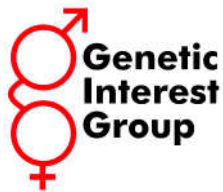


Acknowledgements.

We would like to thank all those who took part in the focus groups, interviews, and on-line survey and all who helped in the development of this guide.

The *Family Route Maps* project is funded by the charity Jeans for Genes and an unrestricted educational grant from Genzyme Therapeutics Ltd.



Genetic Interest Group (GIG)

Unit 4D, Leroy House

436 Essex Road, London, N1 3QP

Tel: 0207 7043141

www.gig.org.uk

Registered Charity No 1114195

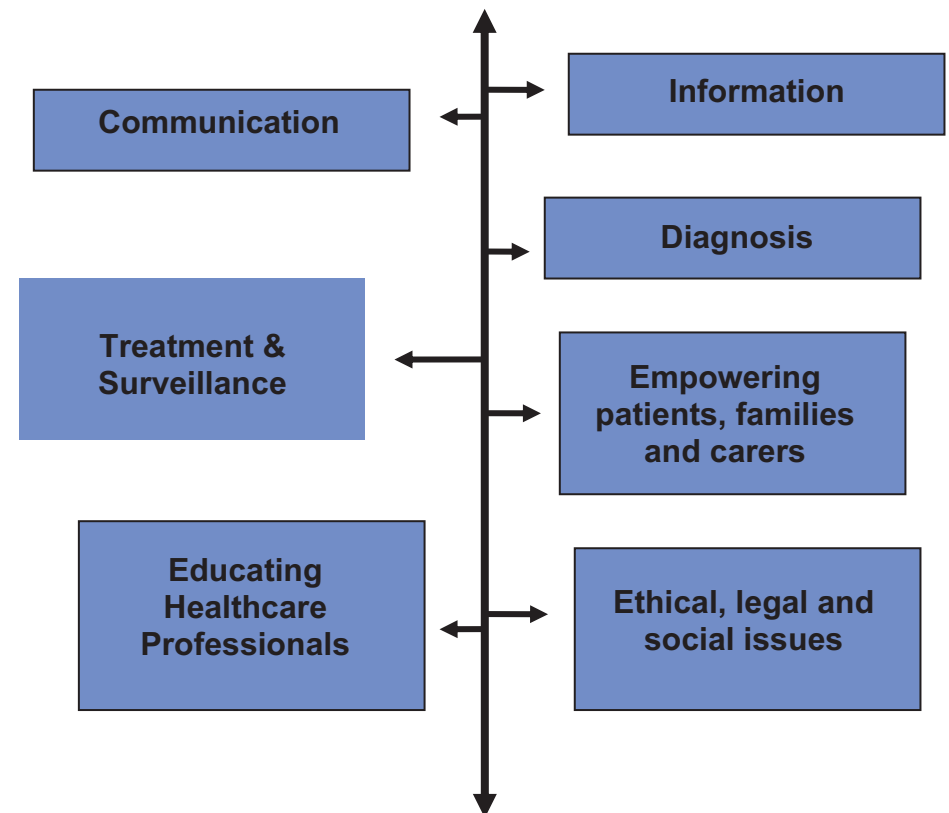
Company Registered by Guarantee 05772999

This leaflet has been prepared in good faith to provide patients with a guide to current services and information. Neither GIG nor NPS (UK) can be held responsible for the accuracy of the information it contains. Links to other organisations are included for information purposes only and are not recommendations from GIG or NPS (UK).

Nail Patella Syndrome

Family Route Map

This Family Route Map is a guide to current services and information.



INTRODUCTION

This leaflet is designed to provide signposts to sources of current information and appropriate services for patients, their families and carers, together with healthcare professionals. The issues and concerns raised during a series of patient focus groups in 2006 organised by the Genetic Interest Group (GIG) identified seven themes (see front cover) which have been used as the basis for developing this resource with the help of patients, families and carers, and clinical staff with experience and expertise of this condition.

Nail Patella Syndrome

Nail Patella Syndrome (also called Fong's Disease, Hereditary Onychoosteodysplasia ["HOOD"] or Turner-Kieser Syndrome) is characterised by several typical abnormalities of the arms and legs as well as kidney disease and glaucoma. It is a complex condition that varies greatly between each individual.

