THE HUMAN GENETIC MUTANT CELL REPOSITORY

List of Genetic Variants, Chromosomal
Aberrations and Normal Cell Cultures
Submitted to the Repository

Fourth Edition
October 1977

U.S. DEPARTMENT OF HEALTH, EDUCATION, AND WELFARE
Public Health Service
National Institutes of Health

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List of Genetic Variants, Chromosomal Aberrations
and Normal Cell Cultures

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Fourth Edition

October 1977

INSTITUTE FOR MEDICAL RESEARCH

Copewood and Davis Streets

Camden, New Jersey 08103

609-966-7377

SPONSORED BY THE NATIONAL INSTITUTE OF GENERAL MEDICAL SCIENCES

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Preface to the Fourth Edition

Acquisition of cell cultures by the Repository and shipment of cell cultures to investigators both continue to increase at an accelerating rate as shown on the graph which follows this preface. In addition to many new cell cultures and supplementary data on others, the present edition contains new features. McKusick's numbers, where applicable, are inserted immediately following the name of each disease. A bracket is placed around cell cultures from members of a family group. HL-A antigens have been determined on many of the lymphocyte cell cultures and this data is recorded in the table on pages 161-163. Eleven cultures are available from both this Repository and from the American Type Culture Collection in Rockville, Maryland under a different numerical designation. To avoid confusion we have listed the ATCC designation under "Remarks".

Camden, N.J., August 1977

Lewis L. Coriell, M.D, Ph.D.

Arthur E. Greene, D.Sc.

THE HUMAN GENETIC MUTANT CELL REPOSITORY

CULTURES SHIPPED ●
CULTURES RECEIVED *

MARCH 1, 1977

I. INTRODUCTION

The Human Genetic Mutant Cell Repository established in 1972 by the National Institute of General Medical Sciences at the Institute for Medical Research, Camden, New Jersey, contains low passage skin fibroblast, lymphoblast, amniotic fluid cell, and a few animal cell cultures stored in liquid nitrogen from hereditary diseases including those with biochemical and chromosome abnormalities and normal controls. Cell cultures stored in the Repository are verified for freedom from contamination, species of origin, karyotype, viability and expression of the biochemical or chromosomal defect. These cells are available to organizations or individuals engaged in health-related research or health delivery concerning early diagnosis, prevention, treatment, counseling and research for control of some of the 2,000 or more inherited diseases that afflict human beings. The problems in many of these diseases are gene abnormalities which alter one or more chemical steps in the normal metabolic sequence of cells. The application of techniques for detailed study of these cellular chemical processes in cell culture is leading to a wealth of information about human genetics and improvement in the prevention and treatment of genetic diseases and probably many other diseases in which genetic factors are not yet recognized to play a significant role. The Repository program has been developed by the National Institute of General Medical Sciences with the help of a scientific advisory committee which periodically reviews the progress of the

collection and provides advice on the acquisition of cell lines.

Cell cultures are listed alphabetically under cell type and biochemical or morphological abnormalities including the genetic mutant (GM) repository number, tissue, passage number, culture medium, submitter, age, sex, race, genetic status, verification and remarks.

Appreciation is expressed to the many investigators who help the NIGMS Advisory Committee and the Repository staff select representative subjects and help validate the genetic defects by providing clinical and laboratory data or assays on the cell culture.

Interested investigators are invited to utilize the Repository as a source of genetic mutant cell cultures.

Lewis L. Coriell, M.D., Ph.D.

Arthur E. Greene, D. Sc.

II. PROCEDURES FOR ORDERING GENETIC MUTANT CULTURES

1. REQUIREMENTS

The cells are distributed only to qualified professional persons who are associated with recognized research, medical, educational or industrial organizations engaged in health-related research or health delivery. Before fibroblast cells can be shipped, the assurance form (Par. 10) must be signed and returned to the Repository. This is adequate for most cell cultures in the Repository. However, lymphoblast or virus transformed cell cultures require prior agreement to observe the Minimum Safety Guidelines (Page 12) and return of the signed Agreement on Lymphoid and Virus Transformed Cells (Par. 11). These forms may be obtained by writing to the Repository, or may be photocopied.

2. PROCEDURE

Requests for cell cultures must be submitted on institutional purchase forms including purchase order numbers. Purchasing agents should indicate the name of the investigator on such orders. Telephone orders will be accepted when accompanied by a purchase order number, and should be confirmed within several days by letter. Each culture requested should include the Genetic Mutant Repository number (GM number), and the diagnosis.

3. ADDRESS

All requisitions should be addressed to:

Dr. Arthur E. Greene

The Human Genetic Mutant Cell Repository

Institute for Medical Research

Copewood and Davis Streets

Camden, New Jersey 08103

Telephone 609-966-7377

4. FEES

The fee for each 25 cm² flask of cells is \$20.00 to non-profit institutions, but is reduced one dollar per culture up to 5 orders received simultaneously i.e. \$19.00 each for an order of 2 cultures, \$16.00 each for 5 or more. Frozen ampules are not shipped, because experience has shown much better success in shipping freshly revived flask cultures. Mass production of cell cultures is not a function of the Repository, (See Appendix E). The Repository is designed to provide only seed cultures. It is suggested that the recipient store aliquots of early passages in liquid nitrogen as insurance against contamination, accidents, artifacts associated with aging, and loss of the culture.

5. SHIPPING CHARGES

The shipping charges are prepaid and will be added to the invoice at the time of billing.

6. HOW SHIPPED

All cell cultures are grown and frozen in antibiotic-free media to aid in detection and prevention of contamination. Cell cultures in the Repository have been tested and found free of mycoplasma, bacteria, molds and fungi during characterization, at the time of frozen storage, and after recovery from liquid nitrogen. When an order is received, a frozen ampule is usually recovered from liquid nitrogen on the following Thursday and the medium is changed on Friday. The following Monday, the culture is inspected, the medium is removed and the T25 flask is filled with fresh medium, packed and shipped, usually by air mail special delivery. The flask is enclosed within two watertight plastic envelopes within a styrofoam box in a cardboard mailer to prevent leakage, overheating or freezing during shipment. A return postcard is enclosed. Please return the postcard with notations about the condition on arrival. Pertinent suggestions to improve the effectiveness of the Repository are appreciated.

7. DESCRIPTIVE DATA

Each shipment contains descriptive data about the cell culture, suggested directions for cell growth and pertinent references when available. Upon receipt cell cultures should be placed in the incubator at 37°C for a few hours or overnight to permit recovery from damage which may have occurred during shipment.

8. REQUEST FOR CITATION AND REPRINTS

It will be greatly appreciated and will make the cell collection more valuable if the source and Repository number is cited in publications in which cell cultures from the Human Genetic Mutant Cell Repository are used. The Repository would appreciate receiving a reprint of such publications.

9. PUBLICATION OF DESCRIPTIVE ARTICLES ON CELL CULTURES STORED IN THE REPOSITORY

Concise descriptions of some of the cultures stored in the Repository are published after characterization in Cytogenetics and Cell Genetics under the name of the individual originally submitting the culture or biopsy. These descriptions include brief clinical histories, assay methods when applicable, family pedigrees, full or partial karyotypes in the case of chromosomal aberrations, and references to previous work involving the cell lines. For a listing of these concise papers, see Appendix C, page 153.

10. ASSURANCE FORM

Before cells can be shipped from the Genetic Mutant Cell Repository the recipient institution must submit a purchase order number and agree to the limitations listed below. This agreement must be renewed annually. Please sign and return original to the Institute for Medical Research, Copewood Street, Camden, New Jersey 08103.

ASSURANCE

As purchaser of cell cultures from the Genetic Mutant Cell Repository we agree that such cells, their progeny, or derivatives will not be used in human experimentation. It is further agreed that if such use is planned, the purchaser will first obtain prior written approval of the Project Officer, N01-GM-6-2119, National Institute of General Medical Sciences.

It is further agreed that cell cultures obtained from the Genetic Mutant Cell Repository will not be resold, but they may be replicated by a third party for the original purchaser. The third party shall not be allowed to resell other than to the original purchaser furnishing the material for replication and the third party shall not use such cells for human experimentation.

Name of Institution

Name of Principal Investigator

Date

Signature of Authorized
Official

Signature of Principal
Investigator

Date

11. AGREEMENT ON LYMPHOID AND VIRUS TRANSFORMED CELLS

This document must be appropriately completed and exchanged before lymphoid and virus transformed cell lines can be transferred from the Human Genetic Mutant Cell Repository to your laboratory.

AGREEMENT

The potential hazardous nature of these human cell lines is unknown. However, in view of this lack of knowledge, it is appropriate that the scientific community be made aware that a potential hazard may exist. Therefore, we recommend that the enclosed guidelines for laboratory procedure be adhered to in the handling of lymphoid and virus transformed cell lines. They are also recommended for handling all cell cultures.

It is understood that you will not further distribute cell lines sent to you to laboratories not under your direct supervision. It is further agreed that all cultures obtained from the Human Genetic Mutant Cell Repository may only be replicated by a third party if the third party also executes an Agreement on Lymphoid and Virus Transformed Cells agreeing to follow the Minimum Safety Guidelines.

As purchaser of cell cultures from the Human Genetic Mutant Cell Repository we agree that such cells, their progeny, or derivatives will not be used in human experimentation. It is

further agreed that if such use is planned, the purchaser will first obtain prior written approval of the Project Officer, NO1-GM-6-2119, National Institute of General Medical Sciences.

This agreement, when appropriately executed, will be on file and will qualify you to receive lymphoid and virus transformed cell lines from the Human Genetic Mutant Cell Repository.

AGREEMENT ON LYMPHOID AND VIRUS TRANSFORMED CELLS

We the signatories below, have read and understand this document and agree that in the handling of cell lines furnished, we will adhere to the procedures and recommendations outlined in the foregoing and the attached "Minimum Safety Guidelines Recommended for Working with Lymphoid and Virus Transformed Human Cell Lines."

A. For the recipient laboratory:

Investigator: Signature _____ Date _____
Title _____

Head of Laboratory or Immediate Supervisor: Signature _____ Date _____
Title _____
Address _____

B. For Human Genetic Mutant Cell Repository:

Signature _____ Date _____
Title _____

III. HUMAN GENETIC MUTANT CELL REPOSITORY

MINIMUM SAFETY GUIDELINES

RECOMMENDED FOR WORKING WITH LYMPHOID

AND VIRUS TRANSFORMED HUMAN CELL LINES*

A. Supervision

1. Administrative Responsibilities

a. Responsibility of Management

Management should establish a biohazards committee to institute and enforce a health and safety policy which includes a specific safety program for work involving human cell lines. The program should meet applicable federal, state and local regulations and include safety training, maintenance of accident records, and provisions for emergency treatment.

b. Responsibility of the Principal Investigator

The principal investigator is responsible for the preparation of safety protocols for the research program under his direction. The protocols should include appropriate procedures for use, storage, decontamination, disposal and emergency treatment. The protocols should be approved by the biohazards

*These guidelines were developed on February 11, 1974 at a special meeting at the National Institutes of Health attended by representatives from the Office of Biohazards of NCI, NIAID, NIGMS and the Advisory Committee to the Human Genetic Mutant Cell Repository.

committee and discussed with the research staff before starting the research program.

2. Medical Surveillance and Screening

a. Physical Examinations

Appropriate pre-employment and periodic medical examinations are desirable for persons working with human cell lines.

b. Work Restrictions

Persons having reduced immunologic competency should be restricted from working with these human cell lines.

c. Serum Collection

Serum should be collected at the time of the pre-employment physical to establish a baseline reference. Serum should be re-collected and stored annually. Serum should be collected immediately after accidental injection or ingestion and at an appropriate interval thereafter. For those individuals exposed to long term lymphoid lines or their derivatives, anti-EB virus titers should be obtained on the collected sera.

3. Laboratory Access

Access to the cell culture area should be restricted to persons directly working with the cell lines, or by specific authorization by the principle investigator or director of the laboratory.

B. Personnel Practices

1. Pipetting

Mechanical pipetting aids rather than mouth pipetting should be used for all pipetting procedures.

2. Eating, Drinking and Smoking

Eating, drinking and smoking should not be allowed in the same areas where cell lines are under study.

3. Protective Clothing

It is recognized that the criteria for protective clothing may vary according to the physical situation of the laboratory and the agents handled. Ideally, adequate protective clothing such as a fully fastened laboratory coat should be worn. This clothing should not be worn outside the work area once the work area has been entered.

C. Physical Control Practices (Recommended for all cell lines, but required for long term lymphoid lines and their derivatives).

1. Ventilated Safety Cabinets or Hoods

Ventilated safety cabinets and hoods and other safety apparatus should be employed and should be tested at least annually to certify correct containment and operation. A list of specifications for satisfactory hoods and instructional materials may be obtained from the Office of Biohazards, NCI.

2. Housekeeping

Appropriate housekeeping procedures which suppress the

formation of aerosols should be used. Working surfaces should be wiped down with an appropriate disinfectant before and after work with each cell culture and at the end of the working day.

3. Decontamination and Disposal

Contaminated glassware and similar materials should be appropriately decontaminated or stored for decontamination before removal from the work area for recycling or disposal. Liquid wastes should be decontaminated either chemically or by heat, before being discharged to the community sanitary sewer system.

4. Protection of Vacuum Lines

Vacuum services, if used, should be protected with disposable absolute air filters and liquid traps. The effluent should be collected in liquid traps containing concentrated disinfectant.

References

Biohazards in Biological Research, ed. A. Hellman, M.N. Oxman and P. Pollack. Cold Spring Harbor Laboratory, New York (1973).

National Cancer Institute Specification: General Purpose Clean Air Biological Safety Cabinet.

IV. SUBMISSION OF SPECIMENS TO THE REPOSITORY

Biopsies are preferred to established cell cultures because they can be processed and stored in lower passage. When submitting a specimen to the Repository, the biopsy should be placed in a 25 cm² tissue culture flask or screw top vial with culture medium containing 100 units/ml of penicillin and 100 mcg/ml of streptomycin. Tape the top or cap of the flask very securely to prevent leakage. Package the biopsy flask in a container so that it will not be broken in transit. Mark on the outside of the package that it should be kept at room temperature and not refrigerated, frozen or overheated.

Early passage cultures may be submitted when a biopsy cannot be obtained from the patient. The culture flask (25 cm² preferably) should be filled to the top with culture medium and shipped as described above. Mail biopsy or culture the same day by air mail special delivery to:

Dr. Arthur E. Greene
Human Genetic Mutant Cell Repository
Institute for Medical Research
Copewood and Davis Streets
Camden, New Jersey 08103
Phone 609-966-7377

Before a biopsy or cell culture can be processed for the

Repository, documented proof of the diagnosis must be provided on a submission sheet which is available from the Institute for Medical Research, or may be photocopied by the submitter from pages 18-22. Without this information the culture cannot be coded into the information retrieval system or be of value to users of the Repository; and to conserve effort and expense the Repository staff are instructed not to process a specimen unless documentation of diagnosis is provided with the specimen. Submitters are therefore requested to fill in all applicable blanks on the submission sheets. Please provide a family genealogy if appropriate.

