



MINUTES

International Confederation of Countries

Advisory Council

12th December, Radegast Lakeview Hotel, Beijing

Attendees

HVP Country Node Representatives

Heather Howard	Australian Node Representative
Martina Witsch-Baumgartner	Austrian Node Representative
Thomy de Ravel de l'Argentiére	Belgian Node Representative
Ming Qi	Chinese Node Representative
Xitao Li	Chinese Node Representative
Andreas Hadjusavvas	Cypriot Node Representative
Sherifa Ahmed Hamed	Egyptian Node Representative
George Patrinos	Greek Node Representative
Fahd Al Mulla	Kuwaiti Node Representative
Zilfalil bin Alwi	Malaysian Node Representative
Tilak Shresta	Nepalese Node Representative
Maria Jesus Sobrido	Spanish Node Representative
Chí Dũng Vĩ	Vietnamese Node Representative

Human Genetics Society Representatives

Ghazi Tadmouri	Assistant Director, Centre for Arab Genomic Studies
Kevin Carpenter	President, Human Genetics Society of Australasia
Wayne Grody	President, American College of Medical Genetics
Lynn Jorde	President, American Society of Human Genetics
Milan Macek	Past-President, European Society of Human Genetics
Yoichi Matsubara	President, International Federation of Human Genetics Societies
Carmencita D. Padilla	Former President, Asia Pacific Society of Human Genetics
Feliciano Ramos	President, Spanish Society of Human Genetics
Augusto Rojas-Martinez	President, Latin American Network of Human Genetics Societies
Raj Ramesar	Liaison & Representative, African Society of Human Genetics
Graham Taylor	President, Human Genome Variation Society

Human Variome Project Interim International Scientific Advisory Committee

Arleen D. Auerbach	USA
Mona El-Ruby	Egypt
David Rimoin	USA
John Burn	UK
Mauno Vihinen	Finland
Garry Cutting	USA

Human Variome Project Staff

Richard GH Cotton	Scientific Director, Human Variome Project
Timothy Smith	Communications Officer, Human Variome Project
Julia Hasler	UNESCO Liaison
Helen Robinson	WHO Liaison

Apologies:

Mireille Claustres	France
Aida Falcón de Vargas	Venezuela

Meeting Opened 0900

1. The Role of The Council

Richard GH Cotton presented the Role of the Council (see supporting documents).

John Burn reminded the Council that a Node is a permanent entity and therefore it is essential that it has the endorsement of the National Society at the time of creation. Work through the IFHGS, ESHG etc. to ensure political buy-in.

2. HVP Country Node Representatives: 5 minute introductions & updates

Australia: Process started by forming a consortium of diagnostic lab heads and clinicians to assess what they need in terms of information. Developed automated collection software and data portal from there. Software available to other nodes for use/modification.

Austria: Austrian Society of Human Genetics (Österreichische Gesellschaft für Humangenetik, ÖGH) – 120 members. 4 person working group formed. Looking into networking, collection of variants, collection of patients, use of international databases, interpretation. Possibility of regional node in South East Europe.

Belgium: 9 centres performing 355 tests. Rare Diseases Plan started by Jean-Jacques Cassiman looking at national registries and centres of excellence. Has strong links with Cartagena.

China: Introduction and update given previously in main meeting.

Cyprus: 1 institution doing testing. Establishing a database.

Egypt: Largest population in Arab world. Consanguinity is about 28-33% and it reaches up to 60% in some areas as in upper Egypt. Node so far run by Egyptian Neurogenetic Disorders Consortium in Upper Egypt. Datasets for other diseases are also currently in development. In conversation with national society to create a unified national repository.

Greece: Developed ETHNOS software platform – available for other Nodes. Have been collecting Greek data since 2003.

Kuwait: Up to 70% of marriages are consanguineous. Have so far achieved infrastructure and training; sending colorectal data to InSiGHT. To develop a Node you need: Leadership, funding inductive environment, collaboration between clinicians and scientists, recognition and support of policy makers.

Malaysia: 1Malaysia Human Genome Variation Consortium launched SNP databases in Oct 2010. Recently the database has been expanded to include gene mutations. Future plans are to include CNV and clinical phenotype databases.

Nepal: 8 member consortium, including government representatives. Active interaction with the Chinese Node. Need training assistance (PhD, MSc).

Spain: Is disseminating the vision of the Human Variome Project. Has regional funding for neurological and psychiatric disorders. Running satellite meetings at the Spanish Society meetings. Talking to existing disease groups and societies. Funding is a problem. A rare disease registry being formed by IRDiRC.

Vietnam: 1 hospital has clinical genetics capacity. Registries for rare diseases exist.

Other Countries Represented

South Africa: Possess reasonable genetics infrastructure. A node is possible in the future.

UAE: Nodes must be viable. Stable support must come for the Government. Scientists alone can't influence. Need international organisations to help, i.e. HVP, WHO.

Japan: Money becoming available for rare diseases. Good time to introduce Human Variome Project.

Philippines: Only 7 clinical geneticists but some registries already. Here to learn more. Will raise the issue at the Asia Pacific Society.

UK: 23 centres. Concept of a Node doesn't really fit, but DMuDB could be it. A "Node" could rather be a person, endorsed by the national society. Each nation needs a Node/Liaison Officer. Big countries will find this difficult.

US: Similar to UK. What is the Human Variome Project adding to efforts in places like the US and UK? Richness in populations, diversity of alleles, consanguinity. Will begin conversation again with NIH and societies.

Finland: No HVP Country Node, but FINDIS (Finnish Disease Database) exists. Trying to develop Node and work with Society.

