



Introduction

The YCC (Y Chromosome Consortium) Newsletter is a forum for communicating progress in the field of evolution of the human Y chromosome. In particular, this Newsletter will contain a regular update of the YCC Repository, the YCC Database and other information (polymorphisms, participants, recent publications, meetings, etc.). The YCC Newsletter will be published twice a year. The Editors welcome contributions to the YCC Newsletter. The next publication deadline is June 1, 1994.

Purpose

A collaborative effort has been initiated to study genetic variation on the human Y chromosome. The overall goal is to use genetic variation on the Y chromosome to test hypotheses on the origin of modern human populations. Studies of Y chromosomal variation will serve to complement the already growing databases of mitochondrial and nuclear DNA sequences.

Two major challenges have emerged: 1) obtaining a representative sample of human populations from a variety of geographic origins, and 2) discovering a sufficient number of informative polymorphisms on the Y chromosome. The observed level of polymorphism is low. The implication of this observation is that large portions of the Y chromosome must be examined in order to sample sufficient genetic variation.

Our approach to meet these challenges is to establish a repository of lymphoblastoid cell lines (the Repository) derived from a sample of individuals representing

indigenous populations from all inhabited continents and to provide DNA isolated from these cell lines to investigators searching for polymorphisms on the Y chromosome. The results of typing the same set of DNAs at many Y-specific loci will be pooled into a common database (the Database), in a similar fashion to the CEPH international collaboration. Many cell lines have already been accessioned in the Repository, but in order to achieve the goal of global diversity, we need the support of anthropological and Y chromosome research communities (see Repository Update).

There are several advantages to a cooperative effort. The rate at which new polymorphisms are identified on the Y chromosome will be accelerated, as will the rate of progress in determining human Y haplotypes. Emphasis will be placed on identifying polymorphisms that are most useful in tracing human histories, i.e., those that have arisen a single time in human evolution. Once several informative polymorphisms have been identified in humans, the ancestral state at each site will be determined by examining the same sites in a sample of great apes. The polymorphism data will be combined into haplotypes, and phylogenetic analysis will be performed using the great apes as outgroups. Ultimately, we will use the information in the Database to determine the time and order of branching of all Y haplotypes in the repository. The frequencies of these and other haplotypes can then be measured in larger population samples.

An additional advantage of this approach is that it will allow for an assessment of the phylogenetic utility of several kinds of polymorphisms; i.e., those in unique DNA, repetitive sequences and microsatellites.

Repository Update

The Repository is located in the Laboratory of Human Genetics at the New York Blood Center. The current estimate for the number of cell lines that the Repository will support is 150. These cell lines will be derived from many indigenous populations. The goal is to accumulate about 20-30 cell lines from each of the major population groups (African, Caucasoid, Asian, Amerindian, Oceanic, and Australoid). Hopefully, each of the major groups will be represented by a sampling of 4-8 sub-populations with 3-5 members from each. The Repository will also contain a few individuals representing species of great apes and somatic cell hybrids carrying a human Y chromosome.

Cell lines are accessioned into the Repository and given a YCC number in the order of their arrival at the Laboratory. They are also given HG numbers which link the cell lines with files containing information from the contributor about the people from which the cell lines were derived. Finally, the code which accompanied the cell line from the contributor is recorded with the YCC and HG numbers. The minimum information we would like to maintain in the files about the people from which the cell lines are derived is 1) language spoken (if native), 2) location where the people live, and 3) the ethnic origin of the people (what the people call themselves). Photographs are welcomed.

The current list of cell lines in the Repository is shown in Table 1 (see Appendix). The Repository is currently under-represented in Asian, Oceanic and Australoid peoples. African, Caucasoid, and Amerindian people are well represented, or will be soon. Potential contributors are encouraged to contact Nathan Ellis at 212-570-3075 or Michael Hammer at 602-621-9828.

At intervals, Maria Proytcheva at the Blood Center will expand the cell lines to a quantity of 200 million cells and then freeze them as pellets. The frozen pellets will be shipped to the University of Arizona where DNA will be prepared. By contributing cell lines to the Y Chromosome Consortium Repository, you will be making DNA samples freely available to any researcher who seeks to study the evolution of the Y chromosome. Those who are interested in the cell lines (for example, to do pulsed-field electrophoresis) will be able to obtain them after we have secured permission from the contributors to the Repository.

DNA Acquisition

Aliquots of DNA isolated from the panel of cell lines will be made available to those searching for or scoring Y chromosome polymorphisms. As a requisite for receiving the DNAs, researchers must agree to score the entire set of DNAs and submit their results to the YCC Database (see below). If you are interested in receiving genomic DNA samples from this collection, a letter of request should be sent to Michael Hammer. Contributions to defray costs already incurred will be accepted. Upon request, a complete set of aliquots containing 10 μ g of DNA will be shipped. Special

requests for larger aliquots (i.e., 100 µg) will be considered; however, we are limiting the number of sets of these larger aliquots at this time.

