

Notes from the FDSSUK Annual Meeting

Saturday 8th October 2016 at

The Holiday Inn Express, Bickenhill Parkway, Birmingham B40 1QA

Kevin Bittlestone welcomed everyone to the meeting and introduced Professor Nick Shaw, Consultant Paediatric Endocrinologist at Birmingham Children's Hospital.

Dr. Shaw introduced himself. He is an Endocrinologist and he treats children with Fibrous Dysplasia and McCune Albright Syndrome, his specialty is the hormonal aspect of the disease. He works as part of a Multidisciplinary Team.

He gave a brief explanation of what Fibrous Dysplasia/McCune Albright (FD/MAS) is and how it occurs. It is an uncommon skeletal disorder which occurs in 1 in 15-30,000 people. He showed us a slide of Fibrous Dysplasia bone and normal bone and explained the difference. He also explained that the disease is caused by a faulty gene called GNAS. The gene mutation happens when the fetus develops in the womb so it is not inherited from our parents nor is it passed on to our children.

An explanation of how FD/MAS can present itself was given.

From an Orthopaedic point of view: A child could have a limp, bone pain, a fracture or a break. Sometimes it can be discovered by accident on an x-ray.

From an Endocrinologist point of view: A child could develop hormonal issues such as thyroid problems, precocious puberty, or growth issues.

From a dermatologist point of view - A child could present with cafe -au-lait skin pigmentations on their skin.

There was a diagram shown of how hormones work and all the different types of hormonal issues were explained. Not all of the hormonal issues occur at an early age.

Precocious Puberty - Girls with high levels of oestrogen may have early menstrual bleeding, early breast development or ovarian cysts. Initial observation should be given followed by a drug to stop these symptoms, until the child is around 12 years old when normal puberty should start. Not all cysts need to be removed, some will disappear by themselves.

Thyroid - Only 2/3 of patients with FD/MAS have abnormal thyroid levels. They will be placed on a hormone replacement medication with regular monitoring. Sometimes iodine treatment will be necessary or even thyroid removal. Thyroid monitoring should be continued throughout adulthood.

Excess Growth Hormone - This condition is less common and in only 21% of patients. It's nearly always associated with FD in the skull bones. Monthly treatment is required to control growth and monitoring.

Cushing's Syndrome - Only occurs in 4% of patients.

Phosphate Wasting - This can be treated with daily supplements and regular monitoring.

Skeletal Problems – Dr. Shaw explained the typical bones that can be affected by FD. An isotope scan should be undertaken to determine which bones are affected. Dr. Shaw explained what an isotope scan is and showed an example of a skull Isotope and a CT scan.

Monostotic FD - When only one bone is affected.

Polyostotic FD - When numerous bones are affected.

Panastotic FD - When the entire skeleton is affected.

A graph showed that the majority of fractures occur between the ages of 6 and 10 years old. There is a reduction in fractures as the patient gets older.

Scoliosis - This can occur in 50% of patients.

It is important that patients with FD in the skull bones are monitored yearly with vision and hearing tests. CT scans should also be carried out yearly to check for optic nerve compression.

Professor Shaw showed us examples of two of his patients. One child had encountered many fractures in various bones and we were shown x-rays of various bits of metalwork that were used to strengthen bones. The other child had encountered lots of rare complications with the heart and spleen.

There is a drug called Denosumab which is currently being trialed and may be used in the future if proved to be successful. It will hopefully show a reduction in growth of tumors and pain but it would need to be monitored throughout patient use.

Questions

What are the risks in fractures for Over 35's? - The patient would need monitoring when menopausal as the risk of fracture goes up and also there is the risk of Osteoporosis.

Will all areas of FD show on an Isotope Scan? - An isotope scan undertaken when the child is young should show up all areas of FD.

Dr. Shaw said that it is important that monitoring should be continued straight from childhood into adulthood so that treatment can be continued. A child should be referred to an adult Dr. once they are approaching adult age.

When using Bisphosphonates, a bone density scan should be done regularly to check bone density of all bones.

Kevin thanked Dr. Shaw for his presentation.

Jon and Mandy Price were introduced. They are Clinical Scientists who were giving a speech on Levels of Radiation in Diagnostic Treatments.