No biopsies or cell cultures submitted to the Human Mutant Cell Repository are to be obtained from a live fetus, defined by the presence of pulse, circulation and other vital signs.

TO BE FILLED IN BY IMR

Date Received _____ / _____ / _____
Mo. Day Year

GM # _____

Contamination ? _____

Failure ? _____

SPECIES: ☒ 1 HUMAN (Go to Item 1) ☐ 2 OTHER (Go to Item 8)
(Specify)

2A. Date of Birth / / or Age: Days Wks. Mos. Yrs.
Mo. Day Year (Circle One)

(Specify)

1	Personal Examination	5	Autopsy Records
2	Hospital Records	6	Private Physician
3	Genetic Clinic Records	7	Other _____
4	Specialist's Report		(Specify)

Date Obtained _____
Mo. Day Year

18

11. Culture Medium in which Submitted:

☐ 1	McCoys	_____ %	☐ 6	RPMI-1640	_____ %
☐ 2	Eagle-Hanks'	_____ %	☐ 7	Fetal Calf Serum	_____ %
☐ 3	Eagle-Earle	_____ %	☐ 1	Inactivated	
☐ 4	Ham's F-12	_____ %	☐ 2	Uninactivated	
☐ 5	Ham's F-10	_____ %			
☐ 8	Other	_____			

(Specify) _____

12. Addition of Following to Culture?

Antibacterial Agents?

- ☐
- 1 No
-
- ☐
- 2 Yes

Antifungal (Yeast)?

- ☐
- 1 No
-
- ☐
- 2 Yes

Other Additives?

- ☐
- 1 No
-
- ☐
- 2 Yes (List) _____

- ☐
- 1 Penicillin _____ %
-
- ☐
- 2 Streptomycin _____ %
-
- ☐
- 3 Gentamycin _____ %
-
- ☐
- 4 Other _____ %

- ☐
- 1 Fungizone _____
-
- ☐
- 2 Mycostatin _____
-
- ☐
- 3 Other _____

(Specify) _____

13. Special Instruction(s) on Handling or Freezing of Culture/Biopsy?

- ☐
- 1 No
- ☐
- 2 Yes

(Specify) _____

14. Have Chromosome Studies ever been Done on this Individual?

- ☐
- 1 Information Not Available (Go to Item 22)
-
- ☐
- 2 No (Go to Item 22)
-
- ☐
- 3 No, but Studies are Underway (Go to Item 22)
-
- ☐
- 4 Yes
-
- ☐
- 1 On Cells from this Culture
-
- ☐
- 2 On Cells from Other Tissue/Samples

15. General Karyotype Results: ☐ 1 Normal

- ☐
- 2 Abnormal
-
- ☐
- 1 Balanced
-
- ☐
- 2 Unbalanced
-
- ☐
- 3 Not Certain

16. Sex Chromosomal Complement: (Check One or More)

☐ 1 XX	☐ 2 XO	☐ 3 XXX
☐ 4 XY	☐ 5 XXY	☐ 6 XYY
☐ 8 Other		☐ 7 XXXY

(Specify) _____

17. Autosomal Mosaicism?

- ☐
- 1 Detected (If so, # and % Sub Lines: _____)

- ☐
- 2 Not Detected
-
- ☐
- 3 Suspected

18. Breakage/Somatic Rearrangements?

☐ Data Not Available☐ Data Available☐ Breaks Observed☐ Somatic Rearrangements Observed☐ Neither Observed

19. Current International Nomenclature: _____

20. The Above Karyotype is Based on ☐ Banded or ☐ Unbanded Technique(s).

21. If Banded, what Staining Method(s) was Used: _____

22. Have Biochemical Studies been Done on this Individual

☐ No (Go to Item 29)☐ Yes☐ On Cells from this Culture (Go to Item 23)☐ On Other Cultures or Tissues from Same Patient (Go to Item 28)

23. Specific Defect Detected or Investigated: _____

24. Assay Method Used: _____

25. Assay Level in this Cell Culture: _____

26. Assay Level in Normal Control Cell Cultures _____

27. Conclusion Based on Biochemical Assay:

☐ Normal☐ Abnormal☐ Inconclusive☐ Consistent with Heterozygosity☐ Consistent with Homozygosity☐ Genotype Uncertain☐ Not Applicable

28. Significant Laboratory Data on Other Cell Cultures or Tissues from this Individual:

Tissue	Assay	Level	Conclusion (See Item 27)

29. Is Pedigree Available? ☐ No ☐ Yes

Relative	Chrom. Studies Done?	Biochem. Studies Done?	Clinical Lab Studies Done?	Cultures Available?	Banked in Camden?
Father					
Mother					
Siblings					
Offspring					
Other (Specify)					

31. Has this Patient/Family been Reported in Literature?

☐ No ☐ Yes

Specify Reference _____

32. Has a Sample from this Patient been Previously Stored in Camden?

☐ No ☐ Yes, GM # _____.

33. Any Additional Comments/Observations/References/Lab Studies on Culture, Patient, Family Etc.?

Enclosures: ☐ Pedigree ☐ Clinical History/Description of Patient and/or Family

☐ Karyotype ☐ Laboratory Data on Patient/Family

☐ Reprint of Reference ☐ Other _____

(Specify)

To encourage storage of unusual cell cultures in the Repository, provision has been made for delayed release to other investigators if the contributor so desires. Please check your preference: a) release culture to anyone requesting it_____, b) release only to contributor during the first year_____. At the conclusion of 1 year the cell culture will be listed in the next printing of the catalog and made available to other investigators unless additional time is specifically requested.

I hereby grant permission for these cells to be stored in a bank of genetic mutant cell cultures and the progeny cells distributed to qualified investigators. Appropriate consent was obtained from the patient from whom the cells were originally obtained, or can be reasonably inferred, for use of these cells for diagnosis, research, teaching or therapy.

No biopsies or cell cultures submitted to the Human Mutant Cell Repository are to be obtained from a live fetus, defined by the presence of pulse, circulation and other vital signs.

Date _____ Submitter _____
(Signature)
Address _____

Telephone Number _____

Mail completed form with, or preferably preceeding shipment of cell cultures, to: Dr. Arthur Greene, Institute for Medical Research, Copewood and Davis Streets, Camden, New Jersey 08103.
Phone: 609-966-7377.

EXPLANATION OF THE CODE INTERPRETING RECORDED

DATA IN THE COLUMNS OF THE CATALOG

COLUMN

GM #: The GM number of the cell culture, refers to the number assigned to the culture when it was received at the Human Genetic Mutant Cell Repository. Fibroblast cultures are all originated from skin unless noted in the Remarks column. Lymphocyte cultures are all established from peripheral blood. McKusick's number is inserted following the name of each disease.*

Passage #: The number of serial in vitro transfers of the cell culture as stored in liquid nitrogen.

Culture Medium: For code see appendix A, page 131.

Submitter Code: A number which identifies the investigator who submitted the biopsy or culture to the Repository. For code see appendix B, page 133.

Age: Age of the donor is expressed in years, or when appropriate in months (mo.), weeks (wk.) or days (da.). F indicates fetus.

Sex: M or F

Race: W-Caucasian; B-Black; O-Oriental; I-Indian (India); P-Puerto Rican. Space left empty if race unknown.

*McKusick, Victor. Mendelian Inheritance in Man 4th Edition.

John's Hopkins Press, Baltimore, London. 1975.

<u>Genetic Status:</u> +	Normal gene
-	Affected gene
y	Hemizygous for X-linked trait
?	At risk for autosomal trait, genetic status not determined
(0)	Carrier for trait as determined by pedigree or clinical diagnosis

The McKusick* number immediately following the name of the disease will indicate the dominant, recessive, or X-linked nature of the disease. The first digit is one for a dominant disease (e.g. Huntington Chorea - 14310), two for a recessive disease (e.g. Tay-Sachs Disease - 27280), and three for an X-linked trait (e.g. Lesch Nyhan Syndrome - 30800).

Verified: B if the defect was verified on the cell culture before freezing, A if verified after recovery from liquid nitrogen. Space left blank if verified on another cell culture or tissues from the same patient or relative, if not verified at all, or if verification is not yet possible because defect is unknown or not yet expressed in culture.

Paris Nomenclature: As described (Paris Conference 1971) and Supplement (1975): Standardization in Human Cytogenetics.

Birth Defects: Original Article Series, VIII:7, 1972. The National Foundation, New York.

*McKusick, Victor. Mendelian Inheritance in Man 4th Edition. John's Hopkins Press, Baltimore, London. 1975.

Balanced, Unbalanced: Balanced (B) or Unbalanced (U) karyotype

Remarks: Any pertinent information not included in preceding columns.

*after GM #: Means a description of this cell culture has been published in Cytogenetics and Cell Genetics. Reference numbers appearing in parentheses at the bottom of the page refer to these publications listed in Appendix C, pages 153-160.

Aging Cell Repository: Cell cultures listed under this heading are of particular interest to investigators interested in studies on aging. Some cultures originally deposited in the GM Repository are also of interest to the Aging Repository. For convenience these cell cultures are listed in both the GM and the Aging (AG) Repository. Many new cell cultures are now being added to the Aging Repository and receive an AG #. Double listings will be made when appropriate. Requests for these cells may be addressed to the Director of the Aging Repository:

Dr. Warren W. Nichols
Institute for Medical Research
Copewood Street
Camden, New Jersey 08103
Telephone 609-966-7377

Procedures and charges are the same as for the Genetic Mutant Cell Repository. The Aging Repository is supported by the National Institute of Aging.

HUMAN FIBROBLAST CULTURES WITH
BIOCHEMICAL MUTANT CONDITIONS

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
DISORDERS OF AMINO ACID METABOLISM									
Alkaptonuria - 20350									
2228	4	B	147	36	M	W	--		
Argininemia - 20780									
954	5	C	6	4	F	W	--		
Argininosuccinicaciduria - 20790									
525	8	C	123	9	M	W	--	A	] Family
533	8	C	123	20	M	W	--	A	
540	8	C	123	63	M	W	+-	B	
Citrullinuria (Citrullinemia) - 21570									
63	17	S	95	9 mo.	F	W	--	B	ATCC CCL 76
1044*	3	A	107	8 mo.	M	W	--	A	Proband; see GM-1204 Lymphoid
1058	3	A	107	27	M	W	+- (O)		Father; see GM-1206 Lymphoid
1679	2	B	157	1 da.	M	W	--		A sib died of Citrullinemia
1684	6	C	114	1 da.	F	W	--	B	See also GM-1685 Lymphoid
Cystinosis									
Type I - (Early Onset Nephropathic Type; Infantile Type) - 21980									
8	5	A	121	5	F	W	--	A	
18	4	A	121	3	M	W	--	A	
20	4	A	121	5 mo.	M	W	--	A	
46	5	A	121	3 1/2	M	W	--	A	
489	10	G	114	1 1/2	M	W	--		Sib of GM-304

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #46

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
<u>DISORDERS OF AMINO ACID METABOLISM</u>									
<u>Cystinosis</u>									
<u>Type I, continued</u>									
304	4	A	114	6 mo.F	M	W	--		Proband, skin; formerly GM-91 Lung Umbilical Cord
90	3	A	114	6 mo.F	M	W	--		Same fetus
93	4	A	114	6 mo.F	M	W	--		
706	3	A	117	1 1/2	M	W	--		
760	2	A	128	6 mo.F	M	W	--		Fibroblasts also available from bone marrow, kidney, thymus, liver, thyroid and amnion
2066	2	B	19	1	F	W	--	A	Biopsy taken at autopsy
<u>Type III - (Benign Type; Adult Type) - 22000</u>									
378	8	G	114	7	F	W	--	B	Sib
379	8	G	114	4	M	W	--	B	Sib
906	11	B	114	32	M	W	+- (O)		Father
907	15	B	114	30	F	W	+- (O)		Mother
<u>Type Unclassified</u>									
908	8	B	114	12	M	W	--	B	Nephropathic
909	9	B	114	42	F	W	+- (O)		Mother
910	9	B	114	49	M	W	+- (O)		Father
<u>Homocystinuria (Cystathionine Synthase Deficiency) - 23620</u>									
342	3	A	66	20	F	W	--		B6 Responsive
423	8	B	114		M		--	A	
424	9	B	114		M		--	A	
584	3	A	38	39	M	W	--	A	Proband
585	3	A	38	65	M	W	+- (O)		Father

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
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DISORDERS OF AMINO ACID METABOLISM

Homocystinuria, continued

594	2	A	86	23	M	W	--	A	Proband
417	4	A	42	42	F	W	+- (O)		Mother
625	3	A	35	35	M	W	+-	A	
720	3	A	38	30	M	W	--		
721	7	A	38	11	F	W	--	B	
722	7	A	38	24	M	W	--	B	Proband
725	7	A	38		F	W	+- (O)		Mother
724	14	A	38		M	W	--		
751	3	A	86	19	M	W	--	A	Sib
752	3	A	86	23	M	W	--	A	Sib
976	3	C	86	6	F	W	+	A	Sib
753	3	C	86	41	M	W	+-	A	Father
813	2	C	38	13	M	W	+-	A	
864	12	A	38	13	F		--	A	
865	5	A	38		M		--	A	
866	8	A	38	10	F		--	A	Sib
867	10	C	38	6	M		--	A	Sib
885	3	C	86	17	M	W	--	A	Proband
883	2	C	38	42	F	W	+- (O)		Mother
1128	3	C	38	11	F	W	--	A	Proband; B6 non-responsive
1129	3	C	38	9	M	W	--		Sib
1126	3	C	38		M	W	+- (O)		Father
1374	4	A	38	13	F	W	--	A	Proband
1375	6	A	38		F	W	+- (O)		Mother
1376	6	A	38		M	W	+- (O)		Father

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
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DISORDERS OF AMINO ACID METABOLISM

Hyperglycinemia, Non-Ketotic - 23830

747	3	A	107	1 1/2	F	W	--		
880	5	C	95	21	M	W	--		
1140	3	A	95	16	M	W	--		
1162	3	A	95	28	M	W	--		Sib Sib Sib Family
1297	8	B	114	6	M		--		
1298	8	B	114	4 1/2	M		--		
1299	7	B	114	2	M		--		
1300	8	B	114	2 1/2	F		--		
1301	8	G	114	2 mo.	F	W	--		
1302	9	B	114	1	F		--		

Hypermethioninemia - 23890

911	2	C	86	3 mo.	M	W	--		
-----	---	---	----	-------	---	---	----	--	--

Hyperphenylalaninemia

6	7	A	95	5	F	W	--		
4	9	A	95	33	M	W	+- (O)		
7	7	A	95	30	F	W	+- (O)		Proband Father Mother Family

