Hypomelanosis of Ito patient information booklet

HITS (UK)
Family support network

Genetic Alliance UK
Supporting. Campaigning. Uniting.

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Introduction

From August 2009 to March 2011 Genetic Alliance UK held a number of consultations with members of the patient support group for Hypomelanosis of Ito (HI) in an effort to identify the core issues and concerns they experienced as a result of living with the condition. The wealth of patient's experiences shared during this time were later analysed to see where gaps in information lay and how best to address these shortcomings.

Informed by this patient-led research, this booklet has been designed to help patients and families affected by Hypomelanosis of Ito (HI) to useful resources relating to health and social care assistance. By way of empowering individuals and families with this information, we aim to improve the quality of life for those affected by the condition.

Information

HI (previously referred to as Incontinentia pigmenti achromians) is a rare condition that is caused by a chromosomal (genetic) change. Although caused by an underlying genetic alteration, HI is not an inherited disease and is not gender-specific. It is characterised by hypopigmentation* (lighter areas) of the skin, neurological problems and frequently associated with asymmetric abnormalities in other organs.

HI is classified as a neurocutaneous* syndrome i.e. a genetic condition which may affect both the skin and brain. Patients with HI have areas of skin lightening (hypopigmentation*), often appearing as unusual patterns such as whorls on the trunk of the body and streaks on the limbs called Blaschko's lines*. This occurs because genes* controlling skin pigmentation are disrupted by the chromosome* change, and this can cause a reduced number of melanocytes*, the cells that make skin pigment called melanin.

The reason that only some areas of skin are lighter is because the chromosome* change only affects a proportion of the cells in the skin and the body. This difference in cells which have the genetic change compared to the remaining cells which are normal is called *mosaicism*, rather like a mosaic which is made up of differently coloured tiles. For this reason HI has also been referred to as Pigmentary Mosaicism Hypopigmentation*, and it may be present from birth, but in 70% of cases it appears within the first year of life.

Sometimes patients may have areas of hyperpigmentation (darker areas of the skin) or areas of both hypo- and hyperpigmentation. This same chromosomal change also causes the neurological and other associated disorders. The most common neurological problems are epilepsy* and learning difficulties, whilst other associated physical

features of the condition include cardiac (heart) abnormalities, asymmetry of the body, scoliosis* (curved spine) and other bone changes, dental problems, craniofacial abnormalities (e.g. cleft palate), kidney and ophthalmic (eye) problems.

A table summarising some of the organ specific problems that are associated with HI can be found in the table below. Please note that this list is not exhaustive.

Organ System	Possible problems seen in HI
Musculoskeletal	Cleft lip or palate*, limb asymmetry/defects, scoliosis, facial disfigurement, joint laxity*, finger/toe abnormalities (such as brachydactyly*, syndactyly*), torticollis*
Heart	Heart valve problems, conduction defects (heart rhythm problems), complex congenital heart diseases
Neurological	Epilepsy*, autism*, learning difficulties, dyskinesia (abnormal movements), microcephaly (small head), macrocephaly (large head), hypotonia (low muscle tone)
Ears	Ear asymmetry, deafness, pre-auricular pit (dimple)
Gastrointestinal	Gastro-osephageal reflux* (acid reflux), Inguinal/umbilical hernia*, intestinal atresia*
Kidneys	Horsehoe-shaped kidney, unilateral agenesis (absent kidney), glomerulocystic disease, hydronephrosis (enlarged kidney)
Skin	Streaky or whorled hypo- or hyperpigmentation
Hair	Colour variation, sparse hair, hypertrichosis (increased body hair)
Nails	Hypoplasia (small nails), anonychia (absent nails), ridging
Teeth	Caries (decay), hypodontia (small and absent teeth), enamel defects

Diagnosis

Due to the wide range of organ systems Hypomelanosis of Ito can affect, diagnosis is often the result of multidisciplinary examination. Dermatologists, neurologists and geneticists are typically the first to make an initial diagnosis, followed by a specialist in the area of the patient's specific symptoms.

Diagnostic criteria include patterned hypopigmentation* (Blaschko's lines*) in conjunction with neurologic deficits plus another major symptom, but clinicians do not universally agree upon this. A Wood's lamp is commonly used in the first step of identification by shining it on the skin lesions*. Blood karyotypes* are the most useful test for determining if the patient has HI, but biopsy* karyotypes and other imaging tests can also be performed to aid in confirmation. However, it is important to note that that there is no single definitive test for HI and that some tests might return inconclusive.

Genetic testing

Genetic testing may be done for HI, but only to confirm its diagnosis rather than to affect treatment. HI is a sporadic genetic condition, which means that it is caused by a chromosome* (genetic) change, which occurs during embryonic development. It is, therefore, not an inherited disorder, so it cannot be passed on to other family members.