Latin America: The Latin American Network of Human Genetic Societies (RELAGH) will coordinate the launching of regional nodes in some countries like Brazil, Mexico, and Venezuela, in partnership with UNESCO and WHO. RELAGH will also encourage the opening of other nodes in different countries of the region. Several American and European colleagues already involved in HVP and related projects expressed their support to collaborate with these regional nodes.

3. Open Discussion

Model System

George Patrinos led the discussion and began by presenting the ETHNOS database platform which has been specifically designed to run NEMDBs (National and Ethnic Mutation Databases). ETHNOS, the Australian system, DMuDB and Findbase are all potential model systems for HVP Country Nodes.

It was noted that a central database will not work (caBIG cited as an example) and the Human Variome Project could endorse a data aggregation system, not a central database. The Council was reminded that this is what the Human Variome Project is doing. The Global Collection Architecture proposed by the Human Variome Project calls for data to be collected by both HVP Country Nodes and gene/disease specific databases and shared or aggregated in a free and open manner with any person or database that wants it. This will include existing central databases, but the Human Variome Project will not be creating a new central database.

Curation needs to be performed by an expert consortium and the most appropriate venue for this is at each gene/disease specific database, rather than an HVP Country Node.

Standards Development

It was recognised that the Human Variome Project's Standards Development Process is a robust and transparent process. The Council should focus on the development of standards, not the implementation.

HVP Country Nodes must share their data with the international gene/disease specific databases. Data must be able to be shared, therefore when developing standards for HVP Country Nodes, they must be developed with the knowledge and recognition of the Standards and Guidelines developed for gene/disease specific databases.

Role of the Council/Human Variome Project

The question was raised as to the scope of discussion – just databasing, or broader. It was agreed that the primary role of the Council was to encourage the development of standards for HVP Country Nodes, but that it also served as a forum for discussion around broader topics, such as assisting developing nations, etc.

This led to further discussion about the role of the Human Variome Project: is the Project just focussing on the collection of information, or is it also concerned with helping countries develop genetics services. It was resolved that the role of the Human Variome Project was not to make it possible for countries to develop genetics services, but to enhance their existing services.

Meeting Adjourned for Lunch

4. Open Discussion

Assistance for Developing Nations

The idea of promoting bioinformatics as a sector of the economy as well as supporting public health services needs to be explored more thoroughly. The needs of developing countries will need careful examination as each country will have different levels of expertise, resources and needs. Plus the ethical, cultural and legal issues and the capacity to deal with these issues, is likely to differ substantially from country to country.

Important to get WHO on board with this initiative. Also need concrete steps taken soon to capture the interest of established players in the US, etc.

Fahd Al Mulla noted that all HVP efforts should be more widely publicised. Fahd also suggested that if a proposal for a specific project around transfer of knowledge and addressing resource disparity was submitted to the Gulf States, with an endorsement from WHO, then there is a high likelihood that it would be funded.

Preliminary discussions between ICO and WHO have indicated that WHO is interested in the needs of developing countries in relation to genetics and genomics and public health. WHO has recently put out a call for input into its 'Grand Challenges in Genomics for Public Health in Developing Countries: Top 10 policy and research priorities to harness genomics for the greatest public health problems' process. Several of attendees said that they had been contacted about this, and almost all present expressed interest in the WHO project and requested more information.

The ICO will follow up with WHO with a view to finding out more.

This led to a discussion of how HVPI could actually assist its members in developing countries, and what concrete steps could be taken. Suggestions included:

- Assistance with the setting up of effective country nodes
- Assistance in collecting, running of samples
- Sharing of expertise
- Setting up formal and informal relationships between established nodes and those with less experienced to facilitate exchange of information and knowledge and develop common understandings
- Assistance in identifying local learning and educational needs; provision of training and educational materials, curricula and course outlines for adaption to local needs; and assistance for conducting and evaluating learning courses
- Assistance with dealing with ethical, social and legal issues
- Assistance with approaching governments for support and involvement
- Assistance in advocacy, public awareness raising with multiple stakeholders

These issues will be taken up by HVPI in furthering its work on assisting developing countries.

Several attendees from developing countries expressed the view that links with WHO are often an important part of promoting innovative public health in their countries and that their local officials were often guided by WHO's views on these matters.

Further discussions were conducted *in camera*.

WHO and UNESCO

On behalf of the ICO, Helen Robinson gave some background on WHO and its ways of working. She indicated that WHO can be thought of as having three levels: the Headquarters in Geneva where the Director-General is based; six regional offices, each with its own Regional-Director; and about 150 country offices in low and middle income countries, that are linked to regional offices and often located inside the national Ministry of Health. This means that Ministers of Health,

health bureaucrats and policy makers are key in the relationship with WHO. WHO's key concerns are the public health implications of any health issue and in addressing the disparities between health outcomes of the rich and poor. This latter area of health and development was driving the WHO Grand Challenges project as many commentators were seeing that they area of genomics research had the potential to further exacerbate the divide between rich and poor nations. There are several concerns:

- emphasis of new technologies, and new skills risked increasing the pressure on already fragile health systems in poorer countries
- the risk that rich countries would step in and take over the field in weaker countries rather than developing lasting capacity there
- that the open and sharing collaborative model and ways of working that underpinned the work on the human genome need to be maintained as this will enhance the benefits for all

Helen also said that she was very interested in taking to anyone involved in the WHO Grand Challenges project. She also suggested that the release of the HPV1 new draft Road Map in January 2012 might provide the opportunity for consortium members to meet with local officials in WHO or their Ministry of Health to discuss genomics and public health and the role of HPV1.

Julia Hasler reported on UNESCO. The Human Variome Project is now a Non-Governmental Organisation in Operational Relations with UNESCO and this is important as an indicator of an official relationship with UNESCO. In terms of raising awareness of Human Variome Project amongst politicians, most countries have permanent delegations to UNESCO in Paris and HVP members could usefully arrange meetings with their delegations (Ambassadors or Science officers) to coincide with HVP4 in June in order to brief them on HVP goals and on their country's involvement in HVP. There could be a possibility to hold an information meeting for all permanent delegations on the Human Variome Project during HVP4.