Isovalericacidemia - 24350

427	7	J	13	9 da.	M	B	--		
947	5	C	2	13	M	W	--		B

Maple Syrup Urine Disease - (Branched-Chain Ketoaciduria) - 24860

296	15	A	33	5	M	B	--		B
297	18	A	33	13 da.	F	W	--		B
16	4	A	95	25	M	W	+- (O)		A
22	5	A	95	25	F	W	+- (O)		A
612	2	A	74	2 mo.	F	W	--		A

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
<u>DISORDERS OF AMINO ACID METABOLISM</u>									
<u>Maple Syrup Urine Disease, continued</u>									
649	3	A	38	9 mo.	M	W	--		Proband Father Mother] Family
650	3	A	38		M	W	+- (O)		
651	3	A	38		F	W	+- (O)		
1000	10	A	27	13	F	W	--	B	Iraqi
1099	3	A	107	7 mo.	F	W	--		
1158	2 IMR	A	86	6	F	W	--	A	Proband, mild variant
1159	2 IMR	A	86		M	W	+- (O)		Father
1364	2	E	107	7	F	B	--		See GM-1366 Lymphoid
1557	12	G	27	5	M	W	--		Variant
1654	2	A	107	5	F	W	--	A	See GM-1655 Lymphoid; two sibs died of MSUD
1744	11	B	77	13 da.	F	W	--	A	
1938	7	B	77	5	M	W	--	A	
2327	3	B	107	4 mo.	F	W	--	A	Pericardium
<u>Methylmalonicaciduria - 25100</u>									
50	10	S	95	1	M	W	--	A	Unresponsive to B12; ATCC CGL 124
212	6	A	95	1	M	W	--	A	Responsive to B12
306	6	A	95	2	M	W	--	A	Responsive to B12
595	2	A	88	7	M	W	--		Proband; Cobalamin A mutant
596	2	A	88	25	F	W	+- (O)		Mother
930	6	B	32	2	F	W	--		Mutase defect; unresponsive to B12
876	5	I	10	1 mo.	M	W	--		Cobalamin B mutant
1673	8	J	88	1 da.	M	W	--	B	Apomutase mutant
1674	7	C	88	1 da.	F	W	--		Cobalamin A mutant
<u>Phenylketonuria - 26160</u>									
937	2	C	107	19 mo.	M	W	--		
2406	2	B	74	2 mo.	M	W	--		Pyloric stenosis

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
DISORDERS OF AMINO ACID METABOLISM									
Propionicacidemia - (Hyperglycinemia with Ketoacidosis & Leukopenia) - 23200									
56	10	A	88	1	F	W	--	B	Proband Mother Father Family
57	10	A	88	6	F	W	--	B	
371	3	A	105	2 mo.	F	W	--	B	
403	7	A	105	24	F	W	+- (O)		
405	7	A	105	25	M	W	+- (O)		
Tyrosinemia - 27670									
286	7	A	95	2 mo.	M		--		
DISORDERS OF CARBOHYDRATE METABOLISM									
Amyloidosis - Dominant									
1998	3	B	155	16	F	W	+- (O)		Non-Portuguese
Aspartylglycosaminuria - 20840									
568	2	A	4	18	F	W	--		
2056	3	B	4	32	M	W	--	B	
2057	4	B	4	26	F	W	--	B	
Fructose -1,6-Diphosphatase Deficiency - 22970									
282	6	A	95	7	F		--		
Fucosidosis - 23000									
289*	3	A	78	31	M	W	+- (O)		Father Mother Son Son Family; normal karyotypes Fucosidosis Type II See GM-1023, GM-1024, GM-1025, GM-1026, Lymphoid
290*	3	A	78	29	F	W	+- (O)		
291*	3	A	78	9	M	W	--	A	
292*	3	A	78	4	M	W	--		
801	6	C	138	15	M	W	--	B	Type II; Spanish-American

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #24

CM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
DISORDERS OF CARBOHYDRATE METABOLISM									
Fucosidosis, continued									
802	5	C	138	7 1/2	M	W	--	B	Type II; Spanish-American
1214	5 IMR	A	108	18	M	W	--		Type II
Galactosemia (Kinase Deficiency) - 23020									
334	5	A	95	22	F	W	--	A	Proband Mother Father] Family
335	3	A	95	53	F	W	+- (O)		
336	4	A	95	62	M	W	+- (O)		
Galactosemia (Transferase Deficiency) - 23040									
52*	11	S	95	8	M	B	--	B	Sib; G6PD Type B; ATCC CCL 133; see also GM-639, SV40 Trans.
1908	2	A	95	15	M	B	--	B	Same patient as GM-52
53*	12	S	95	6	M	B	--	B	Sib; G6PD Type A; ATCC CCL 132; see also GM-638, SV40 Trans.
54	10	A	95	3 mo.	M	W	--	B	Same patient] Duarte variant
1907	3	A	95	6	M	W	--	B	
264	11	J	95	2 1/2	M	W	--	B	
422	3	A	95	9	F	W	--		Sib; G6PD Type A; ATCC CCL 132; see also GM-638, SV40 Trans.
433	3	A	95	10	F	B	--		
438*	6	A	69	46	M	W	+- (O)	B	
439*	6	A	69	41	F	W	+- (O)	B	Father Mother Daughter Son Son Daughter] Family
440*	6	A	69	16	F	W	--	B	
441*	6	A	69	15	M	W	--	B	
442*	6	A	69	12	M	W	++	B	
1533	7	A	69	19	F	W	++	B	
528	4	A	95	2	F	W	--	B	
727	3	A	95	2 mo.	F	W	--	B	

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 25; 34

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 25; 34

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
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DISORDERS OF CARBOHYDRATE METABOLISM

Galactosemia (Transferase Deficiency), continued									
1208	3	C	45	27	M	W	+-	A	Father
1209	2	C	45	1	M	W	--	A	Twin; Proband
1210	2	C	45	1	M	W	--	A	Twin; Proband
1211	3	C	45	4	M	W	+	A	Son
1212	3	C	45	29	F	W	+-	A	Mother
1417	6	A	95	22	F		--		Proband
1418	5	A	95		F		+(O)		Family; slow
1419	5	A	95		M		+(O)		Father growing
1703	4	B	95	9 mo.	M	W	--	B	Cousin 2nd generation
1704	3	B	95	2	M	W	--	B	Cousin with GALT
1741	8	B	77	6 mo.	M	W	--	B	See GM-1743, amniotic fluid cell
1996*	6	J	2	1	M	W	--	A	Double heterozygote; GALT, Duarte
Glucose-6-Phosphate Dehydrogenase Deficiency and Variants (See Biochemical Markers) - 30590									
120	9	C	41	30	F	B			G6PD A/b; PGK 1,2
218	9	C	41	12	M	W			Ca. zero activity
324	5	A	125	22	M	W	Gd-/Gd-		47,XXY; Mediterranean type
325	4	A	125	30	M	W	Gd+/Gd-		47,XXY; Mediterranean type
412	3	A	9	19	M	W	Gd-		Panama type
738	7	A	41		M	W	Gd+		Hektoen variant
888	22	C	125	2 1/2	M		Gd-		New York type

Glycogen Storage Diseases

Type I - Von Gierke Disease - 23220
574 8 C 130
25 M W --

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #48

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
DISORDERS OF CARBOHYDRATE METABOLISM									
Glycogen Storage Diseases									
Type II - Pompe Disease - 23230									
244	4	A	2	5 mo.	F	W	--	A	Mexican
248	3	A	95	4 mo.	M	B	--	A	
338	2	A	8	18 da.	M	B	--		Grows poorly
443	4 IMR	A	99	30	M		--	A	Late onset
1935	3	A	126	30	F	B	--	A	
Type III - Debranching Enzyme Deficiency - 23240									
111	7	A	143	1	F	W	--	A	
226	12	A	143	13	F		--	A	
303	9	A	95	9	F	W	--	A	
573	7	C	62	16	F	W	--	B	
576	6	C	62	7	F	O	--	B	
578	18	C	62	4	M	W	--	B	
683	3 IMR	C	95	9 mo.	M		--		
1702	4	B	95	11	M		--	A	
Type IV - Andersen Disease - 23250									
572	9	C	62	2	F	W	--	A	
Type V - McArdle Disease - 23260									
577	7	C	130	39	M	W	--		
Type VI - Hers Disease - 23270									
579	9	C	62		M		--		
Types VIII, IX - Phosphorylase Kinase Deficiency - 30600									
25	4	A	95	2 1/2	M	W	y-		
575	5	C	62	3	M	W	y-		Mexican

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
DISORDERS OF CARBOHYDRATE METABOLISM									
Hyperpyruvicacidemia									
583	3	A	38	7 mo.	F	W	--		
Lactic-pyruvic Acidosis									
2224	2	B	162	1 wk.	F	W			
Mannosidosis - 24850									
654	3	A	4	7	M	W	--	A	
1851	10	A	6	3	M	W	--	B	
2049	8	G	77	39	M	W	+-	B	Father Proband Sister Family - New variant
2050	9	B	77	11	M	W	--	B	
2051	18	B	77	7	F	W	--	B	
Mucopolipidoses									
Type II - J-Cell Disease - 25250									
87	5	A	77	5 mo.	M	W	--		I-Cell disease lines are difficult to recover from liquid nitrogen Proband Same patient as GM-87 Mother Father Family
521	4	A	95	2	M	W	--		
80	5	A	95	17	F	W	++(O)	A	
81	5	A	95	23	M	W	++(O)		
164	4	A	105	5 1/2	M	W	--	A	
Type III - J-Cell Disease - 25250									
1586	9	B	77	1 mo.	M	W	--		Proband Father Mother Family
1589	7	B	77		M	W	++(O)		
1590	7	B	77		F	W	++(O)		
1494	6	A	57	16	F	W	--	A	Atypical mild variant
1742	3	B	77	4 mo.	F	W	--	B	
2013	8	U	77	1 mo.	M	W	--	B	Proband Mother
2014	8	B	77	18	F	W	+-	B	

GM #	Passage #	Culture Media	Submitter	Age	Sex	Race	Genetic Status	Verified	Remarks
DISORDERS OF CARBOHYDRATE METABOLISM									
Mucopolipidoses									
Type II - I-Cell Disease, continued									
2045	7	H	95	1 da.	M	W	--	B	Proband
2046	6	B	95	25	F	W	+-	B	Mother
2047	4	A	95	26	M	W	+-	B	Father
2145	8	B	77	26	M	W	+- (O)		Father of I-cell
2273	3	B	70	2	M	W	--		Proband
2274	3	B	70		F	W	+- (O)		Mother
Type III - Pseudo-Hurler Polydystrophy - 25260									
2065	3	B	126	9	M	W	--	B	
2425	6	C	134	15	M	W	--	B	
113	4	A	105	2 1/2	F	W	--	A	
1759	2	B	92	13	M	W	--	B	
Type IV									
2048	10	B	77	2	F	W	--	B	
Mucopolysaccharidoses									
Type IH - Hurler Syndrome - 25280									
2	7	A	95	10 mo.	M	W	--	A	Proband
3	8	A	95	26	F	W	+- (O)		Mother
34	6	A	95	9 mo.	F	W	--	A	Proband
31	4	A	95	23	F	W	+- (O)		Mother
42	5	A	95	25	M	W	+- (O)		Father

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
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DISORDERS OF CARBOHYDRATE METABOLISM

Mucopolysaccharidoses

Type IH - Hurler Syndrome, continued

415	2	A	107	4	M	W	--	A	
798	3 IMR	C	62	1	F	W	--	A	Proband Mother Father Family
799	3 IMR	C	62	22	F	W	+- (O)		
800	3 IMR	C	62	27	M	W	+- (O)		
887	5	C	57	7	F	W	--	B	
1053	5	C	57	4	M	W	--	B	
1257	7	K	62	1	F	W	--	A	
1391	2	A	70	9 mo.	F	W	--	A	Proband Mother Father Family
1392	3	A	70	24	F	W	+- (O)		
1393	3	A	70	24	M	W	+- (O)		

Type IH/S - Hurler/Scheie

512	4	A	8	15	F	O	--		
963	3	C	92	5 1/2	M	W	--	A	
1254	9	C	62	4	M	W	--	A	Sib Sib
1255	6	K	62	10	M	W	--	A	
1898	10	B	77	6	M	W	--	B	Proband Mother Father Family
2016	6	B	77	35	F	W	+-	B	
2017	6	B	77	40	M	W	+-	B	

Type IS - Scheie Syndrome - 25310

1256	9	K	62	12	M	W	--	A	
1323	3	A	92	58	M	W	--	A	

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
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DISORDERS OF CARBOHYDRATE METABOLISM

Mucopolysaccharidoses

Type II - Hunter Syndrome - 30990

39	4	A	66	9	M	W	y-	A	
47	6	A	107	9	M	W	y-	A	
140	4	C	105	5	M	B	y-	A	
298	4	A	66	2	M	W	y-	A	
614	3	A	92	18	M	W	y-	A	Proband; mild type]
613	3	A	92		F	W	+- (O)		Mother
615	3	A	92	9	M	W	y-	A	Proband; severe type]
620	3	A	92		F	W	+- (O)		Mother
690	3	A	92	14	M	W	y-	A	
862	2	C	107	11	M	W	y-	A	Mild type
901	11	C	57	5	M	W	y-	B	Proband]
902	6	C	57	28	F	W	+- (O)		Mother
1258	7	K	62	2	M	W	y-	A	
1583	7	B	77	3 1/2	M	W	y-	A	
1927	5	C	96	16	M	W	y-	B	Mild type; uncle]
1928	5	C	96	7	M	W	y-	B	Mild type; nephew]
1929	5	C	96	7	M	W	y-	B	Severe type
2268	8	A	168	5	F	W	-?	A	Rare in females

Type IIIA - Sanfilippo Syndrome, A - 25290

312	2	A	107	3	F	W	--	A	
629	3	A	66	10 1/2	M		--	A	
643	3	A	107	3 1/2	M	W	--	A	
879	7	C	57	3	F	W	--	A	Proband]
886	7	C	57	19	F	W	+- (O)		Mother
903	8	C	57	10	M	W	--	A	
934	2 IMR	C	70	7	F	W	--	A	

