International Genetic Alliance

2009

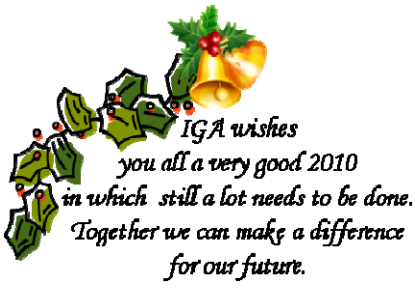
For members and interested parties

 # 7

"seeks a world where genetic conditions are understood, prevented, treated, ameliorated, and cured"

INTRODUCTION

Over the last few months IGA has been involved in a number of international summits and conferences such as on birth defects and disabilities in New Delhi, on progress in life sciences, BioVision in Lyon, on the evolution of medicine in Alexandria, on integration of health and healthcare into societal and political agendas in Berlin. The voice of the patient community was aired in all of these conferences. However - with the exception of the Delhi conference - the invitation to patient representatives for these conferences was limited to one or two persons. This is a denial of the stakeholder status of the patient community. Patient alliances have become a catalyst for spreading excellence e.g. in areas of research and health services. Their role has expanded from the provision of peer to peer support to encompass the stimulation and facilitation of research and development, the development of family-friendly health and social care services and awareness raising amongst the wider public. Increasingly, patient and parent organisations have a role to play in the formulation and implementation of policy at local, regional, national and international levels. Recognition as a vital stakeholder and participation in summits and top conferences on health and healthcare issues should be a matter of course. In the international conference in Delhi on "genetic disorders and disabilities in the developing world" the patient community was an official partner. Patient representatives were well represented in the program and in the audience. It was then and there that IGA launched a declaration on the issues which was published in an extra edition of this newsletter.



EDITORIAL

Written by: Jo-Anne Richards of Southern African Inherited Disorders Association (SAIDA) and Dr. Jai Rup Singh from India

In October 350 participants from 40 countries came together in New Delhi in an attempt to use the best the world can offer to benefit the people with birth defects and disabilities in the developing world.

The International Conference of Birth Defects and Disabilities in Developing countries brought north and south together to explore options for translating research from all over the world into better care and prevention of birth defects, particularly in developing nations.

As the deadline of 2015 approaches, nations are wrestling with how to achieve the Millennium Development Goal of a two-third reduction in childhood mortality. The World Health organisation is preparing a submission to its World Health Assembly, for it to acknowledge the global health significance of congenital disorders and for countries to prioritise services for their care and prevention.

In this spirit, delegates from medical and scientific communities, non-profit organisations and patient- parent bodies came together to share knowledge and experience for the benefit of all. During the pre conference, that was held one day before the main conference, 600 participants attended the several workshops at the All India Institute of Medical Sciences in New Delhi.

Reports on practical collaborations demonstrated how developed and developing worlds can work together to use technology more appropriately, to provide sustainable healthcare in areas that need it most.

Networking between those affected by genetic disorders and doctors and scientists enabled the transfer of knowledge and the

development of projects that promote capacity building in low and middle income countries. Their discussions moved the world closer to the development of an international set of basic quality standards.

The IGA was a strong stakeholder and supporter of the conference, forming the nucleus for patient- parent bodies to demand their right to be heard on policy and practice in health research and services.

With opinions drawn from around the world, the IGA was able to create a declaration, presenting eight topics of urgent interest. A copy of this declaration can be found in the Special Edition #6 of the IGA Newsletter.

The declaration demonstrates how the IGA has emerged as a force for change. The declaration aims to spread excellence and demand that patient-parent bodies have their say in healthcare and social services.

United under the IGA banner, patient-parent organisations have become far more than groups providing support to their peers. In response to global influences and local needs, their role has transformed over the past years.

These groups now facilitate research and the development of family-friendly health services. They raise awareness and have a role to play in formulating policy – not just at local, regional and national levels, but internationally as well.

The IGA will continue to build and support these functions among its members, as those most affected by genetic disorders begin to take their rightful place as important stakeholders within the health community.