UNESCO's interest lies in promotion of international cooperation and capacity building in Science to ensure the use of scientific research for the betterment of society. Besides the continuing cooperation of HVP with the International Basic Sciences Programme of UNESCO under whose auspices the 4th meeting is being held, we should also involve UNESCO's Division of the Ethics of Science and Technology in HVP4.

Raj Ramesar summed up the discussion by stating that we need to use the HVP4 meeting to get buy in to the Project from the political level.

Purpose of Consortium

It was recognised that the benefits of joining the Human Variome Project Consortium need to be better defined and articulated. The ICO needs to be able to sell Consortium. Existing benefits are:

- Voice in the process
- Stand for elections
- Find collaborators

Discussion was had relating to updating the mantra or tag-line of the Human Variome Project. The final suggestion was: sharing genomic knowledge · improving global health

The Council also suggested that the mission and objectives of the Human Variome Project be reviewed to make them “tighter and cleaner.”

ACTIONS FOR ICO: Connect Consortium members with UNESCO
Follow up on WHO *Grand Challenges Round 2*
Develop an action plan for funding *Pathfinder* projects

- Obtain confirmation in writing from China regarding ability to assist
- Coordinate global PR campaign around announcement on DNA Day

Pursue proposal to Gulf States
Define and articulate the purpose of the Consortium
Review mantra, mission and objectives
Develop draft plan for future meetings schedule

Meeting Closed 1700

Supporting Documents

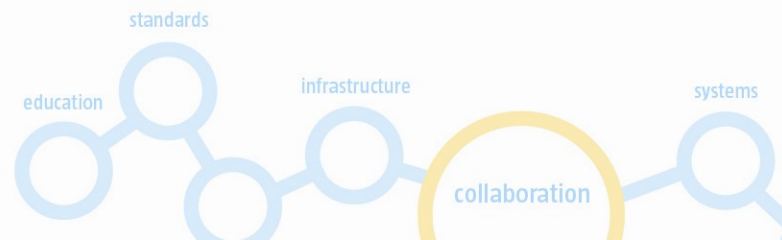
1. Presentation by Richard GH Cotton – “The Role of the Council”

The Role of the Council

Richard G. H. Cotton

Scientific Director

Human Variome Project

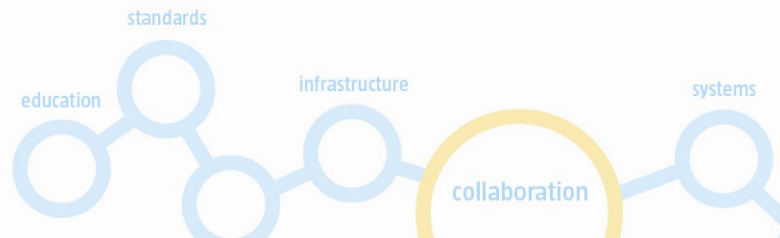
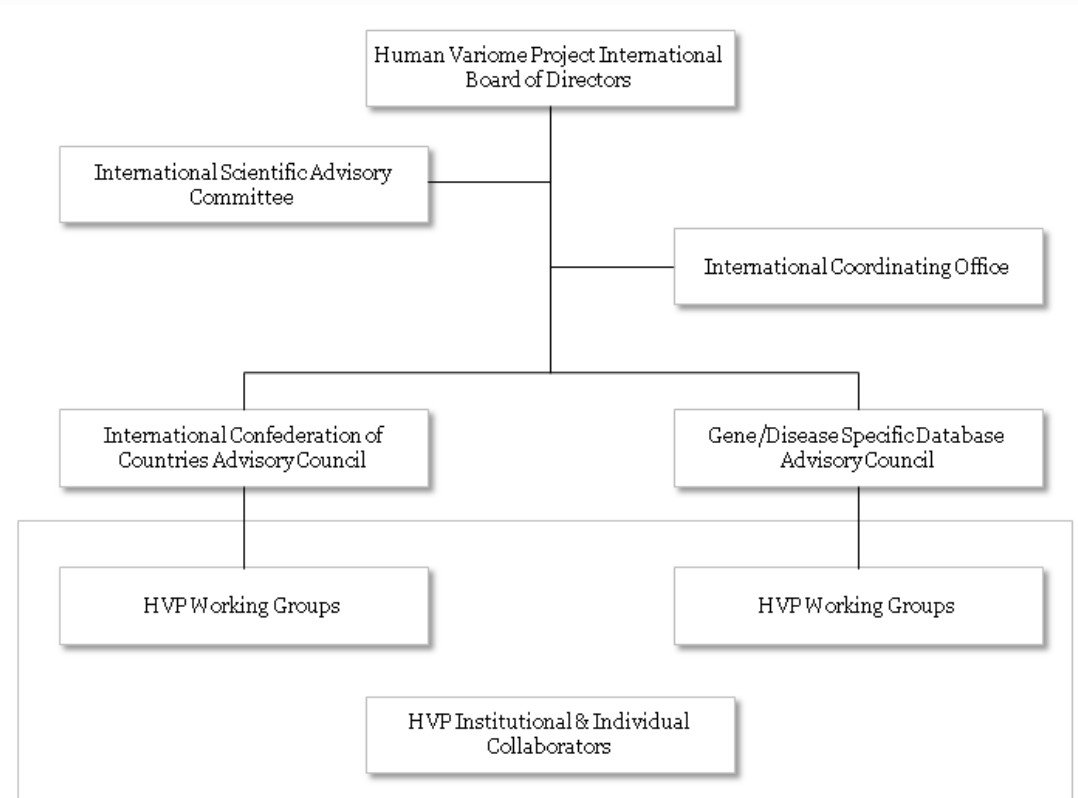