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
DISORDERS OF CARBOHYDRATE METABOLISM									
Mucopolysaccharidoses									
Type IIIA - Sanfilippo Syndrome, A, continued									
1094	3	A	95	5 1/2	F	W	--	A	Proband Same patient as GM-1094 Mother Father Family
1739	3	A	95	6 1/2	F	W	--		
1095	4	A	95		F	W	+-(O)		
1096	3	A	95		M	W	+-(O)		
Type IIIB - Sanfilippo Syndrome, B -25292									
156	5	A	107	7	M	W	--	A	
737	3	A	107	7	M	W	--	A	
1426	3	A	70		F		--		
Type IV - Morquio Syndrome - 25300									
593	3	A	92	7 1/2	F	W	--	A	Clinically atypical Clinically atypical Variant Family
958	2	C	92	12	M	W	--	A	
1259	9	K	62	14	F	W	--		
1361	3	A	92	43	M	W	--	A	
1602	3	A	66	11	F	W	--	B	
Type VI - Maroteaux-Lamy Syndrome - 25320									
519*	9	A	8	4	F	W	--	A	Proband; see GM-1022 Lymphoid Father Mother Sib Sib Family
520*	8	A	8	42	M	W	+-	A	
935*	6	C	8	40	F	W	+-	A	
943*	7	C	8	8	F	W	+-	A	
942*	7	C	8	5	F	W	++	A	
538	9	A	105	1 1/2	F	W	--	B	
552	11	A	105	14	M	W	--		

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #35

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
DISORDERS OF CARBOHYDRATE METABOLISM									
Mucopolysaccharidoses									
Type VII - Beta-Glucuronidase Deficiency - 25322									
151	3	A	66	4 1/2	M	W	--	A	
121*	3	A	126	3	M	B	--	A	Proband Father Mother] Family
1850	3	B	126	28	M	B	+-	A	
2074	4	B	126	21	F	B	+- (O)		
Winchester Disease - 27795									
2295	2	B	180	21	F	P	--		
Neuraminidase Deficiency									
1718	2	B	70	2 mo.	F		--		Proband Mother Father] Family
1719	3	B	70	25	F		+- (O)		
1720	2	B	70	26	M		+- (O)		
Phosphoglycerate Kinase Deficiency - 31180									
743	2	A	98	4	M	O	y-		
DISORDERS OF LIPID METABOLISM									
Carnitine Palmitoyl - CoA Transferase Deficiency									
1763	4	A	95	29	M	W	--		Formerly GN-249
Ceroid-lipofuscinosis - 16235									
741	6	C	66	5	M		--		
Cholesterol Ester Storage Disease of Liver - 21500									
863	5	C	6	11	F	W	--	B	
Fabry Disease (Diffuse Angiokeratoma) - 30150									
107	5	A	105	10	M		y-	A	
881	9	C	108	17	M	W	y-	B	
*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #32									

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #32

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
<u>DISORDERS OF LIPID METABOLISM</u>									
<u>Fabry Disease, continued</u>									
882	10	C	108	40	M	W	y-	B	
1068	9	A	32	35	M	W	y-	B	Proband
1070	8	A	32	52	F	W	+-	B	Mother
<u>Farber Lipogranulomatosis - 22800</u>									
904	14	C	105	8 mo.	F	W	--		Proband
994	11	A	105	29	F	W	+- (O)		Mother
995	11	A	105	36	M	W	+- (O)		Father
2315	8	A	175	7	F	W	--		First cousins
2314	4	A	175	6	F	W	--		Italian descent
2316	2 IMR	A	175	37	M	W	--		Proband; Irish descent
2317	2 IMR	A	175	37	F	W	+- (O)		Father
<u>Gaucher Disease</u>									
<u>Type 2 (Infantile, Cerebral) - 23090</u>									
855	10	C	138	4 mo.	F	W	--	B	
877	2	A	107	1	M	W	--	A	
878	2	C	107		F	W	+- (O)		Proband
1260	2	A	50	11 mo.	F	W	--	A	Mother
<u>Types 1 & 3 (Juvenile & Adult) - 23100</u>									
372	4	A	9	29	M	W	--		
852	5	C	138	20	M	W	--	B	
1607	3	A	47	30	M	W	--	A	
<u>GML Gangliosidosis - Type I - 23050</u>									
806	3	A	126	2 1/2	F	W	--	A	
918	18	C	49	1	M		--		From Canadian Repository

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
<u>DISORDERS OF LIPID METABOLISM</u>									
<u>GM2 Gangliosidosis</u>									
<u>AB Variant</u>									
1675	5	C	8	16 mo.	F	B	--	B	
<u>Type I - Tay-Sachs Disease - 27280, 27290</u>									
77	9	A	95	1	M	B	--	A	Non-Jewish
221	4	A	66	3	M	W	--	A	
502	2	A	66	11 mo.	M	W	--		
514	8	C	68	3	F	W	--	B	Sib, Pericardium
515	5	C	68	1	F	W	--	A	Sib
527	3	A	107	1 1/4	M	W	--		
1108	8	A	68	30	F	W	+- (O)		Mother
1109	7	C	68	31	M	W	+- (O)		Father
1110	7	C	68	10 mo.	M	W	--	B	Proband Non-Jewish
<u>Type II - Sandhoff Disease - 26880</u>									
203	5	A	107	1	M	W	--	A	Proband
204	5	A	107	18	F	W	+- (O)		Mother
294	5	I	17	2	M	W	--		
317	3	A	66	1	F	B	--	A	
470	8	C	9	1	F	W	--		
2094	2	B	181	12	M	W	--	B	Juvenile; variant
<u>Hyperlipoproteinemia</u>									
<u>Type II - Familial Hypercholesterolemia - 14440</u>									
283	5	A	45	28	F		+-	A	For types other than II, inquire Mother
376	5	A	45	6	F		+-	A	Daughter

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
DISORDERS OF LIPID METABOLISM									
Hyperlipoproteinemia									
Type II - Familial Hypercholesterolemia, continued									
488	4	A	84	11	M	W	--	A	Proband; receptor negative
483	4	A	84	37	F	W	+-	A	Mother
966*	11	S	50	12	F	W	--	A	Receptor negative; formerly GM-361*
486	4	A	84	25	F	W	--	A	Receptor negative
1116	9	J	50	7	F	W	--	A	Receptor negative
2000	2	B	50	7	F	W	--	A	Receptor negative
701	7	B	72	6	F	W	--	A	Proband; receptor negative
700	7	B	72	45	F	W	+- (O)		Mother; other family members are available upon inquiry from Repository
1915	3	B	50	13	F	B	--	A	Receptor negative
Krabbe Disease (Globoid Cell Sclerosis) - 24520									
267	7	I	8	36	F	W	+- (O)		
268	6	I	58	41	M	W	+- (O)		
853	8	C	138	1	F	W	--		
854	8	C	138	9 mo.	F	W	--		
1773	2	B	105	1	M	W	--	A	
Metachromatic Leukodystrophy									
Infantile - 25010									
78	7	A	131	3	M	W	--	A	
243	2	A	74	3 1/2	M	W	--	A	

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #26

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
<u>DISORDERS OF LIPID METABOLISM</u>									
<u>Metachromatic Leukodystrophy</u>									
<u>Infantile, continued</u>									
197	3	A	105	4	M	W	--	A	Proband
196	3	A	105	33	M	W	+-(O)		Father
754	8	A	105	37	F	W	+-(O)		Mother; grows poorly
200	4	A	105	5	M	W	+-	B	Brother
560	5 IMR	A	105	27	F	W	+-	B	Aunt
357	9	A	105	3	M	W	+-	B	First cousin
561	5 IMR	A	105	54	F	W	+-	B	Grandmother
640	10	A	105	25	F	W	+-	B	Aunt; Mother of GM-357
905	2	C	122	3	F	W	--	A	
2093	3	B	105	25	M	W	--	A	
2331	5	A	134	5 1/2	M	W	--	A	
<u>Adult - 25000</u>									
132	6	A	105	22	M	W	+-	B	Sib
133	5	A	105	19	M	W	+-	B	Sib
<u>Multiple Sulfatase Deficiency Disease</u>									
915	9	C	105	2	M	W	--	B	
2407	10	A	147	3	F	W	--	B	
<u>Niemann-Pick Disease</u>									
<u>Type A - 25720</u>									
112	3	A	95	10 mo.	M	W	--	A	
370	8	K	2	1	F	W	--	A	
406	9	A	105	2	F	W	--	A	
559	4 IMR	A	105	25	F	W	--	B	
641	19	A	105	4	M	W	--	B	
644	5	A	105	3	F	W	--	B	
898	3	A	105		W		--		

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
DISORDERS OF LIPID METABOLISM									
Niemann-Pick Disease									
Type B - 25720									
1669	12	C	105		F	W	+- (O)	B	
Type C - 25725									
110	6	A	107	9	M	W	--		
645	8	C	105	4	F	W	--	B	
Type Unspecified									
165	3	A	107	7	M	W			Clinically atypical Variant; sib Variant; sib
1612	8	C	138		M	W	--	B	
1613	7	C	138		M	W	--	B	
Refsum Syndrome - 26650									
1007	17	C	129	30	F	W	--	B	
Schilder Disease - 26910									
269	4	A	58	30	F	W	+- (O)		
Wolman Disease - 27800									
1606	4	B	154	7 mo.	F	W	--	B	Proband Mother
2211	4	H	187	21 wk.F	M	W	--		
2121	7	A	187	31	F	W	+- (O)		
DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM									
Adenosine Deaminase Deficiency (Immune Deficiency Disease) - 24275									
469	8	A	118	1	M	W	--	B	
471	4	A	103	1 1/2	F	B	--	B	
2027	9	A	118		F		+-	B	
2028	11	A	118		M		+-	B	

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM									
Adenosine Phosphoribosyltransferase Deficiency - 10260									
517	5	C	71	64	M	B	+-		Enzyme deficient in erythrocytes; normal level in fibroblasts
Inosine Triphosphate Pyrophosphohydrolase (ITPase) Deficiency									
1617	2	B	177	29	F	W			See GM-1619 Lymphoid
Lesch-Nyhan Syndrome (HGPRT Deficiency) - 30800									
377	8	A	77	8 mo.	M	W	y-	B	Proband] Family Mother]
13	7	A	95	34	F	W	+-	B	
1906	3	A	95	15	M	W	y-		Proband] Mother]
14	6	A	95	37	F	W	+- (0)		
68	14	J	27	2	M		y-	A	
152	10	B	101	9	M	W	y-	A	
158	9	B	101	3	M	B	y-	A	Proband; G6PD Type A Mother; G6PD Type AB; HGPRT + coupled with G6PD B; HGPRT - coupled with G6PD A; sister of GM-318
135	8	A	101		F	B	+-	A	
Family									
159	11	B	85	10	M	B	y-	A	Sib; G6PD Type A Sib; G6PD Type A; replaces GM-177; see also GM- SV40 Trans.
2063	10	J	101	5 1/2	M	B	y-	A	
318	13	C	101	40	F	B	+-	A	Mother; G6PD Type AB; HGPRT + coupled with G6PD B; sister of GM-135

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM									
Lesch-Nyhan Syndrome (HGPRT Deficiency), continued									
1390	5	B	90	3 mo.	M	W	y-	A	
537	7	B	31	13	M		y-	A	
1362	3	A	66	10	M	W	y-	A	
1662	2	B	66	9	M	W	y-	A	Variant - Proband Mother Sibling Sibling Family
1659	3	B	66	42	F	W	+-	B	
1661	3	B	66	14	F	W	+-	B	
1660	3	B	66	11	F	W	+-	B	
2226	3	B	66	40	F		+- (O)		Mother Proband
2227	3	B	66	12	M		y-		
2290	2	B	183		M		y-		Skin Lung same fetus; see also GM-2292 Lymphoid & GM-2338 Amniotic
2291	2	B	183		M		y-		
Orotic Aciduria - 25890, 25892									
328	22	A	125	2	M	W	--	B	Medium should contain uridine Medium should contain uridine
632	2	C	115	14	F	W	--		
Xeroderma Pigmentosum - 27870									
Complementation Group A									
518	19	C	23	1	M	W	--	B	De Sanctis-Cacchione XP25R0* ATCC CRL 1261
544	2	A	23	10	M	W	--		De Sanctis-Cacchione XP4LO
710	11	A	11	1 1/2	M	W	--	B	De Sanctis-Cacchione XP26R0
Complementation Group C									
30	3	A	110	24	M	W	--		XP3BE; ATCC CRL 1189

*Nomenclature as per 13th International Congress of Genetics 79:215-225, 1975

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM									
Xeroderma Pigmentosum									
Complementation Group C, continued									
671	4	A	23	15	M	W	--		XP8BE; ATCC CRL 1158; twin
676	3	A	23	15	M	W	--		XP9BE; ATCC CRL 1161; twin
673	4	A	23	21	M	W	--		XP4SL; sib
677	3	A	23	25	M	W	--		XP2BE; ATCC CRL 1166; sib
709	17	A	11	15	M	W	--	B	XP21RO
Complementation Group D									
434	3	A	23	28	M	W	--		De Sanctis-Cacchione XP3NE
435	3	A	23	23	F	W	--		De Sanctis-Cacchione XP2NE
Complementation Group E									
1762	15	A	11	34	F	W	--	B	XP2RO; formerly GM-708; ATCC CRL 1259
Complementation Group Undetermined									
82	5	A	44	12	F	W	--		Proband
241	11	A	44		F	W	++(O)		Mother
436**	3	A	23	22	F	W	--		XP1NE
510	9	J	23	10	M	W	--	A	XP1PW
522**	6	C	37	16	F	B	--		G6PD Type B; phosphoglucomutase I; XP2Nbi
523**	9	A	37	10	F	B	--	A	G6PD Type B; phosphoglucomutase I; XP1Nbi
936**	8	C	46	22 wk.F	F	W	--		Umbilical cord
1213	2	C	133	61	F	W	--		Variant, high repair activity
1227	3	A	133	28	M	W	--	A	XP1KC
1295	4	C	145	6	F	W	--	A	See GM-1646 Lymphoid
1389	4	A	133	21	F	W	--	A	

**Low in unscheduled DNA synthesis

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM									
Xeroderma Pigmentosum									
Complementation Group Undetermined, continued									
1509	4	A	133	38	F	W	--		
1630	2	A	173	3 1/2	M	W	--	A	Proband Father Mother Family
1631	2	A	173	29	M	W	+-	A	
1632	3	C	173	25	F	W	+-	A	
2024	2	B	170	3	M	B	--		Sib Mother Proband Family
2034	4	B	170	28	F	B	+- (O)		
2035	3	B	170	5	M	B	--		
OTHER DISORDERS OF KNOWN BIOCHEMISTRY									
Acatalasia (Acatalasemia) - 20020									
64	5 IMR	L	80		M	W	--	B	Swiss Japanese Japanese Limited supply
65	7 IMR	L	80		M	O	--	B	
66	7 IMR	L	80	60	M	O	--	B	
1931	8	A	169	73	M	W	+- (O)		
Menkes Syndrome (Kinky Hair Disease) - 30940									
220	4	A	74	2 mo.	M	W	y-	A	Sib Sib; see GM-1245 Lymphoid Cousin Family
057	3	A	107	5 da.	M	W	y-		
245	3	A	74	1 mo.	M	W	y-	B	
1981	3	B	107	2	M	W	y-	B	Proband; see GM-1982 Lymphoid Mother; see GM-1984 Lymphoid
1983	3	B	107	26	F	W	+-	B	
Porphyria									
Acute Intermittent Porphyria - 17600									
931	3	B	112	32	F	W	+-	A	Mother Son Daughter Family
932	3	B	112	4	M	W	+-	A	
933	3	B	112	1 1/4	F	W	+-	A	