UPCOMING EVENTS

International Federation of Human Genetics Societies (ICHG/ASHG 2011), Montréal, Quebéc, Canada, 11-15 October 2011

The International Federation of Human Genetics Societies (IFHGS) was founded in 1996. It is an international collaboration of professional human genetics societies. The IFHGS ensures that an International Congress of Human Genetics will take place every five years and that a credible forum for the sharing of ideas about human genetics is provided. "The purpose of The International Federation of Human Genetics Societies is to provide a forum for organized groups dedicated to all aspects of human genetics, including research, clinical practice, and professional and lay education. The Federation will enable communication between its member groups and encourage interaction between workers in genetics fields and in related sciences and will make itself available to promote meetings and publications and other forums which support human genetics research and practice."
For more information visit:
www.ichg2011.org

BioVision Alexandria 2010 – New Life Sciences: Future Prospects, Alexandria, Egypt, 11 – 15 April 2010

The Bibliotheca Alexandria is organising its Fifth International Biennial Conference. The theme of BioVisionAlexandria 2010 will be New Life Sciences: Future Prospects. It will aim to identify and explore the new frontiers and new areas in life sciences that will vastly serve humanity and provide hope for solving the world's most pressing issues. Theconference will focus on three major themes: Health, Food and Agriculture, and Environment. Each of these themes will be addressed by representatives of the greatest minds in industry, science, policy-makers and civil society fields. For information look at: www.bibalex.org/bva2010

REPORTS

The 4th International Conference on 'Birth Defects & Disabilities in the Developing World' was held from 4 – 7 October 2009 in New Delhi, India.

Birth defects or congenital anomalies are the leading cause of death in the first year of life and are also responsible for large numbers of foetal deaths. In the first detailed global

study of the problem (2006) it was reported that annually about 6 percent of the newly-borns worldwide are born with a serious birth defect of genetic or partially genetic origin. Additionally, hundreds of thousands more are born with severe birth defects due to maternal exposure to environmental agents. About 3.3 million children die from birth defects each year while another 3.2 million endure grave disabilities.

Equity in healthcare requires that he burden of birth defects on the individual, family, and community be recognized. Since a large per cent of those born with birth defects are born in medium and low resource countries; the emphasis has to be on optimizing their circumstances. However, the appropriate treatment of those with significant birth defects makes heavy demands on resources. Medical genetics must therefore be integrated into existing healthcare systems to offer counselling. This may lead to prevention of the birth of infants with defects and disabilities that prevent them from realizing the full potential of their lives. There is also the pressing need to develop cheap and reliable diagnostic techniques as well as drugs for a wide range of genetic disorders.

The aim of this conference was to help initiate international linkages for strengthening research and help raise awareness about the causes of birth defects and ways of preventing/treating these. Many scientists, physicians and patient organisations from all over the world were present to share experience and expertise. The International Genetic Alliance (IGA) and the World Alliance of Neuromuscular Disease Associations (WANDA) were also present at the conference. The WANDA had prepared a pré-conference workshop at The All India Institute of Medical Sciences in New Delhi. This workshop was dedicated to *'Muscular Dystrophy – Patient's Roadmap to Treatment'* where representatives of patient organisations addressed the issues involved in finding a treatment. At this moment some promising treatments for neuromuscular diseases like Duchenne Muscular Dystrophy, Spinal Muscular Atrophy and Friedreichs Ataxia are being investigated in clinical trials. Many international collaboration networks, such as the TREAT-NMD network, are involved in finding these treatments. A presentation on Pompe disease, a rare genetic neuromuscular disease, for which now a treatment exists showed that international collaboration among patient organisations is of huge importance to make treatment available for as many people as possible. The International Genetic Alliance was prominent present during the conference. At a well attended session on *'The role of parent/patient organisations in care and prevention of genetic and congenital conditions'* during the conference, members and affiliates of IGA from Brazil, South Africa, China, Australia, UK and the Netherlands told about their experience and contribution to prevent and care for genetic and congenital conditions. Most of the presenters were women who from their own personal experience, because either their child is affected or the woman herself is affected by a disease, show that one active, dedicated parent or one patient can make a difference to a large group of people. This is exactly the strength patient organisations can combine in themselves: personal involvement, dedication, commitment,

