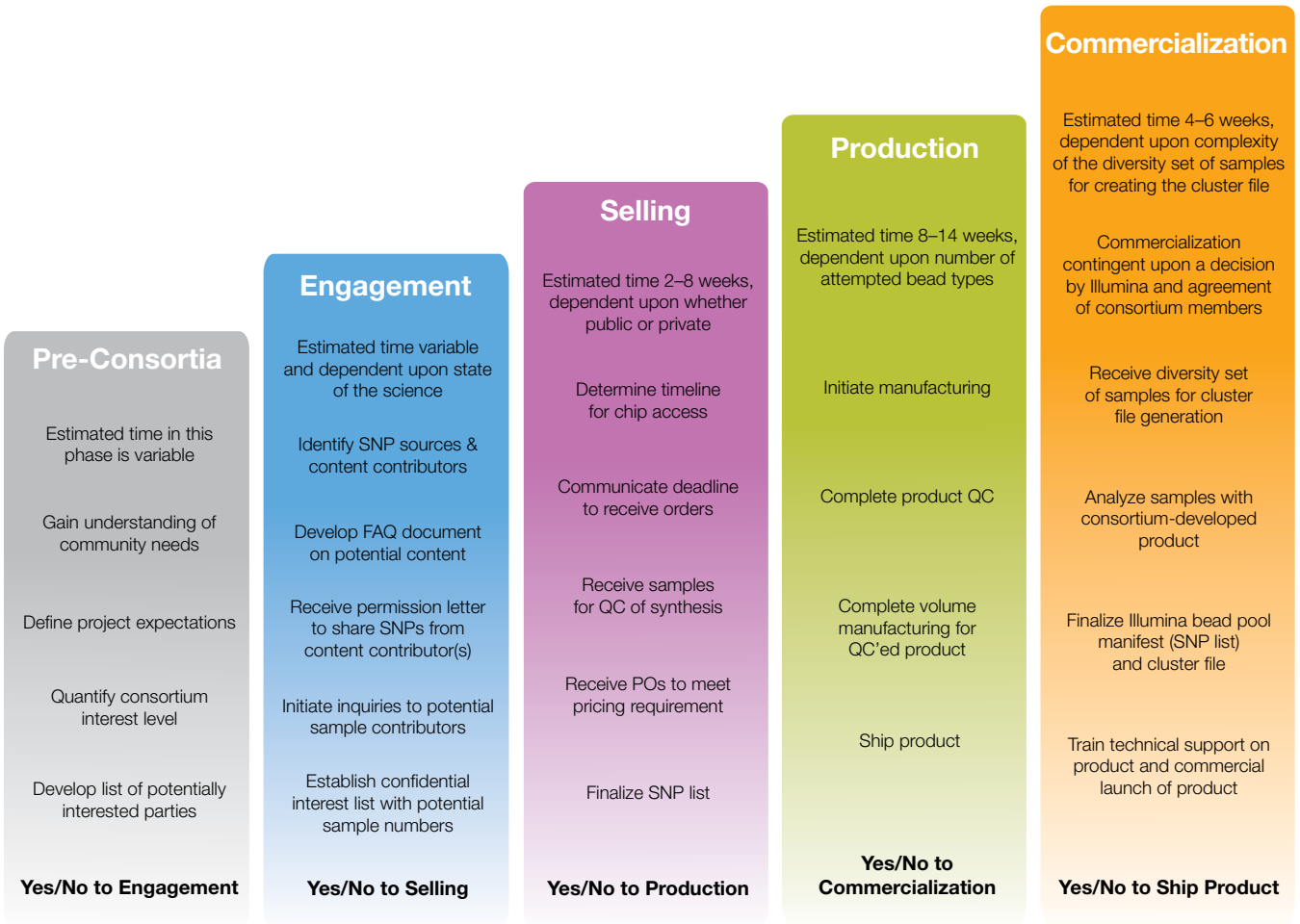
Human Consortia Processes and Milestones

What is a human consortium?

Illumina categorizes projects that directly involve human genetic research or models for human genetic research as “human research”. The Human Consortium program provides a framework for researchers to pool resources in an effort to develop biomarker/SNP genotyping or sequencing tools for a variety of genetic conditions. Confidentiality is maintained throughout the process.

Why is Illumina interested in consortia?