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
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OTHER DISORDERS OF KNOWN BIOCHEMISTRY

Porphyria

Acute Intermittent Porphyria, continued

939	3	C	112	32	F	W	+-	A	Daughter; see GM-2135 Lymphoid Mother; see GM-2134 Lymphoid Father; see GM-2133 Lymphoid
940	2	C	112	61	F	W	++	A	
941	3	C	112	63	M	W	+-	A	
1621	2	B	112	39	F	W	+-	A	Mother Sister Son
1622	3	B	112	19	F	W	+-	A	
1623	2	B	112	16	M	W	+-	A	
1624	3	B	112	22	F	W	+-	A	Mother Son
1625	3	B	112	3	M	W	+-	A	
1647	3	B	112	44	F	W	+-	A	

Cutanea Tarda - 17610

961	3	A	112	70	M	W	+-	
977	5	B	112	81	F	W	+-	
1041	6	B	112	69	F	W	+-	
1082	2	B	112	21	M	W	+-	
1179	3	B	112	36	M	W	+-	
1482	4	B	112	58	M	W	+-	

Hereditary Coproporphyrria - 12130

962	3	C	112	59	F	W	+-	
-----	---	---	-----	----	---	---	----	--

Sickle Cell Anemia (Hemoglobin S) - 14170

2340	10	J	107	4 mo.F	F		+-	
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GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY									
Adrenal Hyperplasia (Adrenogenital Syndrome)									
Type III - 20191									
2241	2	B	107	9 mo.	F	W	--		See GM-2242 Lymphoid
Adrenoleukodystrophy (Addison Disease and Cerebral Sclerosis) - 30010									
623	8	A	105	12	M	W	y-		Proband Mother
337	3	A	105	35	F	W	+- (0)		
Agammaglobulinemia - 30030									
362	8	A	125	21	M	W	y-		
Alzheimer Disease of Brain - 10430									
364	2	A	139	53	M	W	+- (0)		Aunt; see GM-364 Aging Repository Niece
490	3	A	139	33	F	W	+	?	
Basal Cell Nevus Syndrome - 10940									
1552	2	B	104	27	M	W	+- (0)		Basal cell carcinoma; see GM-1656 Lymphoid
1577	6	B	104	39	M	W	+- (0)		
1658	3	B	104	53	M	W	+-	B	
1657	3	B	104	53	M	W	+-	B	
1725	3	B	104	58	F	W	+- (0)		Normal skin See GM-1726 Lymphoid See GM-2099 Lymphoid See GM-2139 Lymphoid
2098	3	B	104	31	M	W	+- (0)		
2138	4	B	104		F	W	+- (0)		
Beckwith-Wiedemann Syndrome (EMG Syndrome) - 22560									
359	3	A	107	8	M	W	--		
Campomelic Dwarf - 21135									
7737	9	A	142	9 da.	M	W	--		Karyotypic abnormalities

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
<u>DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY</u>									
<u>Canavan Disease (Spongy Degeneration of the CNS) - 27190</u>									
59	2	A	95	1	F		--		
60	3	A	95	2	M		--		
<u>Cartilage - Hair Hypoplasia (Metaphyseal Chondrodysplasia, McKusick Type) - 25025</u>									
1671	4	B	70	17	M		--		
<u>CNS Disorder, Unclassified, Hereditary</u>									
1358	3	A	70	15	M	W	--		6 of 16 siblings affected with unclassified leukodystrophy
<u>Chediak - Higashi Syndrome - 21450</u>									
2075	3	B	126	1 1/2	F	W	--		Clinically documented
<u>Chondrodystrophy</u>									
229	3	A	107	9 mo.	M	W			Mother also affected
<u>Cockayne Syndrome - 21640</u>									
739	3	A	115	3	F	W	--		
1098	3	C	6	20	M	W	--		
1428	9	A	90	10	F	B	--		
1629	3	B	155	10	F	W	--		B
1856	4	B	155	13	M	W	--		B
<u>Conradi Syndrome (Chondrodysplasia Punctata) - 21510</u>									
740	2	A	107	1	F	W	--		
<u>Corneal Dystrophy, Macular Type - 21780</u>									
1125	3	C	92	41	F	W	--		Corneal button
<u>Cornelia de Lange Syndrome - 21790</u>									
45	5	A	66	13	M	W	--		

Remarks

Verified

Genetic
Status

Race

Sex

Age

Submitter
CodeCulture
MediaPassage
#GM
#

DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY

Cutis Laxa - 21910

Carrier of X-autosome
translocation

--

F

66

A

4 IMR

480

--

M

147

A

3

1353

--

M

147

C

4

1377

Cystic Fibrosis (Mucoviscidosis) - 21970

--

M

107

A

3

142

--

M

59

B

11

668

--

M

60

A

3

768

--

M

60

A

2

770

--

M

25

I

7

851

--

M

59

B

8

997

--

M

59

B

8

998

--

F

59

B

14

999

--

F

5

B

10

1707

--

M

5

B

9

1009

--

M

5

B

9

1011

--

M

5

B

8

1012

--

M

5

B

12

1708

--

M

5

B

9

1013

--

M

5

G

8

1014

--

F

144

B

3

1348

--

M

178

A

5

1957

--

M

178

A

4

1959

--

M

178

A

4

1958

--

F

144

B

3

1348

--

M

178

A

5

1957

--

M

178

A

4

1959

--

M

178

A

4

1958

--

F

100

A

3

Diabetes Mellitus - 22210

--

M

178

C

4

1828

53

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
<u>DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY</u>									
<u>Diabetes Mellitus, continued</u>									
1486	3	A	34	37	M	W			Maturity onset diabetes; see GM-1241 Lymphoid
<u>Family #1 - Maturity Onset Diabetes</u>									
1122	3	C	34	33	F	W			Sib; see GM-1240 Lymphoid
1219	5	A	34	46	M	W			Sib; see GM-1246 Lymphoid
1237	3	C	34	29	M	W			Sib; see GM-1247 Lymphoid
1430	3	A	34	35	M	W			Sib; see GM-1243 Lymphoid
1955	3	A	34	44	M	W			Sib; see GM-1956 Lymphoid
1497	2	A	34		F	W			Sib; see GM-1498 Lymphoid
1435	5	A	34	22	M	W			See GM-1242 Lymphoid] Sons of
1496	3	A	34	20	M	W			See GM-1244 Lymphoid] GM-1497
1409	3	A	34	15	F	W			Maternal cousin of GM-1435 & GM-1496; only Juvenile Diabetic in family; see also GM-1410 Lymphoid
1837	4	B	34	22	F	W			A paternal second cousin of the 6 sibs; MODY type group II; see also GM-1838 Lymphoid
<u>Family #2 - Juvenile Onset with Optic Atrophy - 22230</u>									
1609	7	A	95	18	F	W			Sib; optic atrophy; see GM-1799 Lymphoid
1610	3	A	95	13	F	W			Sib; optic atrophy; see GM-1795 Lymphoid
1611	4	A	95	15	F	W			Sib; optic atrophy
1701	4	B	95	42	M	W			Normal father; see GM-1797 Lymphoid

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY									
Diabetes Mellitus, continued									
Family #3									
1872	3	A	95	12	F	W			Affected sister
1873	3	A	95	15	F	W			Affected sister
1874	4	A	95	22	F	W			Non-diabetic sister; retarded
1875	4	A	95	17	M	W			Normal brother
1876	4	A	95	18	M	W			Normal brother
1911	3	A	95	20	M	W			Affected brother
1878	4	A	95	39	F	W			Mother
1909	3	A	95	19	M	W			Affected brother; hypertension, short stature
1910	3	A	95	21	F	W			Normal sister
Dysautonomia (Riley-Day Syndrome) - 22390									
732	2	A	107	1	M	W	--		Non-Jewish
850	5	I	25	26	M	W	--		Sib; mild familial type
2341	7	C	191	17	M	W	--		Sib; mild familial type
2342	6	C	191	19	M	W	--		Sib; mild familial type
2343	6	C	191	24	F	W	--		Mild familial type
Dyskeratosis Congenita - 30500									
1774	2	B	107	6 1/2	M	W	y-		Proband; see GM-1775
1786	2	B	107	30	F	W	+- (0)		Lymphoid
1787	2	B	107	78	F	W	+- (0)		Mother
									Great grandmother
Dystonia Musculorum Deformans - 12810, 22450									
2215	4	B	184	30	M		+- (0)		See GM-2217 Lymphoid
2255	3	B	184	14	F		--		Recessive form; see GM-2256
2304	3	B	184	16	F		+- (0)		Lymphoid
2306	2	B	184	13	M	W	+- (0)		See GM-2305 Lymphoid
									See GM-2307 Lymphoid

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY									
Ehler-Danlos Syndrome									
Type I - 13000									
1812	3	B	171		F		+-	B	
2007	6	B	171		F		+-	B	
Type II - 13000									
1691	3	B	171	19	M	W	+-	B	Same patient]
2006	3	B	171	19	M	W	+-	B	
1788	2	B	171	55	M	B	+-	B	
Type IV - 13005									
2207	21	K	147	18	F	W	+- (0)		
Type VI - (Hydroxylysine-deficient Collagen) - 22540									
1790	15	C	147	12	F	W	--		Sib Sib]
1791	15	C	147	9	F	W	--		
Type Unclassified									
733	3	A	143	7	M	W			
	3	C	95	9	F				
2293	2	B	185	14	F	W			
Giant Mitochondrial Disease - 25540									
28	4	A	95	21	F	W	--		
Gouty Arthritis									
432	3	A	120	32	M	W			
Huntington Chorea - 14310									
305	5	A	35	56	F	W	+- (0)		
1061	2	A	136	51	M	W	+- (0)		

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
<u>DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY</u>									
<u>Huntington Chorea, continued</u>									
1083	3	A	136	47	F	W	+- (O)		
1085	3	A	136	44	M	W	+- (O)		
1136	4	A	136	45	M	W	+- (O)		
1168	3	C	136		M	W	+- (O)		
1169	2	C	136	50	M	W	+- (O)		
1170	4	C	136	35	F	W	+- (O)		
1171	3	C	136	37	F	W	+		
1187	3	A	136	43	M	W	+- (O)		
2077	7	B	136	25	F	W	+		
2079	3	B	136	48	F	W	+- (O)		
2147	6	B	186	55	M	W	+- (O)		
2149	3	B	186	54	F	W	++		
2151	3	B	186	26	F	W	+		
2153	3	B	186	40	F	W	++		
2155	3	B	186	20	F	W	+		
2157	4	B	186	16	F	W	+		
2159	3	B	186	19	F	W	+		
2161	3	B	186	21	M	W	+		
2163	3	B	186	12	M	W	+		
<div> <div>Sib</div> <div>Sib</div> </div> <div> <div>Daughter</div> <div>Mother</div> </div> <div> <div>Proband; see GM-2146</div> <div>Lymphoid</div> <div>Normal spouse, see GM-2148 Lymphoid</div> <div>Daughter; see GM-2150</div> <div>Lymphoid</div> </div> <div> <div>Normal widow of deceased proband; see GM-2152</div> <div>Lymphoid</div> <div>Daughter; see GM-2154</div> <div>Lymphoid</div> <div>Daughter; see GM-2156</div> <div>Lymphoid</div> <div>Daughter; see GM-2158</div> <div>Lymphoid</div> <div>Son; see GM-2160 Lymphoid</div> <div>Son; see GM-2162 Lymphoid</div> </div> <div>Family</div>									

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY									
Huntington Chorea, continued									
2165	2	B	186	55	M	W	+- (O)		Proband
2177	6	B	186	26	M	W	+		Son; see GM-2176 Lymphoid
2183	3	B	186	21	F	W	+		Daughter; see GM-2182 Lymphoid
2169	3	B	186	52	M	W	++		Normal spouse of GM-2166 Lymphoid (affected sister of proband); see also GM-2168 Lymphoid
2187	3	B	186	60	F	W	+		Sister of proband; see GM-2186 Lymphoid
2189	2	B	186	63	M	W	++		Normal spouse of GM-2187; see GM-2188 Lymphoid
2171	2	B	186	22	F	W	+		Daughter; see GM-2170 Lymphoid
2173	3	B	186	51	F	W	+- (O)		Proband; see GM-2172 Lymphoid
2175	2	B	186	55	M	W	++		Normal spouse; see GM-2174 Lymphoid
Hypophosphatasia - 14630									
1571	3	A	95		M	B	+-		Fetal
Ichthyosis Congenita - 24230									
222	4	A	116	2 da.	F	W	--		
Incontinentia Pigmenti - 30830									
492	4	A	78	24	F		y-		
1236	9	B	85	3	F	W	y-		

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY									
Leigh Encephalomyelopathy - 25600									
1503	2	A	45	3 1/2	F	W	--		Infantile subacute
Lowe Oculocerebrorenal Syndrome - 30900									
1676	2	B	107	10	M	W	y-		See Aging Repository
Marfan Syndrome - 15470									
35	4	A	26	12	M	W	+- (O)		Sib Sib
36	4	A	26	10	M	W	+- (O)		
Mental Retardation, X-Linked - 30950									
1228	3	C	66	2	M	W	y-		
Muscular Dystrophy									
Becker Type (Progressive Tardive) - 31010									
2298	8	K	202	18	M		y-		
Duchenne Type (Pseudohypertrophic Progressive) - 31020									
2339	8	A	88	20 wk.F	F	M	y-		
Myositis Ossificans Progressiva - 13510									
513	3	A	8	16	M	W	+- (O)		Atypical; See GM-1634, Lymphoid Normal skin; other fibroblasts also available; see also GM-1641 Lymphoid
783	3	A	107		M	B	+- (O)		
Neurofibromatosis (Von Recklinghausen Disease) - 16220									
622	3	A	95	8	M		+- (O)		
1633	3	B	104	61	M	W	+- (O)		
1639	3	B	104	19	F	B	+- (O)		

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY									
Neurofibromatosis (Von Recklinghausen Disease), continued									
1860	6	B	104	41	M	W	+- (0)		Normal skin; other fibroblasts also available; see also GM-1861 Lymphoid
Osteogenesis Imperfecta - 16620									
744	8	C	57	1 da.	F	W	+- (0)		Genetic or acquired Gingiva From skin of non-lesioned leg From skin of lesioned leg Same patient
1093	7	A	95	8	M	W	+- (0)		
1436	3	A	147	24	M	W	+- (0)		
Papular Mucinosis									
948	3	A	124	60	M	W			Genetic or acquired Gingiva From skin of non-lesioned leg From skin of lesioned leg Same patient
950	2	C	124	60	M	W			
951	3	C	124	60	M	W			
Potter Syndrome (Renal Agenesis) - 26670									
630	3	A	95	1 da.	M		--		Multiplication rate slow From Canadian Repository Atypical Atypical
869	3	C	95	1 da.	F	W	--		
Progeria (See Aging Repository) - 26140									
917	14	C	49	17	F		--		From Canadian Repository From Canadian Repository
989	11	A	55	20	M		--		
990	9	C	55		M		--		From Canadian Repository From Canadian Repository
991	8	A	55	4	M		--		
1177	8	C	49	9	M		--		From Canadian Repository From Canadian Repository
1178	18	C	49	34	M		--		
1972	8	B	91	14	F		--		