Representatives of patient organisations, from left to right: Allan Bretag (Australia), Martha Carvalho (Brazil), Jo-Anne Richards (South Africa), Irma Nippert (Germany), Arnold Christianson (South Africa), Dianne Petrie (Australia), Alastair Kent (UK), Ysbrand Poortman (the Netherlands), Sanjana Goyal (India) and Maryze Schoneveld van der Linde (the Netherlands).

knowledge, expertise and power. Since the aim of this conference is to help initiate international linkages for strengthening research and help raise awareness about the causes of birth defects and ways of preventing and treating these, the IGA wrote a declaration on 'Genetic & Congenital Disorders'. This declaration has been signed by the several patient organisations from all kinds of countries and continents that participated actively at the conference. The International Genetic Alliance invites your organisation to sign the declaration as well and to ask your colleague patient

organisations to do the same. This declaration gives us strength and shows the outside world that we do use our strength jointly to support the people we work for.

For signing the declaration, please contact: Ysbrand Poortman: landfort@tiscali.nl or Maryze Schoneveld van der Linde: maryze@pacesworld.com

World Health Summit in Berlin on Health and Human development, October 2009, Berlin, Germany

Mid October 2009 there was a World Health Summit organised in Berlin by the universities Charite (Berllin) and Descartes (Paris) and patronized by Angela Merkel and Nicolas Sarkozy.

The Summit was attended by 700 prominent opinion leaders from politics, economy, science and civil society coming from more than 60 countries.

There were 2 or 3 patient representatives of which one was on the program as a speaker in the one session that involved patients "Patients' needs and health research". The subject was: "Patients as (potential) partners in disease prevention and management". The summit covered many subjects but lost credibility by almost completely ignoring the patient voice. Also the problems of the medium and low income countries could have been given a much more explicit place. It was recommended to found a forum for innovative healthcare provision models in low-wage countries. There was also a plea for international exchange of success factors in healthcare system design. The "regulation density in research" was mentioned as a catastrophy for health research. The Lancet will publish a special WHS- edition while the WHS- organisers will publish a comprehensive report available early next year. The summit led to a decision to have an annual summit in October driven by an alliance of 8 universities (Berlin, Paris, Kyoto, London, Peking, Baltimore, Melbourne, Moscow).

Rare Diseases and Public Policies Conference, 2 November 2009, Buenos Aires, Argentina

On Nov 2nd 2009, the conference on "Rare Diseases and Public Policies" was held in Buenos Aires. The conference was organized by the Health Commission of the Chamber of Representatives, of the National Congress of Argentina. Fundación FOP collaborated in the organization of the event. Patient and parent organizations were invited to participate, as well as health professionals and policy makers. The main objective of the conference was that stakeholders and other people involved in the issue of rare diseases, make and discuss ideas for a bill proposal aimed to improve health and social services to improve the quality of life for patients and their families living with rare diseases in Argentina. The attendance was approximately 100 people.

Lic. Leonardo Gorbacz, Dr Victor Penchazadeh during the opening of the conference.,

Leonardo Gorbacz, representative of the Chamber and author of one of the bill proposals on rare diseases, gave the welcome to the audience. In order to make a contribution to the later discussion of the attendees, the conference began with an academic table with the participation of: Dr. Victor Penschazadeh, Professor of Genetics at University of Columbia, USA and Counselor of International Health Organizations; Dr Luisa Bay from the Argentine Pediatrician Society; Dr Michelini, Director of Maternity and Infancy from Chaco, and in representation of Fundación FOP, Arq. Moira Liljesthröm, who is also patient advocate and Dr Romina Armando. Arq. Liljesthröm & Dr. Armando presented their preliminary results of a research project on rare diseases in Argentina that Fundacion FOP is conducting. This study is being funded by a grant from the National Ministry

of Science, Technology and Productive Innovation.

The main objectives of this research project was to identify the specific problems suffered by people and their families dealing with rare diseases in different social and regional backgrounds in Argentina, in an effort to support well based policies, and to improve the quality of life for people affected. The research objectives were also to identify and provide valuable information to people while focusing on the best means to empower them to become advocates for themselves, through developing their capabilities in organization, interchange of information and communication. In order to achieve the objective to better understand the main problems that emerge from living with a rare disease the research team performed interviews with 322 families affected by 12 selected rare diseases throughout Argentina. During the conference both researchers also showed preliminary results of an on-line survey answered by 950 physicians. This survey asked doctors about, their knowledge of rare diseases, their practices of consulting when they are not able to diagnose by themselves, how they think doctors could get to know more on these pathologies and their knowledge and opinion on parents/ patients organizations.