The Human Variome Project



Sharing data reducing disease

- Representative
- Consultative
- Global

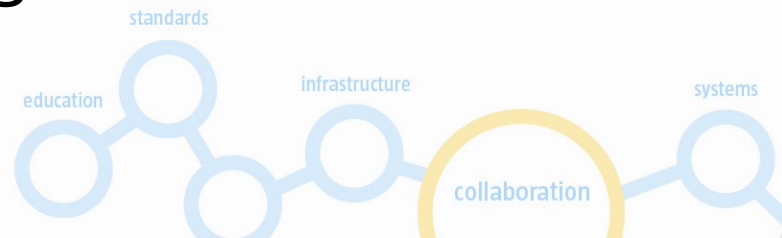


ICC Advisory Council



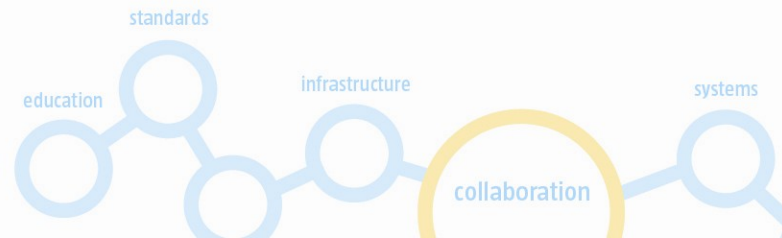
Sharing data reducing disease

- The International Confederation of Countries Advisory Council is composed of one representative from each of the *HVP Country Nodes*
- Representatives are appointed by their respective Nodes
- Chair is elected by the membership of the Human Variome Project Consortium present at the HVP biennial meetings



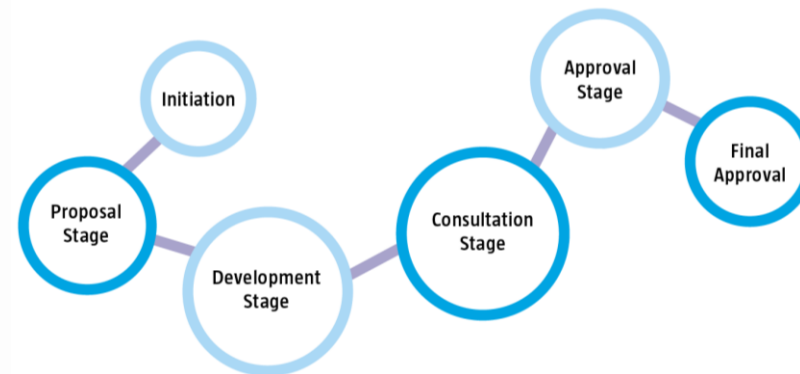
Advisory Councils

- The Advisory Councils of the Human Variome Project are directly responsible for overseeing and directing the work of the various HVP Working Groups that are formed from time to time under their auspice