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY									
Retinoblastoma (See Aging Repository) - 18020									
913	3	C	65	2	M	W	+	+	Skin Conjunctiva Proband; patient also has osteosarcoma Mother
914	3	C	65	2	M	W	+	+	
1879	2	B	155	11	F	W	+- (0)		
1880	3	B	155	37	F	W	+- (0)		
Schizophrenia and Psychiatric Disorders - 18150									
1792	3	B	137	26	M	W			Affected son; see GM-1793 Lymphoid
1833	4	B	137	56	M	W			Proband; atypical psychosis; see GM-1834 Lymphoid
1835	3	B	137	27	F	W			Affected daughter; see GM-1836 Lymphoid
1882	3	B	137	25	F	W			Normal daughter; see GM-1883 Lymphoid
1824	3	B	137	56	M	W			"Carrier" for schizo.; father of GM-1827 Lymphoid: see GM-1825 Lymphoid
1846	5	B	137	20	M	W			Normal first cousin of GM-1827 Lymphoid; see GM-1847 Lymphoid
1844	3	B	137	55	M	W			Affected father of GM-1846; see GM-1845 Lymphoid
Sea-Blue Histiocyte Disease - 26960									
843	3	C	66	10	F	W	--		Sib
844	3	C	66		M	W	--		Sib
1912	3	B	163	24	F	W	--		See GM-1913 Lymphoid
Spinocerebellar Ataxia									
1960	5	B	105	15	F	W			Culture grows slowly

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY									
Testicular Feminization Syndrome - 31370									
1404	4	C	109	20	F	B	y-		X-Linked 46XY
1628	2	A	125	1	F	W	y-		46XY; proband
1948	3	B	125	27	F	W	+- (O)		Mother
1721	2	A	107	15	F	W	y-		46XY
2300	2	B	107	8 mo.	F	B	y-		46XY; skin
2301	4	B	107	8 mo.	F	B	y-		46XY; gonad Same patient
Thanatophoric Dwarfism - 27365									
1422	2	A	107	2 wk.	F	B	--		
Tuberous Sclerosis - 19110									
1643	3	B	104	17	M	B	+- (O)		Normal skin; other fibroblasts also available; see also GM-1636
1644	2	A	104	20	F	B	+- (O)		Lymphoid
									Normal skin; other fibroblasts also available; see also GM-1638
									Lymphoid
Werdnig-Hoffmann Disease									
232	2	A	78	7 mo.	M	W	--		
Wilson Disease (Hepatolenticular Degeneration) - 27790									
32	4	A	95	9	F	W	--		
33	4	A	95	16	M	W	--		
Wiskott-Aldrich Syndrome - 30100									
1598	2	A	107	4	M	W	y-		
Zellweger Syndrome - 21410									
228	3	A	107	2 mo.	M	W	--		

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
SYNDROMES WITH INCREASED CHROMOSOME BREAKAGE									
Ataxia-Telangiectasia - 20890									
367	3	A	107	17	M	W	--		Lung cells
647	7	C	133	17	M	W	--	A	
648	9	C	133	15	M	W	--	A	
1588	13	B	77		M	W	--	A	
1740	18	B	77		M	W	--		
1829	10	C	133	7	M	W	--		
1841	11	C	133	13	M		--		
1936	6	G	77	16	F	W	--		Sib
1937	9	G	77	24	F	W	--		Sib
1970	2	B	155	3	M	W	--		Proband
1977	3	B	155	30	M	W	+- (O)		Father
1986	2	B	155	28	F	W	+- (O)		Mother
2052	3	B	77	15	F	W	--		
Bloom Syndrome - 21090									
811	17	N	44	4	M	W	--	A	
1492	11	B	77		M	W	--		
1493	10	B	77	3	M	W	--		
1620	22	C	44	8 mo.	F	W	--		
Fanconi Anemia (Pancytopenia) - 22765									
368	6	J	133	8	M	B	--	A	
369	11	A	133	6	M	W	--		
391	6	J	133	6	F	W	--		
646	7	C	133	16	F	W	--		

See Aging Repository

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
SYNDROMES WITH INCREASED CHROMOSOME BREAKAGE									
Fanconi Anemia (Pancytopenia), continued									
1309	3	C	133	12	M	B	--		
449	10	J	44	6	F	W	--		
1746	5	A	77	15	M	W	--		A
2053	4	B	77	12	F	W	--		
2061	8	A	133	20	F	W	--		
2361	8	K	188	14	M		--		
2362	11	K	188	14	M		--		
2363	10	K	188	15	M		--		
2364	12	K	188	24	M		--		

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Veri-	Remarks
TRANSLOCATIONS										
Chromosome 1										
97*	3	A	107	1	F	W	46,X,t(X;1)(Xpter>Xq26::1q12>lqter;lpter>lq12::Xq26>Xqter)	B	A	De novo translocation, Beckwith-Wiedemann Syndrome
860	11	B	36	13	F	W	46,XX,der(5),ins(5;1)(q13;q25q32)pat	U	A	Proband; grows slowly
861	7	B	36	40	M	W	46,XY,ins(5;1)(5pter>5q13::1q25>lq32::5q13>5qter;lpter>lq25::1q32>lqter)	B	A	Father
1421	12	A	109	53	M	B	46,XY,t(1;6)(q42;q21)	B	A	
126*	4	A	56	33	M	W	46,XY,t(1;15)(1qter>1p36::15q1>15qter;15pter>15q1::1p36>1pter)	B	A	
201*	10	A	30	13	F	W	47,XX,+21,t(1;17)(1pter>1p32::17p13>17qter;17pter>17p13::1p32>lqter)	B	A	
1356	2	A	66	26	F	W	46,XX,t(1;7)(1pter>1p34::7p13>7qter;7pter>7p13::1p34>lqter)	B	A	

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 7; 11; 10

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Veri- fied	Remarks
<u>TRANSLOCATIONS</u>										
<u>Chromosome 1, continued</u>										
1229	3	B	7	3	F	B	46,XX,-1,+der(1), t(1;2)(p36;q31)pat	U	A	Proband Father
1230	2	B	7	44	M	B	46,XY,t(1;2)(lqter▶ 1p36::2q31▶2qter; 2pter▶2q31::1p36▶ 1pter)	B	A	
1550	6	A	140	31	M	W	46,XY,t(1;3;4)(1pter▶ 1q32::3p21▶3pter; 1qter▶1q32::3p21▶ 3q29::4p14▶4pter; 4qter▶4p14::3q29▶ 3qter)	B	A	
257*	3	A	2	26	F		46,XX,t(1;2)(1pter▶ 1q32::2p23▶2pter; 1qter▶1q32::2p23▶ 2qter)	B	A	
1564	9	C	151	28	F	W	45,XX,t(1;21)(q42or43; q11)			
1813	3	B	66	19	F	W	46,XX,t(1;21)(q12; q22)pat/46,XX,t(1;21) (q12;q22)pat,t(X;15) (p11;q13)	U	A	Menstrual dysfunction; Adj-2 segregation

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #18

GM #	Passage #	Culture Media	Submitter	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Veri- fied	Remarks
<u>TRANSLOCATIONS</u>										
<u>Chromosome 1, continued</u>										
1881	7	A	140	3	M	W	46,XY,t(1;21)(lqter▶ 1p32::21q22▶21qter; 21pter▶21q22::1p32▶ 1pter)	U	A	Suspected Sanfilippo
<u>Chromosome 2</u>										
257*	3	A	2	26	F		46,XX,t(1;2)(q32;p23)		A	t(1;2); see Chromosome 1
1229	3	B	7	3	F	B	46,XX,-1,+der(1), t(1;2)(p36;q31)pat	U	A	Proband; t(1;2); see Chromosome 1
1230	2	B	7	44	M	B	46,XY,t(1;2)(p36;q31)	B	A	Father; t(1;2); see Chromosome 1
501*	3	A	14	4 1/2	M	W	46,XY,der(2),t(2;4) (2qter▶2p25::4q21▶ 4qter)pat	U	A	Proband
1064*	7	A	14	39	M	W	46,XY,t(2;4)(2qter▶ 2p25::4q21▶4qter; 4pter▶4q21::2p25▶ 2pter)	B	A	Father
327*	4	A	141	24	M	W	46,XY,t(2;8)(2pter▶ 2q13::8q24▶8qter; 8pter▶8q24::2q13▶ 2qter)	B	A	

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 18; 38; 39; 44

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Veri- fied	Remarks
TRANSLOCATIONS										
Chromosome 2, continued										
845	7	C	18	18	F	W	46,XX,t(2;8)(2pter▶2q37::8q13▶8qter; 8pter▶8q13::2q37▶2qter)mat	B	A	Proband; primary Amenorrhea
846	8	C	18	54	F	W	46,XX,t(2;8)(2pter▶2q37::8q13▶8qter; 8pter▶8q13::2q37▶2qter)	B	A	Mother; multiple spontaneous abortions
1225	10	B	77	3 mo.	F	I	46,XX,t(2;20)(2qter▶2p21::20p13▶20pter; 20qter▶20p13::2p21▶2pter)		A	Indian (India); de novo; has multiple congenital anomalies
692	5	A	1	8	M	W	45,XY,-21,t(2;21)(q3;q22)mat	U	B	Proband; mentally retarded
693	5	A	1	27	F	W	46,XX,t(2;21)(q3;q22)	B	B	Mother
1579	8	A	30	35	F	W	46,XX,t(2;13)(p1;q34)		B	
1848	3	B	179	35	M	W	46,XY,der(10),t(2;10)(p24;q26)mat	U	B	Cousin
1849	4	B	179	26	M	W	46,XY,der(10),t(2;10)(2pter▶2p24::10q26▶10pter)mat	U	A	Possible red cell acid phosphatase variant
1683	3	B	179	28	F	W	46,XX,t(2;10)(p24;q26)	B	A	Proband; cousin

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Veri-fied	Remarks
<u>TRANSLOCATIONS</u>										
<u>Chromosome 3</u>										
1550	6	A	140	31	M	W	46,XY,t(1;3;4)(q32;p21q29;p14)	B	A	t(1;3;4); see Chromosome 1
194*	15	A	30	19	F	W	46,XX,t(X;3)(Xpter>Xq2::3q1>3qter;3pter>3q1::Xq2>Xqter)	B	A	Possibly reciprocal; 3q12 or 3q13 break; break pt. on X distal to Xq25
<u>Chromosome 4</u>										
1550	6	A	140	31	M	W	46,XY,t(1;3;4)(q32;p21q29;p14)	B	A	t(1;3;4); see Chromosome 1
501*	3	A	14	4 1/2	M	W	46,XY,der(2),t(2;4)(p25;q21)pat	U	A	t(2;4); see Chromosome 2; Proband
1064*	7	A	14	39	M	W	46,XY,t(2;4)(p25;q21)	B	A	t(2;4); see Chromosome 2; Father
1087	3	A	66	2 mo.	F		46,XX,t(4;5)(p;q)			
773*	5	C	66	26	F		46,XX,t(4;7)(4qter>4p16::7q34>7qter;7pter>7q34::4p16>4pter)	B	A	Mother
1220*	3	A	66	2	F		46,XX,der(4),t(4;7)(4qter>4p16::7q34>7qter)mat	U	A	Proband

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 29; 38; 39; 47; 49

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Verified	Remarks
Chromosome 4, continued										
157*	11	A	30	28	M	W	49,XXXY,t(4;11)(4pter>4q35::11q23>11qter;11pter>11q23::4q35>4qter)pat	B	A	Severe mental retardation, sexual infantilism
380*	2	A	78	35	F		46,XX,rcp(4;11)(4pter>4q25::11q13>11qter;11pter>11q13::4q25>4qter)	B	A	
1101	7	C	7	32	M	W	46,XY,t(4;12)(p1;p11)	B		
972	3	C	67	12 1/2	F	B	46,XX,t(4;13)(4pter>4q31::13q14>13qter;13pter>13q14::4q31>4qter)	B	A	
624*	2	A	77	38	F	W	46,XX,t(4;15)(4pter>4q11::15p11>15pter;15qter>15p11::4q11>4qter)	B	A	
98*	5	A	119	10 mo.	M		46,XY,der(21),t(4;21)(p11;p12)mat	U	A	
1091	3	A	58	27	F	W	46,XX,t(4;10)(q32;q22)	B	B	

TRANSLOCATIONS

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 23; 16; 22; 17

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Veri- fied	Remarks
<u>TRANSLOCATIONS</u>										
Chromosome 5										
860	11	B	36	13	F	W	46,XX,der(5),ins(5;1)(q15;q32q25)pat	U	A	t(1;5); grows slowly; see Chromosome 1; Proband
861	7	B	36	40	M	W	46,XY,ins(5;1)(q15;q32q25)	B	A	t(1;5); see Chromosome 1 Father
1087	3	A	66	2 mo.	F		46,XX,t(4;5)(p;q)			t(4;5); see Chromosome 4
1221	6	C	21	1 1/2	M	B	46,XY,-5,+ins(5;6)(q33;q15q27)mat	U	B	Partial trisomy of 6q15q27; Proband
1222	5	C	21	23	F	B	46,XX,ins(5;6)(q33;q15q27)mat	B	B	Mother
1524	3	C	21	19	M	B	46,XY,-5,+ins(5;6)(q33;q15q27)		B	Cousin
71*	15	A	15	19	F	W	46,XX,-5,der(5),t(5;?)(p1;?)mat		A	See Deletions Cri du Chat
589*	2	A	12	17 1/2	M	W	46,XY,t(5;14)(5qter▶5p14::14q21▶14qter;14pter▶14q21::5p14▶5pter)		A	