Jon and Mandy discussed all the different types of imaging that may be used - X-rays, MRI, CT, Ultra Sound and Isotope. Examples of each type of imaging was shown and explained. All images were taken from FD patients. The risks of radiation were shown on a graph and explained. Levels of radiation are low. There are natural levels of radiation around us as well in

some foods and rocks. Before a patient has images taken, a Radiologist will weigh up the levels of radiation involved to keep the patient protected.

Kevin introduced Dr. Juling Ong, Consultant Craniofacial & Paediatric Plastic Surgeon at Great Ormond Street Hospital for Children in London.

Dr. Ong gave a description of FD. He went on to discuss the importance of being treated from very early childhood right through to adulthood. He discussed the presentation of symptoms and the diagnosis of FD in the skull bones.

He showed us photos of severe cases of skull based FD where patients had not been diagnosed or treated early on in childhood. He explained some of the problems and secondary problems that skull based FD can cause if left untreated - breathing difficulties, double vision or blindness, problems with teeth and hearing issues.

A patient with FD in their skull bones needs a team of Dr's to look after their care. As well as a Craniofacial & Maxillofacial surgeon, the patient would also possibly need a Neurosurgeon, an Ophthalmologist and an Audiologist. There is a risk of bleeding when operating on the skull so close monitoring is needed.

Dr. Ong then went to discuss the different types of surgery that a patient may receive. Videos and photo images were used for all examples including extra cranial surgery in which bone graft and plastic implants can be used. Plastic implants are easy to remove. We were shown photos of implants used in reconstructive surgery. It is possible to remove bits of diseased bone and also replace them with live bone and blood supply from the patient's own hip/leg.

A surgeon will always try to mirror each side of the face to keep the patients appearance as normal as possible and images of implants used to adjust the eye position were shown. This is extremely difficult when re-growth appears but the patient should have regular scans and measurements taken. It is good to use 3D Photography as well. Every patient is different, FD varies so much and re-growth can occur.

Dr. Ong gave a detailed talk about optic nerve compression and why it is vital to have regular eye examinations.

The achievements of a Craniofacial Surgeon are:

To try and maintain a normal appearance for the patient.

To try and have minimal impact on the patient's life.

Check for secondary complications such as blindness and monitor them closely.

Kevin introduced our final speaker of the day, Dr. M. K. Javaid, Consultant Rheumatologist at The Nuffield Orthopaedic Centre in Oxford.

Dr. Javaid gave an introduction to the Rudy Study and then gave an update on what has been happening over the last year. The Rudy Study is made up of a team of Dr's, Researchers and Patients who come together to get a better understanding of pain in rare diseases. Patients are

asked to complete a number of questionnaires based on their pain. No one knows why FD is so painful. Patients can set up a profile page and complete the questionnaires there.

There is a new diagnostic history page on the website where patients can enter their diagnosis details. Now partners of FD patients can join in the study too. Patients can now join the forum too.

We have started asking patients to send us a copy of their clinic letters. We want to find out more details about the patients, where they have FD, what treatment and care they receive and what other health conditions or secondary conditions they may have. There is a skeleton diagram on the patient profiles where they can colour in all the bones that hurt.

Dr. Javaid explained that a patient pathway/guidelines needs to be put together to help Dr's understand the disease and give the correct treatment advice. These guidelines would provide information to Dr's in the UK on diagnosis, assessment, treatments and monitoring. Lots of Dr's and GP's have no experience in the disease and patients are not getting the correct diagnosis or the correct treatment that they need. Patients would be able to use these guidelines to help them get the correct treatment.

In the future there is the hope for research and trials for new painkillers for FD.

Dr. Javaid also has an ongoing research project with the James Lind Alliance. Three rare patient groups have come together with clinicians to decide which questions are the most important when dealing with a rare disease. They are in the process of putting together a survey where patients can write down which are the most important questions to them. The results will then be analysed.

Dr. Javaid also works alongside other Dr's in Europe and The USA.

Questions

When will the pathway/guidelines be ready? - One year. We are hoping to have a Dr's draft ready in the spring.

Do we know how many FD/MAS patients there are in the UK? - Not sure of the exact number of patients in the UK. We would need to look at GP research networks.

Are we getting better at diagnosis in the UK? - There is some improvement but a lot of work still needs to be done.

Can we get access to treatment and experienced Dr's? In some areas of the UK there is no treatment available in local hospitals and patients cannot be referred to other hospitals as they are too far away from where they live - That's what the pathway is for. We would like to see Centre's of excellence across the UK which will specialise in FD and hopefully be available for all patients.