Nail Patella Syndrome UK is a registered charity working to promote awareness of the condition. The charity was officially registered in January 2003 and the main aims and objectives are to increase awareness and raise funds to allow the valuable research work to continue. Another very important aspect of their work is to host medical conferences every two years, bringing together healthcare professionals with expertise in Nail Patella Syndrome (NPS) and individuals and families living with the condition, providing a platform for sharing experiences and information. The charity has a website www.npsuk.org

You can contact the charity either by phoning **0800 121 8298** or by email to npsuk_info@yahoo.co.uk or by writing to:
PO Box 26415, East Kilbride, Glasgow, G74 1YW.

TIP

MedicAlert Foundation: www.medicalert.org.uk
MedicAlert ID emblems enable emergency medical staff to call a 24-hour helpline number to obtain detailed information on your medical condition, a summary of which is engraved on the emblem.

TIP

UK schools have a Special Educational Needs Coordinator (SENCO) whose role is to ensure every effort is made to tailor the individual needs, physical, emotional or educational, of a child within that school. You can ask to speak to the SENCO at your child's school any time.

Links

Other useful organisations:

Birth Defects Foundation www.bdfnewlife.co.uk Tel: 08700 707020

British Society for Human Genetics (BSHG): www.bshg.org.uk

Carers UK www.carersuk.org Tel: 0808 808 7777

Citizens Advice Bureau www.citizensadvice.org.uk

Contact-A-Family: www.cafamily.org.uk Tel: 020 7608 8700

Directgov www.direct.gov.uk provide a website with help on benefits and entitlements

Disability Benefits Office Tel: 08457 123456

Disablement Income Group Scotland www.digscotland.org.uk
Tel: 0131 555 2811

GIG (Genetic Interest Group) have a range of patient leaflets on their website www.gig.org.uk/eurogentest_patientleaflets.htm Tel: 020 7704 3141

International Glaucoma Association www.glaucoma-association.com
Tel: 01233 648170

Nail Patella Syndrome Worldwide www.nailpatella.org

Parents Centre www.parentscentre.gov.uk a website with resources and links to specialist help with education, money matters and more

The Family Fund www.familyfund.org.uk Tel: 0845 1304542

The UKGTN (UK Genetic Testing Network) has produced a patient leaflet www.ukgt.nhs.uk/gtn/UKGTN-information/Patient-leaflet.html

You're ABLE, Information, products and services for disabled people www.youreable.com has a website with Forums and Discussion

Wellchild www.wellchild.org.uk Tel: 0808 801 0330

OTHER INFORMATION

Insurance

It is common to have difficulties finding adequate and affordable insurance policies once diagnosed with a genetic condition. The following companies offer travel insurance for people with pre-existing conditions. There may be others available and this list should not be taken as a recommendation:

- The insurance group Banner - see www.bannergroup.com have a simple policy that anyone declared fit for travel by Interhealth, see www.interhealth.org.uk is covered as normal by them. They don't have extra rates for particular conditions.
- Free Spirit (AXA Insurance UK plc) www.free-spirit.com for people with medical conditions and/or disabilities
Tel: 0845 2305000, email: sales@pjhayman.com
- AllClearPlus (Groupama Insurance Co Ltd) www.allcleartravel.co.uk provides a special policy.
Tel: 08712 088500, email: info@bishops courtas.co.uk
- Freedom Insurance www.freedominsure.co.uk offer travel insurance. Tel: 01223 454290

Financial help

CAB (Citizens Advice Bureau) can help with advice locally about benefits and can be found through your phone directory.

Psychological help

Psychological counselling is not commonly offered on diagnosis or even later on. If you feel that you need extra help in coming to terms with your condition or any other aspect of your life, don't be afraid to ask for this via your GP.

Education

If you have concerns about Attention Deficit Disorder (with or without hyperactivity) possibly affecting your child or if they are experiencing other problems at school discuss this with your Community Paediatrician or ask for a referral to specialist services from your GP or Health Visitor.