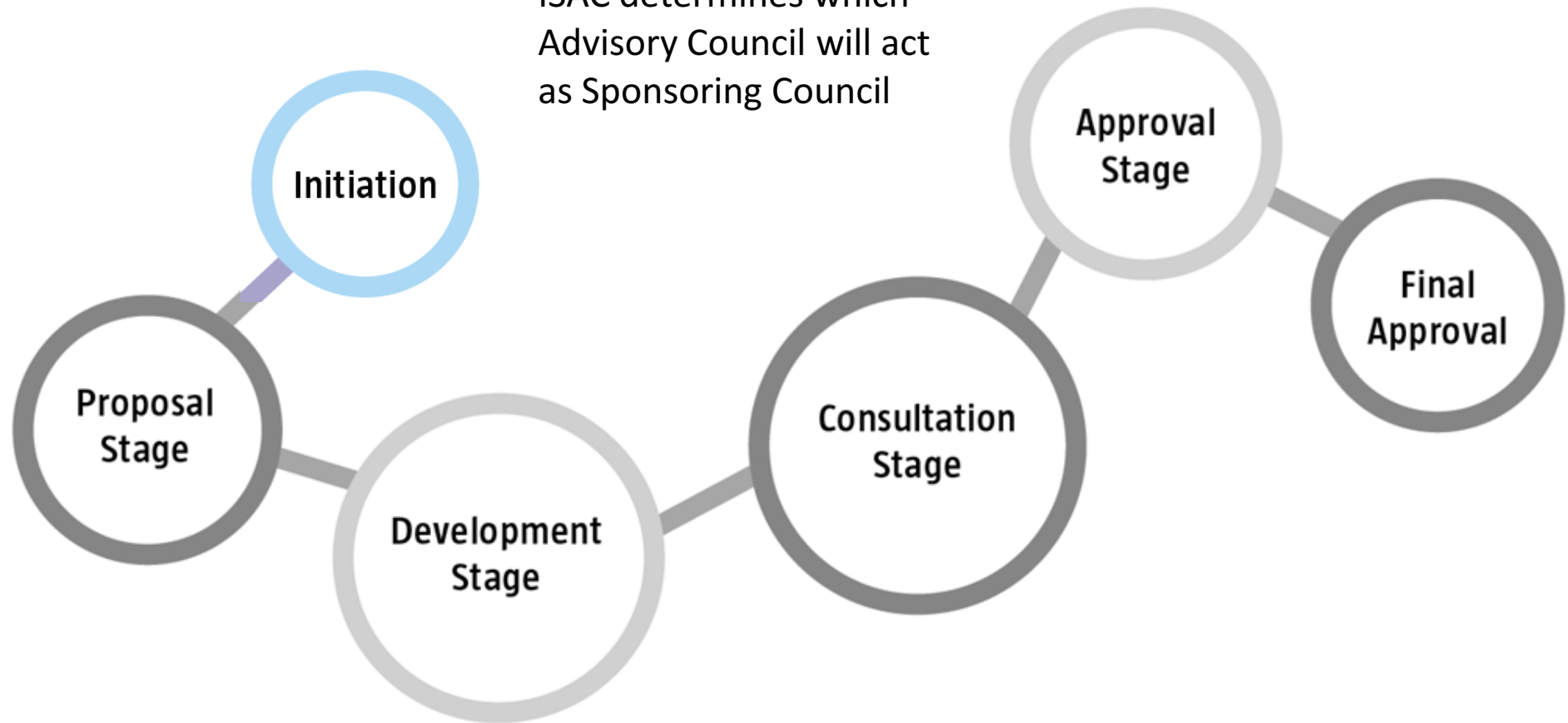


Primary Responsibility

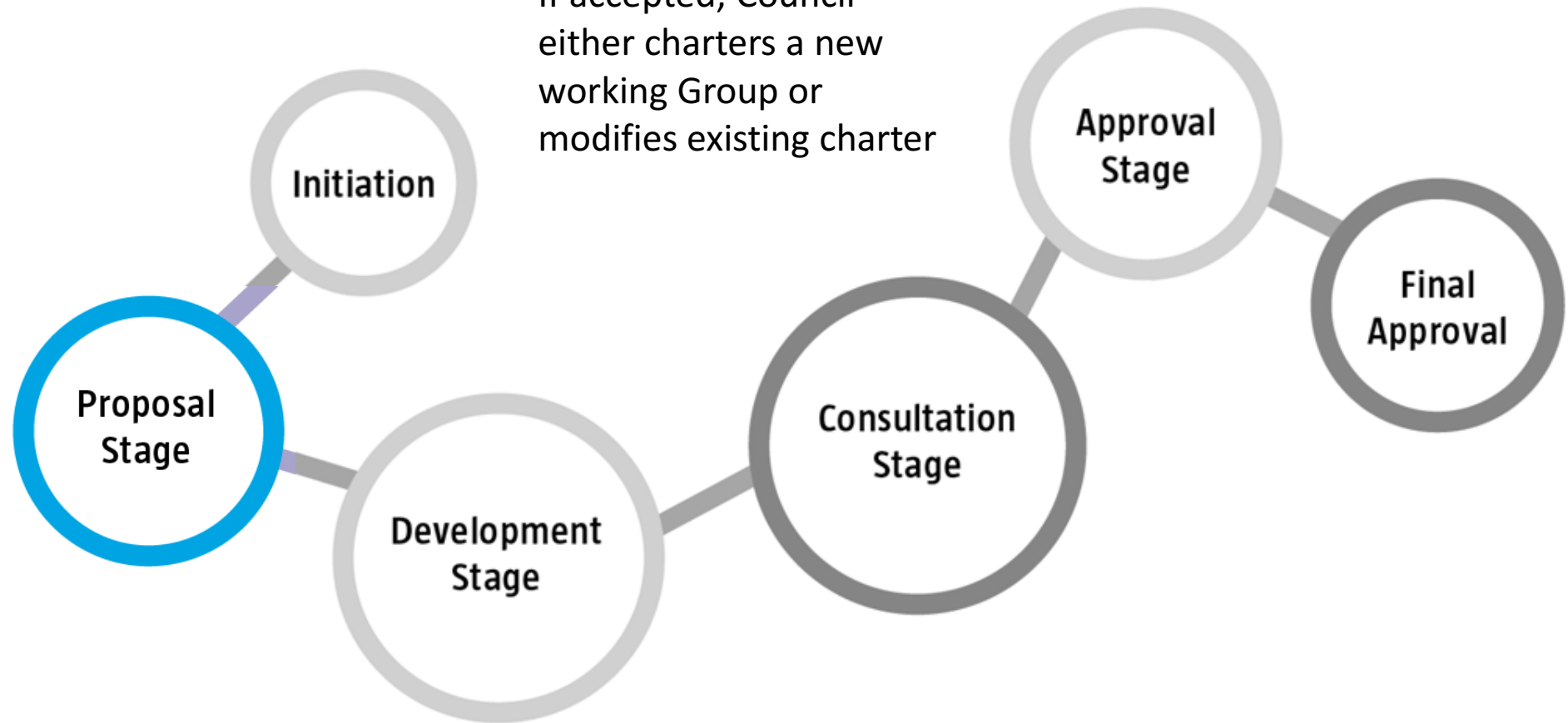
- Oversee the creation of HVP Standards and Guidelines for the creation, operation, and maintenance of HVP Country Nodes
 - Process for this described in *PD06-2011: Standards Development Process*