A blood test to assess the chromosomes for a mosaic abnormality (also called a blood karyotype*) is the most useful and definitive test for determining if a patient has HI, but skin biopsy* and other imaging tests may also be necessary to aid in confirmation.

A clinical geneticist can provide support and help coordinate the appropriate patient-centred care amongst the various specialists who may be needed. Information about genetic testing, genetic counselling and clinics can be found at www.genetests.org and a directory of genetic centres is provided at www.geneticalliance.org.uk/services.htm.

Prognosis

The impact of HI on the individual patient can vary dramatically depending on the severity of the case and the associated disorders. Prognosis is dependent on the presentation of the individual patient's symptoms. In majority of cases, hypopigmentation of the skin is a consistent symptom.

NOTE: Descriptions explaining what each of these heath care professionals do can be found in the glossary.

Treatment

Currently there is no treatment or drug therapy available for patients with Hypomelanosis of Ito, so emphasis is placed on managing the associated symptoms. Due to the great variability of features associated with HI, condition management must be specifically tailored to each patient.

For patients with associated epileptic seizures, anticonvulsant* medicine is a common therapy. There are some other abnormalities that can be surgically improved or corrected depending on the type and severity. Some examples of surgically reparable conditions include cleft palate/lip, cataracts and musculoskeletal* deformities.

Although there is currently no treatment specifically for the hypopigmentation of the skin in HI patients, research is ongoing and there are cover-up cosmetics available to help even-out the skin tone.

NOTE: It is important for HI patients to seek support from specialists in the area of their associated conditions. This might include referrals to the following healthcare professionals:

Audiologists	Chiropractors
Dermatologists	Dentists
Dieticians	Neurologists
Occupational Therapists (OT)	Ophthalmologists
Orthopaedists	Psychiatrists
Paediatricians	Physiotherapists
Speech and Language Therapists (SALT)	

Patients, families and carers

Patient Support Groups

Support groups are useful tools in helping patients and families learn how to successfully and holistically manage HI. They provide a community sharing similar experiences that can help the members cope with the physical and emotional challenges that can often accompany the diagnosis of this condition. These groups can also provide a network for sharing resources and making referrals.

HITS (UK) Family Support group was established in 2001 by families for families. Their aim is to provide mutual consolation and assistance by uniting families affected by Hypomelanosis of Ito through telephone, email, letter, internet chat rooms and newsletters. Their goal is to create a network that offers a wide variety of support and information for patients, families, carers and healthcare professionals. The group can be found at:
www.e-fervour.com/hits Tel: 01803 401018

The Ectodermal Dysplasia Society facilitates fundraising and research while providing networking opportunities and medical references to patients impacted by genetic conditions that affect the normal function of skin, hair, nails and/or teeth.

www.etcodermaldysplasia.org Tel: 0124 226 1332

Unique operates email and telephone help lines to connect families with rare genetic diseases and healthcare professionals.

www.rarechromo.org Tel: 0188 333 0768

Other Useful Support Groups

Action for Children is an organisation designed to help families with disabled children and offer Children's Centres and parenting programmes.

www.actionforchildren.org.uk Tel: 0300 123 2112

Contact A Family supplies advice, support and referrals for families with children affected by any type of disease or condition.

www.cafamily.org.uk Free Helpline: 0808 808 3555
Tel: 0207 608 8700

The Parent Centre supplies support and information for parents in relation to supporting their child, particularly those affected by disease.

www.theparentcentre.co.uk Tel: 0131 664 5388

Well Child is a paediatric support network for families with chronically sick children. They have information on symptom management methods and clinician referrals.

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

You're Able is an online forum for disabled patients and families with resource information on online support groups.

www.youreable.com

Carers

Some patients may require the aid of a carer depending on the severity of their HI and associated symptoms. Carers can also benefit from additional information and support in providing the best care for the affected individual.

Carers UK is a support group that provides information, resources, networking opportunities and professional advice for all types of carers.

www.carersuk.org Tel: 0207 378 4999

The Princess Royal Trust for Carers is a resource for carers that guides them through the process of finding appropriate services and understanding legislation governing their entitlements.

www.carers.org Tel: 0844 800 4361

HDS trains and provides specialised carers for homecare and community support for all types of disabled adults and children.

www.hdsservices.co.uk Tel: 0208 518 6164

Education

Patients with learning difficulties associated with Hypomelanosis of Ito, especially children, may have special educational needs (SEN) and require additional support. Teachers of the Deaf, Qualified Teachers of the Visually Impaired and SEN coordinators may play an integral role in supporting your child's learning.