After the opening session, the audience's attended three different workshops to discuss proposals related to access to diagnosis, treatment and information on rare diseases. Some of the new proposals that emerged from the discussion at the workshops, were added to the bill.

The event showed to be a very good and democratic way to outline public policies with the participation of stakeholders and other people involved in the issue of rare diseases. On November 25 the bill was treated in the Chamber of Representatives and was approved. Hopefully next year it will be treated in the Chamber of Senators to its definitive approval.

Moira Liljesthröm, Fundación FOP, Argentina
www.fundacionfop.org.ar

Official Launch EuroGenGuide, 27 November 2009, Warsaw, Poland

On November the 27th EuroGenGuide was officially launched to the public with a conference on EuroGenGuide – 'Patient Led Education and Development for Genetic Testing in Research and Medicine'. At the meeting around 55 people representing patient organisations, companies, politics and science were present. They came from countries like Poland, Russia, Belgium, The Netherlands, UK, Germany and Italy.

Representatives of organisations, from left to right: Alastair Kent (GIG), Alex McKeown (GIG), Kim Wever (VSOP), Georges Veckmans (Rare Disorders Belgium), Michael Livingstone (Heart EU), Genevieve Pierquin (Rare Disorders Belgium), Elia Casati (Policlinico Milan), Domenico Coviello (Policlinico Milan), Selena Freisens (CEEGN), Svetlana I Karimova (Rare Diseases Russia). In the front: Maryze Schoneveld van der Linde (WANDA).

Genomic research offers many new opportunities in the treatment of genetic diseases, which have become one of the greatest challenges of modern medicine. However, information about new and innovative treatments do not currently reach all patients and health professionals in Europe. EuroGenGuide, a platform for information and knowledge about genetic disorders and treatments for them, has been created in order to contribute to solving this problem. Genetic disorders are relatively rare: they affect fewer than 5 people in 10 thousand. Yet affecting millions of people around the world. Over 30 million people suffer from rare genetic disorders in Europe alone. In Poland, between 1.3 and 2.6 million patients have a genetic condition, with most of these being young children. Their parents often have difficulty in finding help and the right information. Physicians too need support to know how and where they can help their patients get the treatment that they need. A considerable number of cancers can also be caused by genetic abnormalities. 1 in 3 people will develop cancer at some point in their life. This number could be reduced with a better understanding of the genetic causes of cancer, and the relationship between

environmental factors and genetics. The main task for EuroGenGuide is to provide comprehensive, professional information on diagnosis and treatment of genetic disorders in Europe. This will allow patients and their families to make free and well-informed choices about their treatment, and will enable specialists to help those in need in the most appropriate way. EuroGenGuide has been helping people since 2008. As many as 76% of the users claim that the website allows them to find the information they need and 73% are of opinion that the information they have found is presented in an understandable way. Until April 2009 the website has been visited by 30,222 users a month. The project is coordinated by Genetic Interest Group, the national charity for all those affected by genetic disease in the UK. The EuroGenGuide team consists of various organisations of patients and parents, physicians, scientists, researchers and biotechnologists from all over Europe. The European patients' contribution to the project has considerably improved the level of knowledge in all aspects of human genetics.

It is of great importance for European communities to increase the efficient detection and treatment of genetic disorders, an to ensure the application of proper diagnostic techniques such that students of medicine in future receive better education in the field of genetics.
EuroGenGuide: www.eurogenguide.org.uk

SUPPORT NEEDED

The Washington-based **Genetic Alliance**, together with **Oregon Health and Science University (OHSU)**, are currently conducting an online study that hopes to lead to improved medical care for families with genetic diseases. The survey, which takes 10-15 minutes to complete, asks members of the public with an interest in genetic disease a number of questions about the responsibility for sharing genetic-risk information within families-and, particularly, when it is or is not morally acceptable to share such information. The link to the survey can be found below:
www.surveymonkey.com/s.aspx?sm=C_2fGj736r_2fLAHxJnp1YMBRQ_3d_3d