The Database

A database of genotypes of all individuals represented in the Repository will be maintained at the University of Arizona. The format will be a spreadsheet with the YCC numbers down the left side and the loci tested along the top. The Database will be accessible to all participants by anonymous ftp at [joplin.biosci.arizona.edu](ftp://joplin.biosci.arizona.edu) in the YCC directory. The database will become available also in Gopher or Mosaic at [joplin.biosci.arizona.edu](ftp://joplin.biosci.arizona.edu). The Database will be updated several times a year depending on volume and will be published in the YCC Newsletter. When sufficient data has been collected, results from analyses of the Database will be submitted for publication by the "Y Chromosome Consortium" as a group.

Y Chromosome Polymorphisms

A list of Y chromosome polymorphisms, compiled by Chris Tyler-Smith, is presented in Table 2 (see Appendix). The list describes polymorphisms in the literature up to January 1994. Pseudoautosomal polymorphisms have been excluded, but variants observed on a single Y chromosome are included. DXYS1Y (pDP34) and DYS153 (CRI-J192) polymorphisms have also been reported but they have not been described in detail. The editors request information on these and other Y chromosome polymorphisms. This information will be published in the YCC Newsletter.

Minutes

Round table discussion on the "Evolution of the Y Chromosome" at the American Society of Human Genetics meeting in New Orleans (October 5-9, 1993).

Attending: Al Agellon (U. Arizona), Roxane Bonner (U. Arizona), Benny Chan (U. Texas, Houston), Michael Cummings (U. Illinois, Chicago), Nathan Ellis (New York Blood Center), Robert Erickson (U. Arizona), Patricia Fetchner (Johns Hopkins), Mary Haag (All Child. Hosp., St. Petersburg), Michael Hammer (U. Arizona), Yufeng Huang (Boston U.), Li Jin (U. Texas, Houston), Judith and Kenneth Kidd (Yale), Harry Ostrer (NYU), Andres Ruiz (Stanford), Frank Schneiders (Inst. Hum. Genet., Hannover), Chris Tyler-Smith (Oxford), and Doug Vollrath (Stanford).

The Human Y Chromosome Consortium met for the second time at the ASHG meeting. Chris Tyler-Smith presented his Y chromosome polymorphism data and phylogenetic analysis. Nathan Ellis gave an overview of the cell line repository which now has over 50 lines; he noted that the Repository has a poor representation of Australoid, Oceanic, Asian, and perhaps Caucasoid peoples. Michael Hammer gave a short data presentation and the YCC Database was discussed. Then the floor was opened for discussion. A number of issues were raised including the logistics of cell line accession and DNA acquisition. Mary Haag offered to look at cytogenetic variation of Y chromosomes in the repository. Judy and Ken Kidd discussed the Genome Diversity Project and the possibility of joining efforts in the collection of cell lines in the future. Finally, we discussed the presentation of Y polymorphism data at a future symposium. Nathan Ellis and Michael Hammer submitted a proposal to the ASHG program committee entitled "Tracing Human History: Perspectives from the Y chromosome, mtDNA and the autosomes". It included a list of potential speakers and topics. Unfortunately, there are about 30 other proposals competing for 6 workshops. It was decided that it would be best to investigate other possible meetings which would be appropriate for a Y chromosome symposium. One possibility is the American Association of Physical Anthropologist meetings.

Announcement

The editors request that participants submit a brief statement of their research interests (e.g., populations and polymorphisms being investigated) for publication in the next edition of the YCC Newsletter. Please submit by June 1, 1994.

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Appendix

Table 1. The origins of cell lines contained in the Y Chromosome Consortium Repository.

HG#	YCC#	Source code	Language	Geographic origin	Ethnic origin
2214	01	Yale No. 117		North America	Amerindian
2215	02	Yale No. 123		North America	Amerindian
2216	03	Yale No. 139		North America	Amerindian
2217	04	Yale No. 218		North America	Amerindian
2255	05	NASAM	!Kung	Namibia	San
2256	06	JK736	Aka	Bagandu, CAR	Biaka Pygmy
2257	07	JK741	Aka	Bagandu, CAR	Biaka Pygmy
2258	08	JK1029	Niger/Kordofanian	Ituri, Zaire	Mbuti Pygmy
2259	09	JK1031	Niger/Kordofanian	Ituri, Zaire	Mbuti Pygmy
2260	10	JK965	Nasioi	Bougainville, Solomon Isl.	Melanesian
2261	11	JK971	Nasioi	Bougainville, Solomon Isl.	Melanesian
2262	12	JK1364	Tupi	Rondonia, Brazil	Karitiana
2263	13	JK1370	Tupi	Rondonia, Brazil	Karitiana
2264	14	JK1493	Tupi	Rondonia, Brazil	Surui
2265	15	JK1498	Tupi	Rondonia, Brazil	Surui
2266	16	JK1504	Tupi	Rondonia, Brazil	Surui
2267	17	JK1624	Yucatec	Campeche, Yucatan	Mayan
2268	18	JK1626	Yucatec	Campeche, Yucatan	Mayan
2486	19	SAIMR01/JR020	!Kung	Namibia	Tsumkwe San
2487	20	SAIMR02/JR031	!Kung	Namibia	Tsumkwe San
2488	21	SAIMR03/JR321	!Kung	Namibia	Tsumkwe San
2489	22	SAIMR04/JR323	!Kung	Namibia	Tsumkwe San
2494	23	TiMu/AR01		Tucson, Arizona	Navaho
2513	24	HaMi/AR02		Tucson, Arizona	Ashkenazi Jewish
2496	25	AnPa/AR03		Tucson, Arizona	Tohono O'Odham
2497	26	RoDa/AR04		Tucson, Arizona	Irish/English
2498	27	McAa/AR05		Tucson, Arizona	Porch Creek
2588	28	SAIMR13/JR354	!Kung	Namibia	Tsumkwe San
2589	29	SAIMR14/JR077	!Kung	Namibia	Tsumkwe San
2590	30	SAIMR15/JR305	!Kung	Namibia	Tsumkwe San
2591	31	SAIMR16/LD148	Bantu	South Africa	Herero