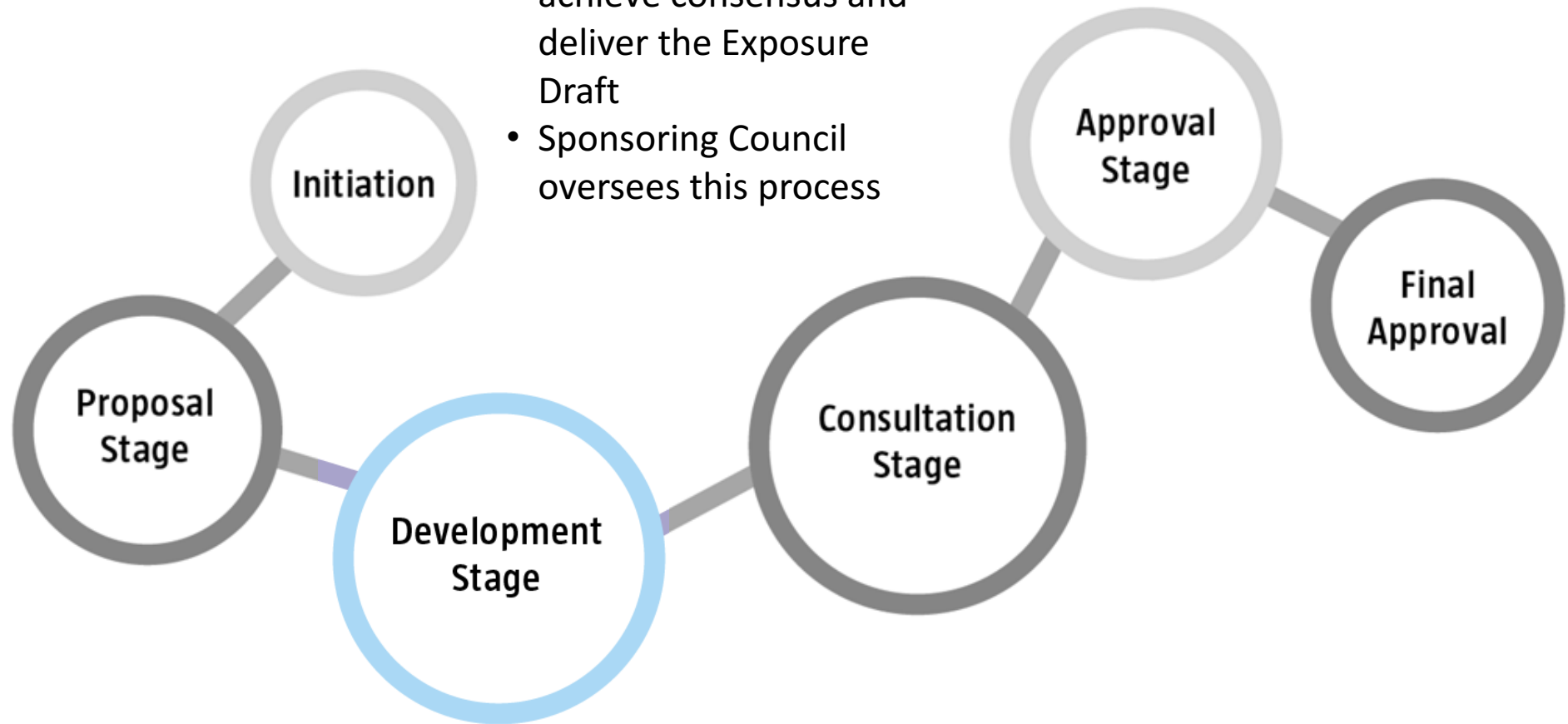
- A Consortium member submits an Activity Proposal to the ISAC
- ISAC determines which Advisory Council will act as Sponsoring Council



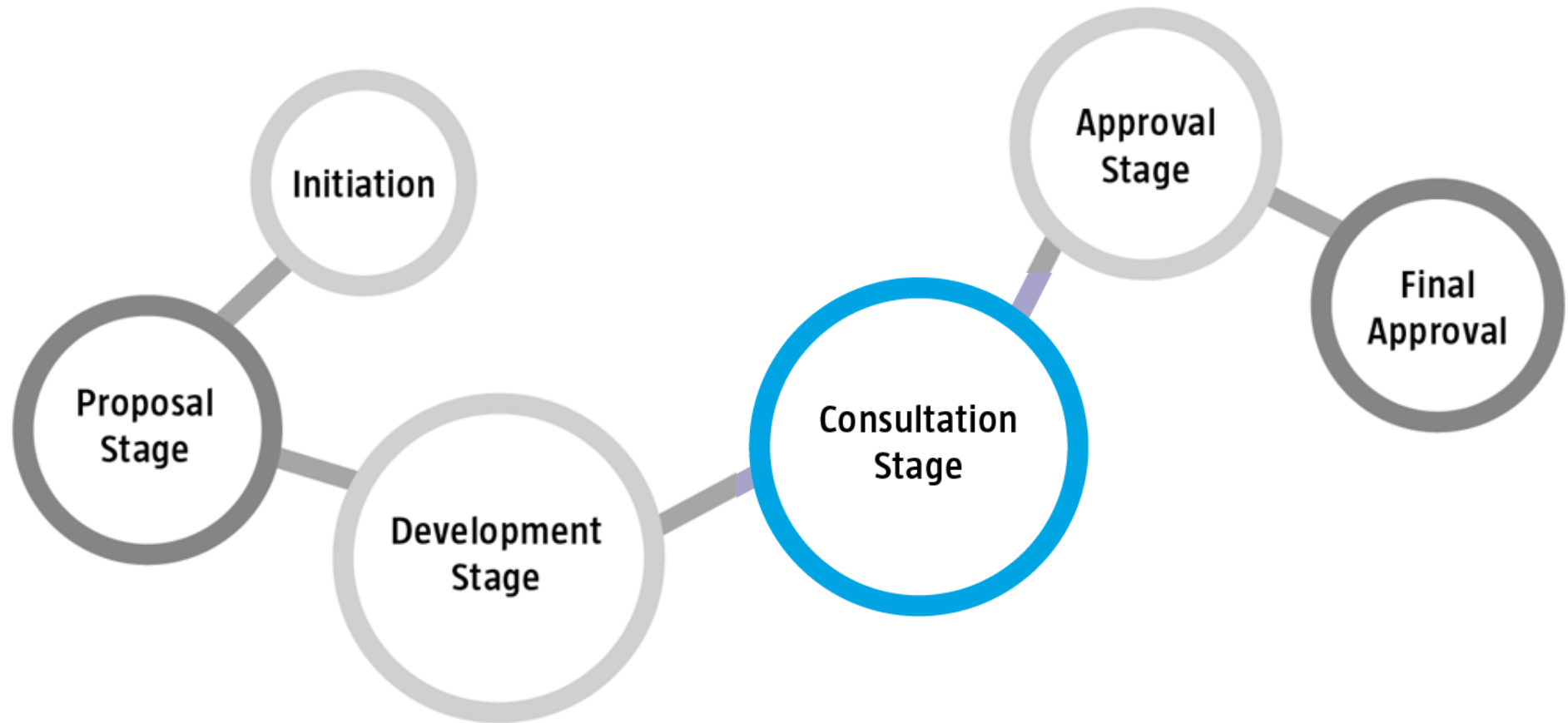
- Sponsoring Council considers Activity Proposal
- If accepted, Council either charters a new working Group or modifies existing charter