NEWS

Genetic Alliance awarded. Patient and Provider Education at the Heart of New Prenatal and Early Childhood Diagnosis Program

The U.S. Health and Human Services, Health Resources and Services Administration (HRSA), Genetic Services Branch, awarded Genetic Alliance, in partnership with the National Coalition for Health Professional Education in Genetics and other partners, a four year $3.28 million cooperative agreement for the implementation of the Prenatally and Postnatally Diagnosed Conditions Awareness Act (PPDCAA). PPDCAA is dedicated to improving the system of information and support for prenatal and early childhood diagnosis. The partners will develop models for diagnosing congenital conditions, beginning with: Down syndrome, spina bifida and dwarfism. The program will create replicable, sustainable mechanisms for patient and provider education.

Receiving prenatal or early childhood diagnosis is a life-changing event with countless challenges," explained Sharon Terry, president and chief executive officer of nonprofit health advocacy organization Genetic Alliance. "Families have to navigate a fractured healthcare system to attain the services and care their children deserve. The Congenital Conditions Program will provide individuals, families and providers with access to accurate, evidence-based, timely, unbiased, quality information to foster informed decision making that will impact all aspects of a child's life."

The goal of the Congenital Conditions Program is to collect and disseminate evidence-based information, while coordinating the availability of supportive services for parents whose child receives a diagnosis prenatally, at birth, or up to one year after birth. Program partners will work with HRSA to expand and improve:

- Awareness of national and local peer-support programs.
- Creation of awareness and educational programs for healthcare providers who provide, interpret and inform parents of confirmatory diagnosis results.
- Public use of the Genetics and Rare Diseases Information Center.

- Integration of the Quality Assessment Toolbox, as developed through a Center for Disease Control and Prevention cooperative agreement for the Access to Credible Genetics Resources Network.

This initiative provides an opportunity for family-centered groups to work with health professionals, regional and state programs, federal agencies, and other stakeholders in a cohesive program that increases awareness, educates families and providers, and delivers information during a critical time in a child's development," said Joseph D. McInerney, executive director of the National Coalition for Health Professional Education in Genetics. DCAA paved the way for funding supporting the diagnosis of conditions that impact many individuals throughout their lifespan. Down syndrome, the most commonly occurring chromosomal condition in the U.S., occurs in one in every 733 babies annually. Today there are more than 400,000 people living with Down syndrome. The National Center for Health Statistics indicates that spina bifida, a condition that affects the neural tube, is found in one newborn in 2,500. Types of dwarfism occur in one in every 95,000 newborns. Although the program will focus on these three conditions initially, it will create and solidify a model that can be applied to other conditions. I look forward to building on the work of organizations committed to Down syndrome, spina bifida, dwarfism, and other congenital conditions. This project will be fueled by collaboration," said Natasha Bonhomme, project director of the Congenital Conditions Program and the vice president of strategic development at Genetic Alliance.
For more information about Genetic Alliance, visit www.geneticalliance.org

BOOKS

THE VOICE OF 12,000 PATIENTS – Experiences and Expectations of Rare Disease Patients on Diagnosis and Care in Europe
This book can be downloaded fully or by sections from the Eurordis website at: http://www.eurordis.org/article.php3?id_article=1960

IT'S MY LIFE
'It's My Life' portrays a revolution that is taking place now! Twelve patients with a rare hereditary disorder all over the world tell in their stories the determination to take charge, to make a real difference.
This book can be ordered at any local bookstore. Copies ordered via the **European Genetics Alliances' Network** (EGAN) will bring a donation for each copy towards patient support activities. Your order can be send to egan@egan.eu
The price for each book will be € 15 (± US $ 20) excluding shipping costs.

BECOME A MEMBER OF

The international multidisciplinary community genetics network. It's aim is to facilitate communication between all those working in the field of community genetics (and genomics). To become a member you only need to send an e-mail mentioning this desire to commgennet@gmail.com As a member you are invited to stimulate your colleagues to become a member, or to send me the e-mail address(es) of (the) person(s) you propose.