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 36; 41

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Veri-fied	Remarks
TRANSLOCATIONS										
Chromosome 5, continued										
344*	4	A	66	6 1/2	M	W	45,XY,del(5)(pter ▶p14:),t(5;15)(5qter ▶5p14:l5q1▶15qter), t(9;11)(9qter▶9p24:; 11q23▶11qter;11pter▶ 11q23::9p24▶9pter)	U	A	Cri du Chat
1535	6	A	2	16	F	B	46,XX,der(12),t(5;12) (q31;q24)mat,9qh+,21s+	U		Proband
1536	6	C	2	38	F	B	46,XX,t(5;12)(q31;q24), 9qh+,21s+	B		Mother
1678	3	B	140	33	F	F	46,XX,t(5;10)(p15;p13)	B	B	
Chromosome 6										
1421	12	A	109	53	M	B	46,XV,t(1;6)(q43;q21)	B	A	t(1;6); see Chromosome 1
1137	6	D	17	1 mo.	M	B	48,XXY,+21,recp(6;10) (6qter▶6p22 or 24:: 10p12▶10pter;10qter▶ 10p12::6p22 or 24▶6pter)		B	
1524	3	C	21	19	M	B	46,XY,-5,+ins(5;6) (q33;q15q27)mat	U	B	t(5;6); Cousin; see Chromosome 5
1221	6	C	21	1 1/2	M	B	46,XY,-5,+ins(5;6) (q33;q15q27)mat	U	B	t(5;6); Proband; see Chromosome 5
1222	5	C	21	23	F	B	46,XX,ins(5;6)(q33; q15q27)mat	B	B	t(5;6); Mother see Chromosome 5
*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #27										

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Verified	Remarks
<u>TRANSLOCATIONS</u>										
Chromosome 6, continued										
144*	5	A	12	34	F	W	46,XX,der(6),t(6;21)(21qter>21q11::6p25>6qter)mat	U	A	13% of cells found to be balanced in passage 1 after recovery; these tend to increase with passage in culture
610*	7	A	75	3 mo.	F	W	46,XX,t(6;18)(6qter>6q21::18p11>18pter; 6qter>6q21::18p11>18qter)	B	A	
1605	3	A	166	34	M	W	46,XY,t(6;11)(6qter>6p2::11q23>11qter; 11pter>11q23::6p2>6pter)	B	B	
2068	5	J	88	4	F	W	46,XX,t(6;7)(6qter>6q27::7q22>7qter; 7pter>7q22::6q27>6qter)	B	A	
1139	4	A	95	21	F	W	46,XX,t(15;17)(q15;p13),t(6;13)(p21;q34)		A	Culture is mosaic 64% carry only the t(15;17) 36% carry both
Chromosome 7										
1356	2	A	66	26	F	W	46,XX,t(1;7)(p34;p13)	B	A	t(1;7); see Chromosome 1

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 6; 28

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Veri- fied	Remarks
<u>TRANSLOCATIONS</u>										
Chromosome 7, continued										
773	5	C	66	26	F		46,XX,t(4;7)(p16;q34)	B	A	t(4;7); see Chromosome 4; Mother
1220	3	A	66	2	F		46,XX,der(4),t(4;7)(p16;q34)mat	U	A	t(4;7); see Chromosome 4; Proband
44*	5	A	97	6	M	W	46,XY,t(7;10)(7qter>7p2::10q11>10qter; 10pter>10q11::7p2>7pter)	B	A	Congenital malformations, severe retardation although apparently balanced
1696	9	C	176	30	F	W	46,X,t(X;7)(Xqter>Xq21::7p22>7qter; Xpter>Xq21::7p22>7pter)	B	B	
2068	5	J	88	4	F	W	46,XX,t(6;7)(q27;q22)	B	A	t(6;7); see Chromosome 6
657	3	A	107	4 wk.	M	W	46,XY,t(7;18)(7pter>7q36::18q21>18qter; 18pter>18q21::7q36>7qter)	U	A	
Chromosome 8										
845	7	C	18	18	F	W	46,XX,t(2;8)(q37;q13)mat	B	A	t(2;8); Proband; see Chromosome 2
846	8	C	18	54	F	W	46,XX,t(2;8)(q37;q13)	B	A	t(2;8); Mother; see Chromosome 2
327	4	A	141	24	M	W	46,XY,t(2;8)(q13;q24)	B	A	t(2;8); see Chromosome 2

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #12

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Veri- fied	Remarks
<u>TRANSLOCATIONS</u>										
<u>Chromosome 8, continued</u>										
1512	6	A	30	26	M	W	46,XY,t(8;10)(p21;p15)	B	B	
213*	5	A	82	35	M	W	46,XY,t(8;12)(8pter▶ 8p23::12p11▶12qter; 12pter▶12p11::8p23▶ 8qter)	B	A	
<u>Chromosome 9</u>										
1387	4	A	61	14	F	W	46,XX,-13,+der(9), t(9;13)(q22;q12)mat	U	B	Result of adj-2 seg.
988*	10	G	82	3 wk.	M	W	46,XY,t(9;17)(9qter▶ 9p13::17q25▶17qter; 17pter▶17q25::9p13▶ 9pter)	B	A	Multiple congenital anomalies; 13% of cells are polyploid
705	13	B	16	10 mo.	F	W	46,X,t(X;9)(q12;p24)	B	B	Grows slowly
1414	5	C	68	28	F	B	46,X,-X,+der(9),rcp (X;9)(q11;q32)mat	U	B	Proband; Turner's Syndrome
1429	8	A	68	55	F	B	47,X,+10,rcp(X;9) (q13;q34)	B	A	Mother; mosaic, 14% do not show +10
1664	6	C	113	40	M	W	46,XY,t(9;13)(q13;q12)	B	B	See Chromosome 13
1734	3	B	140	40	M		46,XY,der(15),t(9;15) (p11;q11)mat	U	B	

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 8; 42

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Verified	Remarks
<u>TRANSLOCATIONS</u>										
<u>Chromosome 9, continued</u>										
1750	2	B	70	2 1/2	M	W	47,XY,+der(14)t(9;14)(p24;q22)mat	U	B	Proband
1751	2	B	70	26	F	W	46,XX,t(9;14)(p24;q22)	B	B	Mother
1892	5	B	132	37	F	W	46,XX,rcp(9;18)(p24;q12)	B	B	see Chromosome 14
<u>Chromosome 10</u>										
1137	6	D	17	1 mo.	M	B	48,XXY,+21,rcp(6;10)(p22 or 24;p12)		B	t(6;10); see Chromosome 6
44*	5	A	97	6	M	W	46,XY,t(7;10)(p2;q11)	B	A	t(7;10); see Chromosome 7
1512	6	A	30	26	M	W	46,XY,t(8;10)(p21;p15)	B	B	t(8;10); see Chromosome 8
1396	3	A	95	3	M	W	46,XY,der(10),t(10;16)(q26;q22)pat		A	
216*	4	A	143	29	M	W	46,XY,t(10;17)(10pter▶10q24::17p13▶17pter;17qter▶17p13::10q24▶10qter)	B	A	Father
217*	6	A	143	9 da.	M	W	46,XY,-17,+der(17),t(10;17)(17qter▶17p13::10q24▶10qter)pat	U	A	Proband, deceased; culture grows slowly

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 12; 13; 14

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Veri- fied	Remarks
TRANSLOCATIONS										
Chromosome 10, continued										
959*	6	C	143	11	M	W	same karyotype as GM-217, pat	U	A	Paternal fourth cousin of GM-217
1413	8	B	132	1 1/2	F	B	46,XX,der(10),rcp(10;21)(q26;q21)mat	U	B	Proband
1580	6	B	132	48	F	B	46,XX,rcp(10;21)(q26;q21)	B	B	Grandmother of proband; aunt of GM-1399
1399	5	B	132	20	M	B	47,XY,+der(21),rcp(10;21)(q26;q21)mat	U	B	Nephew of GM-1580; Cousin of GM-1413
984	4	C	95	34	F	W	46,XX,t(10;21)(q24;q22)	B	Mother Proband	
983	3	C	95	14 mo.	M	W	46,XY,der(21),t(10;21)(q24;q22)mat	U		
1678	3	B	140	33	F		46,XX,t(5;10)(p15;p13)		See Chromosome 5	
1848	3	B	179	35	M	W	46,XY,der(10),t(2;10)(p24;q26)mat	U	B	See Chromosome 2 Cousin
1849	4	B	179	26	M	W	46,XY,der(10),t(2;10)(p24;q26)mat	U	A	See Chromosome 2 Cousin
1683	3	B	179	28	F	W	46,XX,t(2;10)(p24;q26)	B	A	See Chromosome 2 proband

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #37

G#	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Veri- fied	Remarks
<u>TRANSLOCATIONS</u>										
<u>Chromosome 10, continued</u>										
1091	<u>3</u>	A	58	27	F	W	46,XX,t(4;10)(q32;q22)	B	B	
157*	<u>11</u>	A	30	28	M	W	49,XXXXY,t(4;11)(q35;q23)pat	B	A	t(4;11); see Chromosome 4
380*	2	A	78	35	F		46,XX,rcp(4;11)(q25;q13)	B	A	t(4;11); see Chromosome 4
980	2	A	12	33	F	W	45,XX,-22,t(11;22)(11pter>11q25::22q11>22qter)	U	B	
1605	3	A	166	34	M	W	46,XY,t(6;11)(6qter>6p2::11q23>11qter;11pter>11q23::6p2>6pter)	B	B	t(6;11); see Chromosome 6
<u>Chromosome 12</u>										
1101	<u>7</u>	C	7	32	M	W	46,XY,t(4;12)(p1;p11)	B		t(4;12); see Chromosome 4
1535	6	A	2	16	F	B	46,XX,der(12),t(5;12)mat, 9qh+,21s+	U	B	Proband; t(5;12); see Chromosome 5
1536	6	C	2	38	F	B	46,XX,t(5;12), 9qh+,21s+	B	B	Mother; t(5;12); see Chromosome 5
213*	5	A	82	35	M	W	46,XY,t(8;12)(p23;p11)	B	A	t(8;12); see Chromosome 8
1665	3	B	140	28	F	W	46,XX,t(12;21)(12p21q)	B	B	

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 23; 16; 8

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Veri-fied	Remarks
<u>TRANSLOCATIONS</u>										
<u>Chromosome 13</u>										
1579	8	A	30	35	F	W	46,XX,t(2;13)(p1;q34)		B	t(2;13); see Chromosome 2
972	3	C	67	12 1/2	F	B	46,XX,t(4;13)(q31;q14)	B	B	t(4;13); see Chromosome 4
1387	4	A	61	14	F	W	46,XX,-13,*der(9), t(9;13)(q22;q12)mat	U	B	t(9;13); see Chromosome 9
1555	5	C	113	13	M	W	47,XY,+der(13),t(13;17)(q14;p13)mat	U	B	Sib
1663	13	A	113	14	M	W	46,XY,+der(17),t(13;17)(q14;p13)mat	U	B	Proband; partial trisomy 13
85	5	A	143	2 wk.	M	B	46,XY,-13,*t(13q13q)	U	A	Clinical Trisomy 13
1296	4	C	12	47	F	W	45,XX,t(13q15q)	B		
1224	7	B	77	3 mo.	F	W	46,XX,der(13),t(13;18)(q32;q11)pat	U	A	
392*	9	A	2	1	F	W	45,XX,t(13;22)(13qter►cen►22qter)	B	A	
627*	3	A	58	30	F	B	46,XX,t(13;22)(13pter►13q22::22q13►22qter; 22pter►22q13::13q22►13qter)	B	A	
1664	6	C	113	40	M	W	46,XY,t(9;13)(q13;q12)	B	B	See Chromosome 9

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 19; 30

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Veri-fied	Remarks
<u>TRANSLOCATIONS</u>										
<u>Chromosome 13, continued</u>										
2018	12	B	77	2	F	W	45,XX,tan(13;13)(pter ▶q34::q12▶qter)	U	A	
1139	4	A	95	21	F	W	46,XX,t(15;17)/46,XX, t(15;17),t(6;13)		A	t(15;17);t(6;13) see Chromosome 6
<u>Chromosome 14</u>										
589*	2	A	12	17	1/2	M	46,XY,t(5;14)(p14;q21)		A	t(5;14); see Chromosome 5
479*	12	C	48	35	F	W	45,XX,t(14;15)(14qter▶ cen▶15qter)		A	
981	3	C	148	4	F	B	46,XX,der(14),t(14; 20)(p11;p11),inv(9) (pter▶p13::q13▶p13:: q13▶qter)	U	A	Proband
982	3	C	148	Adult	F	B	46,XX,t(14;20)(p11;p11)	B	A	Mother
5*	5	A	111	30	F	W	45,XX,t(14;22)(14qter▶ cen▶22qter)		A	
73*	5	C	111	Adult	F	W	46,X,t(X;14)(Xpter▶ Xq13::14qter;14pter▶ 14q32::Xq13▶Xqter)	B	A	Mother
74*	10	S	111	23	M	W	47,Y,der(X),der(14), +der(14),t(X;14)(q13; q32)mat	U	A	Proband; Klinefelter Syndrome

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 41; 33; 3; 1; 2

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Veri- fied	Remarks
TRANSLOCATIONS										
Chromosome 14, continued										
1750	2	B	70	2 1/2	M	W	47,XY,+der(14),t(9;14)(p24;q22)mat	U	B	Proband
1751	2	B	174	26	F	W	46,XX,t(9;14)(p24;q22)	B	B	Mother; t(9;14); see Chromosome 9
2044	2	B	164	6 1/2	M		45,XY,-16,t(14;16)(16;18)(14qter▶14p12::16p11▶16pter;18qter▶18p1::16q12▶16qter)	U	A	
Chromosome 15										
126*	4	A	56	33	M	W	46,XY,t(1;15)(p36;q1)	B	A	t(1;15); see Chromosome 1
624*	2	A	77	38	F	W	46,XX,t(4;15)(q11;p11)	B	A	t(4;15); see Chromosome 4
344*	4	A	66	6 1/2	M	W	45,XY,del(5),t(5;15)(p14;q1),t(9;11)(p24;q23)	U	A	t(5;15)+t(9;11); see Chromosome 5
1296	4	C	12	47	F	W	45,XX,t(13q15q)	B		t(13;15); see Chromosome 13
479	12	C	48	35	F	W	45,XX,t(14q15q)		A	t(14;15); see Chromosome 14
11139	4	A	95	21	F	W	46,XX,t(15;17)(q22;p13)/46,XX,t(15;17),t(6;13)	B	A	t(15;17)t(6;13); see Chromosome 6
17*	4	A	12	2 1/2	M	W	45,XY,t(15;18)(15qter▶15q1::18q23▶18pter)	U	A	

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 11; 22; 27; 4

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Verified	Remarks
<u>TRANSLOCATIONS</u>										
<u>Chromosome 15, continued</u>										
118*	5	A	52	29	F	W	46,XX,-15,+der(Y), t(Y;15)(qter▶Yq11:: 15p1▶15qter)pat	U	A	Uterine tissue
1734	3	B	140		M		46,XY,der(15),t(9;15) (p11;q11)mat	U	B	t(9;15); see Chromosome 9
1813	3	B	66	19	F	W	46,XX,t(1;21)(q12;q22) pat/46,XX,t(1;21)(q12; q22)pat,t(X;15)(p11;q13)	U	A	See Chromosome 1
Chromosome 16	1396	A	95	3	M	W	46,XX,der(10),t(10;16) (q26;q22)pat		A	t(10;16); see Chromosome 10
2044	2	B	164	6 1/2	M		45,XY,-16,t(14;16) (16;18)(p12;p11q12;p1)	U	A	t(14;16)t(16;18); see Chromosome 14
Chromosome 17	201*	A	30	13	F	W	47,XX,+21,t(1;17) (p32;p13)	B	A	t(1;17); see Chromosome 1