Every school within the UK should have a Special Education Needs Coordinator (SENCO) who is responsible for advising parents and teachers about suitable programmes that will meet the child's specific educational and physical needs.

Further details about the role of a SENCO and how to access this service can be found at:

www.teachersmedia.co.uk/SEN.

Additional Education Resources

National Association of Special Education Needs promotes and provides education, training and developmental support to those with special education needs, particularly children. They operate in nearly all UK schools.

www.nasen.org.uk Tel: 0182 731 1500

National Portage Association Portage is a home-visiting educational service for pre-school children with additional support needs and their families.

www.portage.org.uk Tel: 0121 244 1807

SEMERC is a member-based charity offering networking, information and support for families of children with special needs. Special Needs covers a broad spectrum including learning/education, physical and behavioural difficulties

www.specialkidsintheuk.org

ACE (Advisory Centre for Education) is an organisation that provides advice and support to parents and carers with special needs children attending state-funded schools.

www.ace-ed.org.uk Tel: 0808 800 5793

Network 81 is a group that focuses on promoting the educational needs of disabled children, assisting parents in getting the necessary educational support for the children and linking families with similar needs together.

www.network81.org Tel: 0845 077 4055

IPSEA is a charity that provides assistance and advice to parents whose children have special educational needs. They offer a helpline and a tribunal support service.

www.ipsea.org.uk Tel: 0800 018 4016

Parents for Inclusion is a network of families and parents with children who are disabled and/or have special educational needs. They link families for support and offer a helpline.

www.parentsforinclusion.org Tel: 0800 652 3145

Financial assistance

Depending on the neurologic impairments and severity of their associated symptoms patients and families with HI may require many hospital visits and procedures throughout the patient's lifetime. Managing HI can be financially stressful, but there are financial aid services available to provide support.

The Citizens Advice Bureau (CAB) has information about these services and can also offer advice regarding welfare benefits, taxes and debts.

www.citizensadvice.org.uk Tel: 0844 477 2020 (Wales)

Tel: 0844 411 1444 (England)

Turn2us is a not-for-profit organisation that helps people in gaining access to grants, welfare benefits and other financial aid.

www.turn2us.org.uk Tel: 020 8834 9200

The Family Fund is a charity that provides grants to low-income families with disabled and severely ill children. They also help families obtain household items and other necessities.

www.familyfund.org.uk Tel: 0845 130 4542

Insurance

For patients with rare genetic conditions, finding appropriate and sufficient insurance can be difficult, but there are life and travel insurance options available. Some companies offer alternatives to Life Cover, but it is important to investigate several options to find the policy that best suits your family.

At Genetic Alliance UK we have worked closely with insurance companies to help make insurance for those affected by genetic conditions more affordable. The two companies we have worked with are Insurancewith and Allclear who offer travel and life insurance respectively.

We have found that they are helpful and understand most issues surrounding insurance for those with genetic conditions, although we would always recommend having a look at other companies and policies to find the policy that is right for you before buying insurance.

Further guidance relating to insurance concerns can be found at www.geneticalliance.org.uk/insurance.

Insurance Groups

Insurancewith is focused on helping families and individuals with severe medical conditions secure travel insurance at the best possible rates.

www.insurancewith.com Tel: 0845 2307 159

Allclear is an insurance company specifically created to help those both serious medical conditions purchase life insurance at a very competitive rate.

www.allclearlife.co.uk

Free Spirit is a travel insurance agency designed to meet the needs of individuals with medical conditions.

www.free-spirit.com Tel: 0845 230 5000

Additional resources

Genetic Alliance UK is the national charity of over 140 patient organisations, supporting all of those affected by genetic conditions. We do this by supporting patient groups, campaigning/raising awareness and uniting groups and individuals with genetic conditions.

www.geneticalliance.org.uk Tel: 0207 704 3141

National Education and Development Centre is an NHS organisation that supports awareness, understanding and education regarding genetics and genetic conditions. It is a useful resource for understanding the causes and development of HI in greater detail.

www.geneticseducation.nhs.uk

Skin Care Campaign is an organisation that focuses on raising awareness and improving healthcare for individuals with skin conditions and diseases.

www.skincarecampaign.org

Rare Disease UK was established by Genetic Alliance UK together with other key stakeholders. As the national alliance for people with rare diseases (and all who support them) it campaigns for communities of patients and families suffering from rare diseases, and provides them

with support and care networking opportunities.

www.raredisease.org.uk Tel: 0207 704 3141

Glossary

Anticonvulsant: Medication designed to control abnormal electrical or neuronal activity in the brain that characterises a seizure.

Audiologist: A specialist clinician who diagnoses and treats patients with hearing impairments.