Exercise

Care should be taken to ensure exercises are suitable for an individual with NPS, always check with your lead healthcare professional. The NPS Worldwide website has a list of suggested exercises www.nailpatella.org/workout.html

INFORMATION

Nail Patella Syndrome (NPS) is a genetic condition which affects the nails, knees, elbows and can cause iliac horns (bony protrusions on the pelvis bones). There are, however, many other features that may be seen in this condition and other body systems such as the kidneys and eyes may be affected. In the skeletal system, tendons, ligaments and muscles can be affected as well as bones. The severity of the features of NPS and how often each feature occurs, varies between one person with NPS and another, even within the same family. A person with NPS does not have to have all of the features of the condition.

NPS is inherited in an autosomal dominant manner, that is, the risk of transmission is 50% per pregnancy (just like flipping a coin). Approximately 20% of cases are sporadic, being the result of a new mutation (gene change).

The condition is found in approximately 1 in 50,000 of the population and occurs in all ethnic groups.

The Genetic Interest Group (GIG) website has a series of leaflets explaining more about inherited conditions and includes;

- 'Dominant Inheritance',
- glossary of terms used in genetics, and
- useful questions to ask when going for an appointment.

Please see www.gig.org.uk/eurogentest_patientleaflets.htm

Genetic Testing:

Genetic testing and counselling is available at 23 regional NHS genetic centres throughout the UK; please see the website of www.bshg.org.uk for a list and also in their section 'For Patients' there is an explanation of what Genetic Counselling is and is not. A referral to a genetic centre is usually made through your GP or specialist. Importantly, if you were seen several years ago in a Clinical Genetics Unit you may wish to be reviewed and in this case it might be possible to contact them directly yourself. There are a number of issues surrounding genetic testing particularly in relation to children and as such, many patients may wish to be seen and counselled by a consultant clinical geneticist as early on as possible.

DIAGNOSIS, TREATMENT AND SURVEILLANCE

It is very common for families to remain undiagnosed for several generations despite having been seen by doctors from a variety of disciplines. However, screening is recommended for all patients diagnosed with NPS.

Recommendations for the care of patients with NPS have been published by Sweeney et al in 2003 in the Journal of Medical Genetics. This includes:

- Annual screening for renal disease from birth.
- Screening for glaucoma in adulthood every 2 years.
- Before treatment such as surgery or intense physiotherapy is considered Magnetic Resonance Imaging (MRI) should be undertaken to determine abnormal anatomy.
- Genetic Counselling should be offered to all patients.

TIP

Genetic Counsellors (part of the NHS Clinical Genetics Services) can help with concerns around the impact on you and your family of being diagnosed with a genetic condition and may be able to give you information about where to go for support.

In pregnancy there is an increased frequency of preeclampsia (high blood pressure, swelling and proteinuria). Antenatal care should include surveillance by the adult nephrology (kidney specialist) team.

Contact Nail Patella Syndrome (UK) for further information on centres of expertise or clinicians who have experience of diagnosing and managing the condition: Tel: 0800 121 82 98.

In the UK, patients have the right to ask for a second opinion via their first specialist or through their GP. They are not, however, entitled to receive that second opinion if the clinicians do not think it necessary. A patient may appeal against any decision via their Primary Care Trust (PCT). For more information on the Referral Process please see: www.gig.org.uk/docs/referrals.pdf

INFORMATION FOR HEALTHCARE PROFESSIONALS

Information and articles about NPS are available from the NPS (UK) website www.npsuk.org/medical_info.html

or at the following websites;

GeneReviews www.genetests.org (One of the authors is Dr Elizabeth Sweeney, Clinical Geneticist, UK.)

OMIM — Online Mendelian Inheritance in Man www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=161200

Orphanet (European database) www.orpha.net
Free-access website providing information on rare diseases

The National Organization for Rare Diseases www.rarediseases.org

The Renal Unit of the Royal Infirmary of Edinburgh www.edren.org provides further information about the possible renal complications

Information about Education for Healthcare Professionals:

The NHS National Genetics Education and Development Centre www.geneticseducation.nhs.uk provide information and resources for healthcare professionals.

British Society for Human Genetics www.bshg.org.uk

Genetics in Family Medicine: The Australian Handbook for General Practitioners www.gpgenetics.edu.au

TIP

Say how you feel: if you don't want your picture taken; or if you don't want medical students participating in your appointments; or clinical staff discussing your child in front of them, don't be afraid to say so.