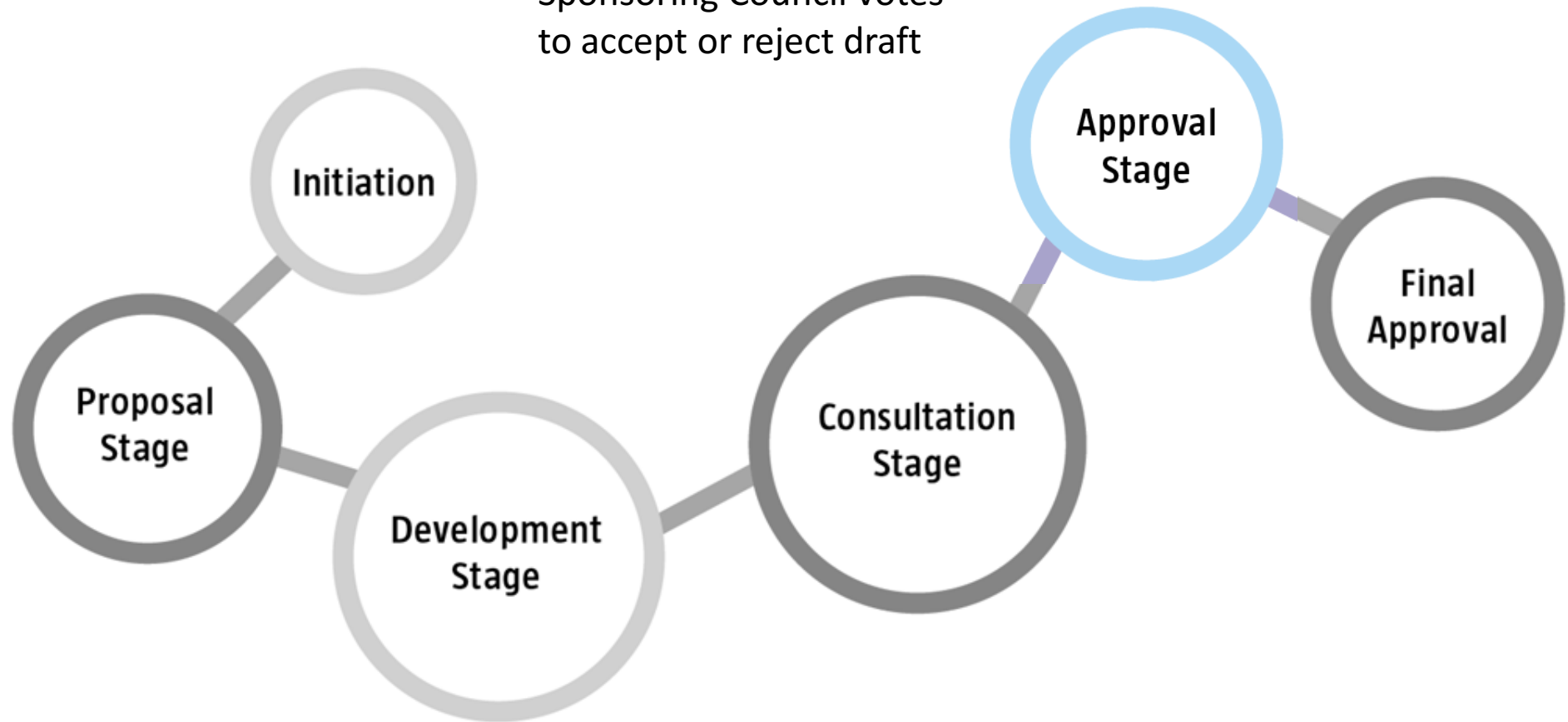
- Working Group interactively develops Working Drafts until they achieve consensus and deliver the Exposure Draft
- Sponsoring Council oversees this process