Supporting genotyping tools for human genetic research is strategically vital to Illumina’s mission. The needs for each research community can be different. Within each community, researchers may have overlapping goals that may be best addressed by a group effort and a single tool or set of common tools. Consortia help reduce the barrier of entry for individual researchers by enabling development of genetic analysis tools available to the community.



The process for creating a new product through a consortium occurs over a series of strategic phases. During each phase, a set of milestones must be met before the process can continue on to the next phase. This ensures the greatest probability for success by enabling consortium members to make well-informed decisions along the path to product development.

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The word “consortium” is frequently used to describe many community efforts. How does Illumina qualify potential consortia?

Illumina relies on the wider research community to identify genetic conditions that are a good fit for a consortium approach. In many cases, existing consortia will approach Illumina to inquire about developing a tool through a community effort. Conversely, Illumina will sometimes drive the formation of a consortium if many independent researchers’ inquiries suggest that pooling efforts might be a useful approach. There are a number of flexible options for a consortium approach. The consortium can remain confidential or open, as preferred by consortium members.

Are there particular products that consortia use, or can any of the products be accessed?

Typically an iSelect® custom genotyping BeadChip is developed that can meet the research goals of multiple groups. These BeadChips are based on Illumina’s BeadArray™ technology and are compatible with the proven Infinium® HD assay. The iSelect platform supports densities of 3,000 to 1,000,000 attempted bead types. Depending upon the content, this means that up to 1,000,000 SNPs and insertions/deletions (indels) can be simultaneously queried per sample.

Benefits, Roles, and Responsibilities

What is the benefit to participants of a consortium approach?

Researchers can typically access a higher density of SNP markers and a lower per-sample price than the budget of a single contributor might otherwise allow. Through the consortium, the cost of bead pool synthesis is spread over more samples than might be possible from a single institution. The minimum order for participating in a consortium is 48 samples, providing that all orders for a private iSelect BeadChip total at least 1,152.

What types of projects are appropriate for a consortium approach?

The consortium approach can be applied to any project that would benefit from pooling resources to develop a genotyping tool that will meet the needs of multiple researchers.

Is it mandatory for consortia to be willing to commercialize content in order to participate?

While several of Illumina’s commercial genotyping projects were developed through consortia, commercializing the content is not a requirement. In fact, private consortia are an integral part of Illumina’s overall mission. Illumina maintains strict confidentiality around private consortia, so these projects are not publicized. The only requirement for content (candidate SNPs or indels) in consortia projects is that it can be shared with other members of the consortium. Depending upon the goals of the content contributors, content may be maintained confidentially within a small group or content can be made publicly available. If consortium members are amenable to sharing content publicly, the decision to create a commercial product will be at Illumina’s discretion. Some consortia may decide to share their content via a “controlled sell,” in which consortia members determine who in the research community can have access to the final product.

What is a “controlled sell” consortium product?

Illumina works closely with consortia members to determine the access level that the research community may have to the final product. There are three levels of access: first, and most public, is a commercially-available product, which is available to all members of the research community. On the other end of the spectrum is a private product, which is only available to the consortia members. In the middle is the “controlled sell,” in which consortia members (or a steering committee) determine and communicate to Illumina who has access to the final product.

What is the difference between a “content contributor” and a “sample contributor” in a consortium?

Content contributors are typically small subsets of researchers that have driven the discovery efforts of variants comprising a given phenotype. Sample contributors include anyone in the research community interested in the targeted condition or genetic trait. Illumina requests permission from content contributors to share final content with sample contributors. For sample contributors, Illumina will communicate a deadline by which purchase orders (POs) must be received in order to participate in the consortium. The final sample count for all orders received by the PO deadline will determine the per sample price.

What are the responsibilities of consortium members?

Content contributors should be referenced in any publications, public presentations, press releases, or public announcements resulting from use of the genotyping content. Illumina’s standard terms and conditions for products and services will also apply.

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Forming, Joining or Participating in Consortia

Do I need to have my draft genome and all SNPs in hand prior to inquiring about a consortium?

It is not necessary to have all content defined prior to exploring the consortium option with Illumina. In fact, many groups benefit from Illumina's experience in helping define a streamlined path for discovering, validating, and finalizing content.

What if the timelines of the community change after entering into a consortium with Illumina? For example, what if we evaluate the available data and believe that additional SNP/biomarker discovery is needed prior to finalizing content?