Autism: A disorder of neural development characterised by impaired social interaction and communication, and by restricted and repetitive behaviour. These signs all begin before a child is three years old.

Biopsy: The removal of a small sample of tissue from a patient for close examination and testing during the diagnostic process.

Blaschko's Lines: Skin lines or stripes that become visible when skin diseases manifest themselves and are caused by migrating embryonic cells; they can appear as a lack of pigmentation or as a darkening of the skin.

Brachydactyly: Shortening of the fingers or toes due to underdevelopment of the bones in the hands or feet

Chiropractor: A health care professional who diagnoses, treats and helps prevent musculoskeletal problems, especially focusing on the spine.

Chromosome: Structures found in the nucleus of cells composed of DNA and proteins. Normally humans have 46 chromosomes in each cell, 23 from each parent. Of these, 22 are autosomes and 1 is a sex chromosome.

Cleft lip palate: A separation or split in either the upper lip or the roof of the mouth (palate) or sometimes both.

Dermatologist: A specialist clinician who diagnoses and treats conditions affecting the skin, including the scalp and nails.

Dietician: A health care professional who focuses on proper food and nutrition in order to promote good health.

Epilepsy: A disorder characterised by abnormal electrical discharges in the brain that typically present as seizures.

Fetal development: The growth of a human embryo or fetus into an infant child in its mother's womb.

Gastro-oesophageal reflux: A condition where the lower oesophageal sphincter (the muscular ring at the lower end of the oesophagus) is abnormally relaxed and allows the stomach's acidic contents to flow back or 'reflux' into the gullet (oesophagus).

Gene: The fundamental physical and functional unit of heredity. A gene is an ordered sequence of DNA located in a particular position on a particular chromosome that encodes a specific functional product (i.e., a protein or RNA molecule).

Geneticist: A specialist clinician who diagnoses and treats patients affected by hereditary and genetic conditions.

Hyperpigmentation: The darkening of an area of skin or nails caused by increased melanin production by melanocytes

Hypopigmentation: The loss of skin colour or pigment due to a lack of melanin production by melanocytes.

Inguinal hernia: Occurs when part of the bowel (part of your digestive system) pokes through your lower abdomen into the groin (the area at the front of the body where the thighs meet the abdomen).

Intestinal atresia: The failure of a portion of the intestinal tract to completely form. It occurs most frequently in the ileum (lower part of the small intestine)

Joint laxity: A lack of stability in a joint

Karyotype: A photomicrograph of an individual's chromosomes arranged showing the number, size, and shape of each chromosome type; used in low-resolution physical mapping to correlate gross chromosomal abnormalities with the characteristics of specific diseases.

Skin Lesion: Any abnormal skin tissue; in HI patients, this term refers to the lines and patterns of hypopigmented skin.

Melanocyte: A skin cell that produces melanin, an important colour pigment in the skin.

Musculoskeletal: Having to do with both the muscles and the skeleton/bones.

Mutation: A spontaneous, permanent change in the DNA sequence of a gene.

Neurocutaneous: A disease or syndrome affecting the skin and nervous system.

Neurologist: A specialised clinician who diagnoses and treats conditions affecting the nervous system, which includes the brain, spinal cord and nerves throughout the body.

Occupational Therapist: a specialist therapist that aids physically and/or mentally disabled patients perform and maintain daily life activities.

Ophthalmology: A clinical specialty that is concerned with the structure, function and treatment of the eyes.

Orthopaedist: A specialized clinician and surgeon who diagnoses and treats disorders impacting the musculoskeletal system.

Otolaryngology: A clinical specialty that is concerned with the structure, function and treatment of the ears, nose and throat; clinicians are sometimes referred to as ENT clinicians.

Physiotherapist: A specialist therapist that treats patients with physical impairments and disabilities.

Special Education Needs Coordinator (SENCO): The coordinator in collaboration with the head teacher and governing body, plays a key role in determining the strategic development of the SEN policy and provision in the school in order to raise the achievement of children with SEN.

Scoliosis: A medical condition in which an individual's spine is curved from side to side; it is a musculoskeletal disorder.

Syndactyly: A condition where two or more fingers/ toes are fussed together

Torticollis: Means 'twisted neck'. The most common cause is acute torticollis, often called 'wry neck'. This is a common cause of neck pain and stiffness.

Umbilical hernia: Occurs when fatty tissue or a part of the bowel pokes through the abdomen near the navel (belly button)

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To find out more information on this project please go to www.geneticalliance.org.uk/projects/facilitatingnetworks.htm

Or contact:

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www.geneticalliance.org.uk

HITS (UK) patient support network

www.efervour.com/hits

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