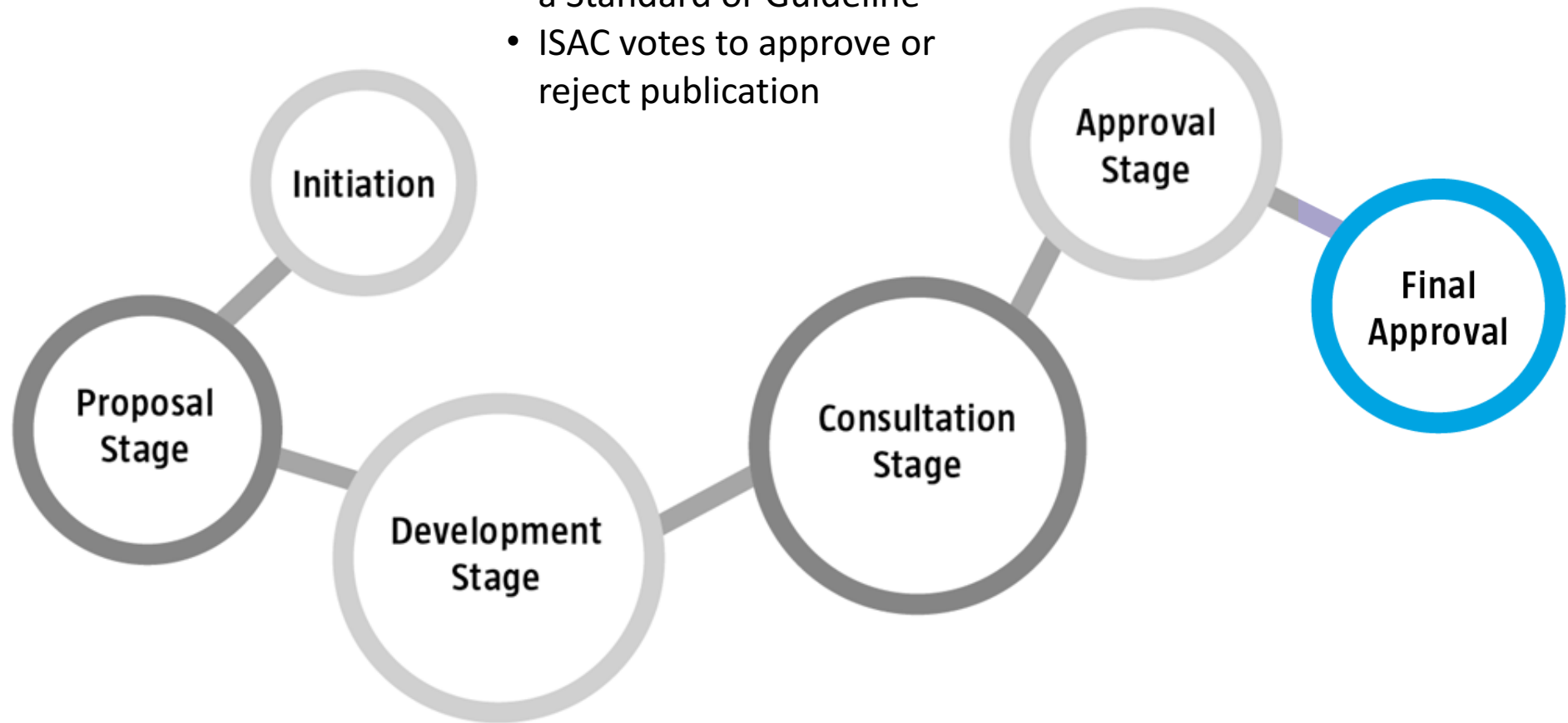
- Exposure Draft is released to HVP Consortium for review and comment



- Working Groups submits approved exposure draft to Sponsoring Council
- Sponsoring Council votes to accept or reject draft



- Sponsoring Council recommends publication to ISAC of draft as either a Standard or Guideline
- ISAC votes to approve or reject publication



Timelines

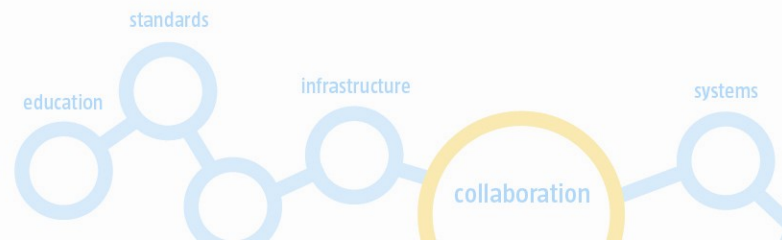
Development Stage (where the work gets done) is the longest

– Up to two years to achieve consensus

PLUS:

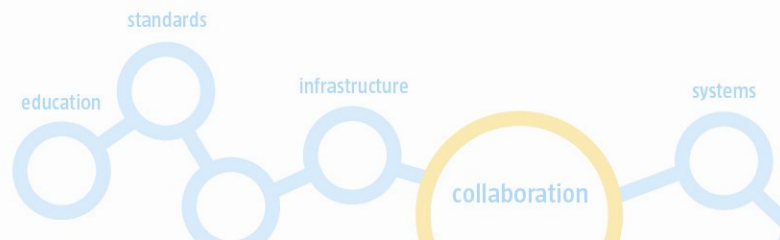
- 60 days for Community Consultation
- 60 days to respond to Community comments
 - or a further 6 months if Community rejects draft
- Time for AC and ISAC to vote

A long time – but time is needed to ensure consensus



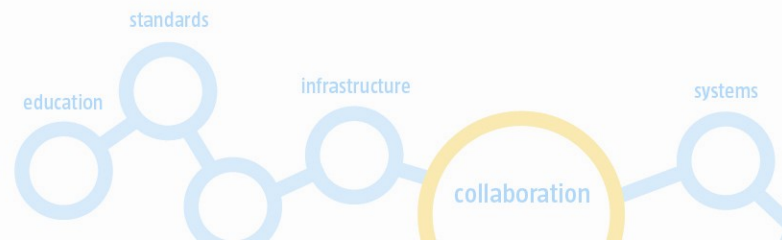
In the meantime...

The International Scientific Advisory Committee is able to make recommendations of best practice that should be followed until such time as an appropriate HVP Standard or Guideline is produced.



Other Responsibilities

- Provide advice and guidance to ISAC on matters concerning collection and sharing of data from individual countries
- Actively encourage the creation of new HVP Country Nodes
- Ensure all HVP Country Nodes comply with published HVP Standards and Guidelines
- Work with ICO to develop relevant education and training programmes



ICO Support

- The International Coordinating Office will provide secretarial support to the Council
- ICO can help arrange meetings and provide telephone and video conferencing facilities
- Our new website (March/April 2012) will have a suite of tools to enable communication and collaboration amongst Council members
- ICO will be appointing 1.0 FTE Staff member