The **Genetic Alliance Network**. Genetic Alliance offers a wide array of programs, resources, and events for all the individuals and organizations in our network, including:
- Annual Conference, - Genetic Alliance Webinars, - MemberForum, - Discussion listserv, - Weekly Bulletin, - Policy Bulletin, and quarterly newsletter, - WikiAdvocacy and WikiGenetics, - Disease InfoSearch
To join the network, please visit: http://www.geneticalliance.org/join

International Alliance of Patients' Organisations (IAPO) is a unique global alliance representing patients of all nationalities across all disease areas and promoting patient-centred health care around the world. If you like to receive IAPO's newsletter, please go to this website: http://www.patientsorganizations.org/index.pl?n=210;section=3 to submit your request.

RELEVANT WEBSITES

ORGANISATIONS

BIO – a biotechnology industry organisation whose members are involved in research and development of innovative healthcare
Website: www.bio.org/healthcare/issues/

EGAN – focuses on genetics, genomics and medical biotechnology and their implications.
Website: www.egan.eu

International Alliance Patient Organizations (IAPO) - global alliance representing patients worldwide across all disease areas
Website: www.patientsorganizations.org

March of Dimes – is an organisation for pregnancy and baby health
Website: www.marchofdimes.com

UNESCO – gives information on life sciences and the implications on all species.
Website: www.unesco.org/shs/bioethics

WAO – for the prevention and treatment of genetic and congenital conditions.
Website: www.world-alliance.org

Genetic Interest Group (GIG) – is a national alliance of patient organisations of genetic disorders.
Website: www.gig.org.uk

Erfocentrum – Dutch national genetic resource and information centre.
Website: www.erfocentrum.nl/english.php

WHO – its Genomic Resource Centre provides information and raises awareness on human genetics and human genomics.
Website: www.who.int/genomics/en

MAJOR PROJECTS with International dimension and genetic alliances' engagement:

CAPABILITY – Capacity Building for the Transfer of Genetic Knowledge into Practice and Prevention
Website: www.capabilitynet.eu

PATIENTPARTNER – to promote the role of organisations in the clinical trials context.
Website: www.patientpartner-europe.eu
EuroGenGuide – contains information about genetic testing, counselling and research across Europe.
Website: www.eurogenguide.eu

EuroGenTest – harmonizing genetic testing across Europe.
Website: www.eurogentest.org
Human Genome Project Information – informs about project facts, education, medicine and the new genetics.
Website: www.genomics.energy.gov

RARE DISORDERS:

Orphanet – the portal for rare diseases and orphan drugs
Website: www.orpha.net

International Birth Defects Information Systems (IBIS) – to promote better care and prevention of birth defects through information sharing
Website: www.ibis-birthdefects.org

IGA MEMBERS:

Europe: European Genetic Alliances' Network (EGAN),
Website: www.egan.eu
Central & Eastern Europe: Central & Eastern European Genetic Network (CEE GN),
Website: www.ceegn.org
Asia: Indian Genetic Society
Africa: Southern African Inherited Disorders Association (SAIDA),
Website: www.saida.org
Latin America: Brazilian Genetic Alliance (BGA),
Website: www.abg.org.br
North America & International: Genetic Alliance,
Website: www.geneticalliance.org
Middle East: Iranian Genetic Alliance of Parent/Patient Organisations & Gulf State Alliance
Australia and New Zealand: Association of Genetic Support of Australasia (AGSA),
Website: www.agsa-geneticsupport.org.au & New Zealand Organisation for Rare Disorders (NZORD),
Website: www.nzord.org.nz

UPCOMING INTERNATIONAL EVENTS with IGA - involvement and partnering

11 – 15 April 2010, Alexandria, Egypt
BioVisionAlexandria 2010 – New Life Sciences: Future Prospects

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3 – 6 May 2010, Chicago, USA
BIO International Convention

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4 - 6 October 2010, Paris, France
EuroBio

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6 - 9 October2010, Brussels, Belgium.
1st European Congress on Preconception Care and Preconception Health

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10 - 12 October 2010, Berlin, Germany
World Health Summit

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27 - 30 March 2011, Lyon, France
World Life Sciences' Forum, Biovision

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11 – 15 October 2011, Montréal, Canada
International Federation of Human Genetics Societies (ICHG/ASHG)

IGA CONTACT DETAILS

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