Table 1. (continued)

HG#	YCC#	Source code	Language	Geographic origin	Ethnic origin
2592	32	SAIMR17/Alb77	Bantu	South Africa	S. Sotho
2593	33	SAIMR18/Alb74	Bantu	South Africa	Pedi
2595	34	SAIMR07/JR306	!Kung	Namibia	Tsumkwe San
2596	35	SAIMR08/JR054	!Kung	Namibia	Tsumkwe San
2597	36	SAIMR19/Alb47	Bantu	South Africa	Tswana
2598	37	SAIMR20/WK122	Bantu	South Africa	Ovambo
2609	38	SAIMR05/JR013	!Kung	Namibia	Tsumkwe San
2610	39	SAIMR06/JR301	!Kung	Namibia	Tsumkwe San
2611	40	SAIMR09/LD156	Bantu	South Africa	Herero
2612	41	SAIMR10/LD145	Bantu	South Africa	Herero
2613	42	SAIMR11/Alb27	Bantu	South Africa	Zulu
2614	43	SAIMR12/Alb55	Bantu	South Africa	Tswana
2632	44	SAIMR22/LD185	Bantu	South Africa	Herero
2633	45	SAIMR23/LD188	Bantu	South Africa	Herero
2634	46	SAIMR21/LD147	Bantu	South Africa	Herero
2643	47	JK3146		Siberia	Yakut
2644	48	JK3149		Siberia	Yakut
2645	49	JK3150		Siberia	Yakut
2646	50	JK3151		Siberia	Yakut
2647	51	JK3152		Siberia	Yakut
2648	52	JK3158		Krasnador (Black Sea)	Adygeans
2649	53	JK3159		Krasnador (Black Sea)	Adygeans
2650	54	JK3160		Krasnador (Black Sea)	Adygeans
2651	55	JK3161		Krasnador (Black Sea)	Adygeans
2652	56	JK3168		Krasnador (Black Sea)	Adygeans

A strike-through indicates the cell line was entered into the Repostory but the cells did not proliferate.

Table 2. Polymorphisms on the human Y chromosome.

A. Conventional RFLPs.				
locus	probe	enzyme	comments	reference
DYS11	12f2	TaqI, EcoRI	insertion/deletion, probe gives extensive cross-hybridisation	2
DYS1	49f, 49a	TaqI	complex pattern, many variable fragments low stringency conditions required	6, 10
		PvuII		14
		BglII, HindIII, PstI, SstI		15
DYS7	50f2	EcoRI	fragment C absent in one individual	3
		TaqI	novel fragment in two samples low stringency conditions required	5
DYZ3	YalphaI	HindIII	additional site in some subunits of tandem array very high stringency required	16
DXYS5Y	47z	StuI	point mutation	9
DYZ8	21A1	TaqI	complex polymorphism	4
DYS21	116/21	PstI	one individual only	7
DYS150	13/0.9	BglI	one individual only	7
DYS152	CRI-J177	BglII, HindIII, PstI	insertion/deletion	1
DYS287	pYAP	EcoRV, TaqI	Alu sequence insertion	12
DYS27	1/2	TaqI	point mutation in tandem array	5
-	92R7	HindIII	point mutation	8

B. Polymorphisms detected by PFGE.

locus	probe	enzyme	comments	reference
DYZ3	YalphaI	several	array size polymorphism	16, 11
		AvaII	point mutation	16
		EcoO109I	point mutation	11
DYZ5	Y-190	XbaI	array size polymorphism	17
-	poxY1	BglII, XbaI	detects one variable DYZ1 fragment	11
-	RBF2	SfiI	detects two independent variable loci	5
DYS1	49f	SfiI	one or two independently variable loci	5
-	RBF4	SfiI	one hypervariable locus	5
-	M911	XbaI	nature of polymorphism uncertain	8
DYZ1	HY10	many	complex hypervariable pattern	8
DYZ2	HY2.1	many	complex hypervariable pattern	8

C. Polymorphisms detected by PCR.

locus	locus name	comments	reference
-	27H39LR	GATA repeat	13
-	YCAI	CA repeat	8

-	YCAII	CA repeat, two Y loci amplified	8
-	YCAIII	CA repeat, two Y loci amplified	8

Table 2. References

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