Illumina works closely with content contributors to revisit timelines to ensure that goals of the consortium and the research community as a whole are best addressed. We offer customers the ability to augment content on existing BeadChips with the latest advances from genome-wide association studies, next-generation whole-genome sequencing, and exome sequencing studies for variant confirmation, fine mapping, and target validation. iSelect + add-on content provides unprecedented flexibility to customers who want to add newly discovered content to a custom array after the initial design period is completed.

Access and Pricing

I only need to run one plate (96 samples) and have previously not been able to meet the minimum sample number needed for running an iSelect project with Illumina. How can I benefit from a consortium?

Researchers with as few as 48 samples can benefit from a consortium. Since the bead pool and the BeadChips will be manufactured for the entire consortium at once, as long as the minimum sample order (1,152 samples) is met by the community, individual researchers can order as few as 48 samples' worth of BeadChips.

How many samples can I assay on a BeadChip?

For SNP/indel content between 3,000 and 90,000 attempted bead types, a 24-sample BeadChip format is used. For SNP/indel content between 90,001 and 250,000 attempted bead types, a 12-sample BeadChip format is used. For SNP/indel content between 250,001 and 1,000,000 attempted bead types, a 4-sample BeadChip format is used.

Samples per BeadChip

Attempted Bead Types per Sample	BeadChip Format
3,000 to 90,000	24-sample
90,001 to 250,000	12-sample
250,001 to 1,000,000	4-sample

What do I need to run the chips once the consortium BeadChips are shipped to me?

The per-sample price includes BeadChips and all reagents required to run the Infinium HD assay. Illumina recommends a minimum of 200 ng of DNA (> 50 ng/µl as measured by a fluorescent method of quantification) for best results. Access to an Illumina BeadArray Reader, iScan, or HiScan™SQ system is required for BeadChip scanning. For help finding a local service provider or core facility, or for information about Illumina's FastTrack Services, please contact Illumina Tech Support or your local account manager.

How do I design my genotyping tool? How are SNPs and insertions/deletions (indels) probes designed?

Researchers can design probes for SNPs or indels using the Assay Design Tool (ADT), available through iCom, Illumina Tech Support, or FastTrack Services. The ADT assigns a score for each submitted design that gives a relative confidence that a probe will successfully query a particular SNP or indel based on the surrounding sequence. The output file finalized by content contributors can then be submitted to the Illumina orders group to place the final order.

How are SNPs selected and content validated?

Research communities considering a high-density genotyping tool often have access to several sources of SNP content with varying levels of validation. The state of the research will dictate whether additional validation might be useful before finalizing the SNP list. Illumina will work with content contributors to reduce risks before finalizing content. Design scores generated by ADT for each potential SNP are well-established indicators of successful probe design.

What is the consortium price?

Consortium pricing will depend upon the number of total attempted bead types and the number of samples submitted by the deadline determined by the consortium. For any given consortium opportunity, the appropriate density of markers and anticipated sample numbers will differ. Illumina will work directly with content and sample contributors to help identify possible pricing scenarios.

What is the minimum order I can place?

Minimum orders are for 48 samples (the smallest reagent kit configuration), provided that the minimum sample number (1,152) is achieved by the consortium. Re-orders can be placed for up to one year after bead pool manufacture.

Is there a limit to how many samples I can order at the consortium price?

There is no limit to the number of samples that can be ordered, provided the order is in 48-sample increments (47-sample increments for FastTrack Services). 48 samples is the smallest reagent kit configuration for a shipment.

Can I re-order additional BeadChips?

The bead pool is manufactured in liquid phase. Once the bead pool is manufactured, the life of the liquid phase is 1 year. When orders are filled, the liquid bead pool is stabilized onto BeadChips, which go through quality control and remain under warranty for a minimum of 6 months from the date of manufacture. This means that the effective time period during which experiments can be run on a bead pool is 18 months. Re-orders can be submitted during the lifetime of the bead pool.

Data Analysis and Sharing

My research competes with other interested parties, so I do not want my sample numbers widely publicized. Is it a requirement that other members know about my experiments?

Any information on sample numbers or experiments from each institution will be kept confidential. Individual consortium members can decide whether they are comfortable revealing the goals of their experiment to other consortium participants. Illumina will only divulge the total sample numbers pooled by all participants that will determine the per-sample price. Illumina will request written or email permission from researchers to discuss research objectives among all consortium members.

How do I participate?

Researchers interested in the Consortium program can contact consortiamanager@illumina.com. A specialist will be able to answer any additional questions and help place orders.

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