame Ziekten

Algemeen Nut Beogende Instelling
ANBI

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VSOP ALLIANTIE VOOR ERFELIJKHEIDSVRAAGSTUKKEN