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 31; 10

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Veri- fied	Remarks
<u>TRANSLOCATIONS</u>										
Chromosome 17, continued										
216*	4	A	143	29	M	W	46,XY,t(10;17)(q24;p13)	B	A	t(10;17); Father see Chromosome 10
217*	6	A	143	9 da.	M	W	46,XY,-17,+der(17), t(10;17)(q24;p13)pat	U	A	t(10;17); Proband see Chromosome 10
959	6	C	143	11	M	W	46,XY,-17,+der(17), t(10;17)(q24;p13)pat	U	A	t(10;17); pat 4th cousin; see Chromosome 10
1555	5	C	113	13	M	W	47,XY,+der(13),t(13;17)(q14;p13)mat	U	B	t(13;17); Sib; see Chromosome 13
1663	13	A	113	14	M	W	46,XY,-17,+der(17), t(13;17)(q14;p13)mat	U	B	t(13;17); Proband; see Chromosome 13
1139	4	A	95	21	F	W	46,XX,t(15;17)(q22;p13)/46,XX,t(15;17), t(6;13)	B	A	t(15;17); t(6;13) see Chromosome 6
271*	3	A	58	28	F	W	46,XX,t(17;19)(17pter▶17q23::19p13▶19pter; 19qter▶19p13::17q23▶17qter)	B	A	

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 13; 14; 37; 15

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Veri- fied	Remarks
<u>TRANSLOCATIONS</u>										
<u>Chromosome 17, continued</u>										
119*	4	A	87	31	F	W	46,XX,t(17;22)(17qter▶ 17p13::22q11▶22qter; 22pter▶22q11::17p13▶ 17pter)	B	A	
988*	10	G	82	3 wk.	M	W	46,XY,t(9;17)(p13;q25)	B	A	t(9;17); see Chromosome 9
<u>Chromosome 18</u>										
610*	7	A	75	3 mo.	F	W	46,XX,t(6;18)(q21;p11)	B	A	t(6;18); see Chromosome 6
1224	7	B	77	3 mo.	F	W	46,XX,der(13),t(13;18) (q32;q11)pat	U	A	t(13;18); see Chromosome 13
17*	4	A	12	2 1/2	M	W	45,XY,t(15;18)(q1;q23)	U	A	t(15;18); see Chromosome 15
657	3	A	107	4 wk.	M	W	46,XY,t(7;18)(q36;q21)	U	A	t(7;18); see Chromosome 7
1892	5	B	132	37	F	W	46,XX,rcp(9;18)(p24; q12)	B	B	t(9;18) see Chromosome 9
2044	2	B	164	6 1/2	M		45,XY,-16,t(14;16) (16;18)(p12;p11q12;p1)	U	A	t(14;16)t(16;18); see Chromosome 14
Chromosome 19										
271*	3	A	58	28	F	W	46,XX,t(17;19)(q23;p13)	B	A	t(17;19); see Chromosome 17

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 9; 42; 28; 4; 15

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Verified	Remarks
<u>TRANSLOCATIONS</u>									
<u>Chromosome 19, continued</u>									
89	5	A	43	Adult	F	W	46,X,t(X;19)(Xpter▶Xq2::19ql3or19pl3▶19qter or 19pter?; 19pter or 19qter▶19pl3or19ql3::Xq2▶Xqter)	B	A
1225	10	B	77	3 mo.	F	I	46,XX,t(2;20)(p21;p13)	A	t(2;20); see Chromosome 2
981	3	C	148	4	F	B	46,XX,der(14),t(14;20)(p11;p11)mat,inv(9)(p13ql3)	U	A
982	3	C	148	Adult	F	B	46,XX,t(14;20)(p11;p11)	B	A
<u>Chromosome 21</u>									
1564	9	C	151	28	F		45,XX,t(1;21)(q42or43;q11)		t(1;21); see Chromosome 1
692	5	A	1	8	M	W	45,XY,-21,t(2;21)(q3;q22)mat	U	B
693	5	A	1	27	F	W	46,XX,t(2;21)(q3;q22)	B	B
98*	5	A	119	10 mo.	M		46,XY,der(21),t(4;21)(p11;p12)mat	U	A
144*	5	A	12	34	F	W	46,XX,der(6),t(6;21)(p25;q11)mat	U	A

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 17; 6

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Veri- fied	Remarks
<u>TRANSLOCATIONS</u>										
<u>Chromosome 21, continued</u>										
1413	8	B	132	1 1/2	F	B	46,XX,der(10),rcp(10; 21)(q26;q21)mat	U	B	t(10;21); Proband; see Chromosome 10
1580	6	B	132	38	F	B	46,XX,rcp(10;21)(q26; q21)	B	B	t(10;21); Grandmother; see Chromosome 10
1399	5	B	132	20	M	B	47,XY,+der(21),rcp(10; 21)(q26;q21)mat	U	B	t(10;21); Cousin; see Chromosome 10
1581	7	B	132	25	F	B	46,X,t(X;21)(q11;p11?)		B	Mother
1730	11	C	132	5	F	B	46,XX,der(21),t(X;21) (q11;p11?)mat		B	Proband
1665	3	B	140	28	F	W	46,XX,t(12;21)(12p21q)	B	B	t(12;21); see Chromosome 12
1700	3	B	95	6	M		46,XY,t(21;22)(p12;q11)	B	B	
1881	7	A	140	3	M		46,XY,t(1;21)(p32;q22)		A	t(1;21); suspected Sanfilippo; see Chromosome 1
2058	5	B	165	11 da.	F	W	46,XX,/46,XX,t(21;21)	U	B	87%/13% at passage 8; see Trisomy 21, Down's syndrome.
1813	3	B	66	19	F	W	46,XX,t(1;21)(q12;q22) pat/46,XX,t(1;21)(q12; q22)pat,t(X;15)(p11;q13)	U	A	See Chromosome 1

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Veri-fied	Remarks
<u>TRANSLOCATIONS</u>										
<u>Chromosome 22</u>										
980	2	A	12	33	F	W	45, XX, -22, t(11;22)(q25;q11)	U	B	t(11;22); see Chromosome 11
392*	9	A	2	1	F	W	45, XX, t(13;22)(13q22q)	B	A	t(13;22); see Chromosome 13
627*	3	A	58	30	F	B	46, XX, t(13;22)(q22;q13)	B	A	t(13;22); see Chromosome 13
5*	5	A	111	30	F	W	45, XX, t(14;22)(14q22q)		A	t(14;22); see Chromosome 14
119*	4	A	87	31	F	W	46, XX, t(17;22)(p13;q11)	B	A	t(17;22); see Chromosome 17
1700	3	B	95	6	M		46, XY, t(21;22)(p12;q11)	B	B	t(21;22); see Chromosome 21
<u>Chromosome X</u>										
97*	3	A	107	1	F	W	46, X, t(X;1)(q26;q12)	B	A	t(X;1); see Chromosome 1
194*	15	A	30	19	F	W	46, XX, t(X;3)(q2;q1)	B	A	t(X;3); see Chromosome 3
705	13	B	16	10 mo.	F	W	46, X, t(X;9)(q12;p24)	B	B	t(X;9); see Chromosome 9
1414	5	C	68	28	F	B	46, X, -X, +der(9), rcp (X;9)(q11;q32)mat	U	B	t(X;9); Proband; see Chromosome 9
1429	8	A	68	55	F	B	47, X, +10, rcp(X;9)(q13;q34)	B	A	t(X;9); Mosaic Mother; see Chromosome 9

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 19; 30; 3; 9; 7; 29

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Veri- fied	Remarks
<u>TRANSLOCATIONS</u>										
<u>Chromosome X, continued</u>										
73*	5	C	111	Adult	F	W	46,X,t(X;14)(q13;q32)	B	A	t(X;14); Mother; see Chromosome 14
74*	10	S	111	23	M	W	47,Y,+der(14),t(X;14)(q13;q32)mat	U	A	t(X;14); Proband; see Chromosome 14
89	5	A	43	Adult	F	W	46,X,t(X;19)(q2;q13or p13)	B	A	t(X;19); see Chromosome 19
1581	4	IMR	132	25	F	B	46,X,t(X;21)(q11;p11?)		B	t(X;21); Mother; see Chromosome 21
1730	11	C	132	5	F	B	46,XX,der(21),t(X;21)(q11;p11?)mat		B	t(X;21); Proband; see Chromosome 21
1696	9	C	176	30	F	W	46,X,t(X;7)(q21;p22)		B	t(X;7); see Chromosome 7
2103	6	C	109	19	F	W	46,X,-Y,t(X;Y)(Yqter>Yq11::Xp11>Xq22)	U	B	2° Amenorrhea; see Chromosome Y
1813	3	B	66	19	F	W	46,XX,t(1;21)(q12;q22)pat/46,XX,t(1;21)(q12;q22)pat,t(X;15)(p11;q13)	U	A	See Chromosome 1
<u>Chromosome Y</u>										
118*	5	A	52	29	F	W	46,XX,-15,+der(Y),t(Y;15)(q11;p1)	U	A	t(Y;15); see Chromosome 15
2103	6	C	109	19	F	W	46,X,-Y,t(X;Y)(q11;p11)	U	B	t(X;Y); see Chromosome X

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 1; 2; 31

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Veri-fied	Remarks
<u>INVERSIONS</u>										
<u>Chromosome 2</u>										
735	17	A	77	17	F	W	46,Xdic,Xqi,inv(2)(p15q21)mat	B	B	Turner Syndrome
<u>Chromosome 3</u>										
1252	3	C	106	22	F	B	46,XX,inv(3)(p25q25),inv(9)(p11q13)	B	A	Mother; see Chromosome 9
1253	3	C	106	4 mo.	M	B	46,XY,rec(3),dup q,inv(3)(p25q25)mat	U	A	Proband; see Chromosome 9; Father is XYY (GM-1250)
<u>Chromosome 4</u>										
413	4	B	56	1	M		46,XY,inv(4)(pter>p14::q22>p14::q22>pter)mat	B	B	Proband; has minor malformations
414	4	B	56	23	F		46,XX,inv(4)(pter>p14::q22>p14::q22>pter)mat	B	B	Mother
<u>Chromosome 9</u>										
445	7	A	30	26	M	W	46,XY,inv(9)(p13q13)		B	Father; clinically normal
447	7	A	30	1	M	W	46,XY,inv(9)(p13q13)		B	Proband; congenital malformations
450	7	A	30	33	M	W	46,XY,inv(9)(p13q13)	B	B	Same patient; clinically normal
451	7	A	30	33	M	W	46,XY,inv(9)(p13q13)	B	B	Same patient; testicular tissue

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Veri-fied	Remarks
<u>INVERSIONS</u>										
<u>Chromosome 9, continued</u>										
453	7	A	30	24	F	W	46,XX,inv(9)(p13q13)	B	B	Clinically normal
1251	2	C	106	4	M	B	46,XY,inv(9)(p11q13)mat	B	A	Half brother of proband
1252	3	C	106	22	F	B	46,XX,inv(3)(p25q25),inv(9)(p11q13)	B	A	Mother of proband & sib; see Inversion Chromosome 3
1253	3	C	106	4 mo.	M	B	46,XY,rec(3),dup q,inv(3)(p25q25)mat	U	A	Proband; father is XYY (GM-1250); see Inversion Chromosome 3
1918	6	B	61	28	M	B	47,XYqs,+21,inv(9)(p13q21)			See Trisomy, Chromosome 21
1920	2	B	61	23	M	B	47,XY,+21,inv(9)(p13q13)mat			See Trisomy, Chromosome 21
981	3	C	148	4	F	B	46,XX,der(14)t(14;20)(p11;p11)inv(9)(p13q13)	U	A	See t(14;20)
<u>Chromosome 10</u>										
446	7	A	30	26	M	W	46,XY,inv(10)(p11q11)	B	B	Clinically normal
448	7	A	30	1	M	W	46,XY,inv(10)(p11q21)pat	B	B	Proband; congenital malformations;
452	7	A	30	22	M	W	46,XY,inv(10)(p11q21)	B	B	Clinically normal; father
<u>Chromosome 13</u>										
437	3	A	66	32	F	W	46,XX,inv(13)(p13q21)	B	B	

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Verified	Remarks
<u>INVERSIONS</u>									
Chromosome 13, continued									
1570	2	A	119	5 mo.F	M		46,XY,rec(13)dup p,inv (13)(p12q32)		Paternal karyotype is 46,XY,inv(13) (p12q32)

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Verified	Remarks
<u>MONOSOMIES</u>									
<u>Chromosome 21</u>									
137	4	A	75	5 1/2	M	W	45,XY,-21	A	
230	9	J	88	14 mo.	F	W	45,XX,-21	A	
<u>Chromosome X</u>									
775	4	C	107	13	F		45,X	A	
993	2	A	107	9	F	B	45,X	A	
1176	2	A	107	8	F	W	45,X	B	
857	#	A	95	1 da.	F	W	45,X	B	
563	4	A	66	14	F	B	45,X	B	Same patient; only X0 cells found; tissue from right gonad Same patient; 4 cells, 46XX/46 cells, 45X0; tissue from left gonad See iso-X
562	4	A	66	14	F	B	45,X/46,XX	B	
339	12	A	30	25	F	W	45,X/46,X,i(Xq)		
314	16	A	30	60	F	W	45,X/46,X,dup(X)(qter ▶pter::pter▶qter)	B	Ovarian dysgenesis without Turner stigmata
1441	2	A	74	3 1/2 mo.	M	W	45,X/46,X,iso(Y)		See iso-Y; tissue from right testis
978	7	C	143	15	F	W	46,XY/45,X		Karyotype based on leukocytes, tissue from right gonad; see Gonadal Dysgenesis
1723	2	B	203	23	F	W	45,X	A	

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Verified	Remarks
<u>TRISOMY/POLYSOMY</u>									
<u>Chromosome 8</u> 496	<u>3</u>	A	56	2	M		47,XY,+8/46,XY		2 cell lines in lymphocytes; 83% have +8; in skin fibroblasts 81% have +8
2030	7	A	30	23	M	W	46,XY/47,XY,+8	B	5%/95% in fibroblasts; mental retardation
<u>Chromosome 9</u> 2329	<u>6</u>	J	39	18 da.	F	W	47,XX,+9/46,XX	B	10% of fibroblasts show the +9
<u>Chromosome 10</u> 1429	<u>8</u>	A	68	55	F	B	47,X,+10,rcp(X;9)(q13;q34)	A	See Chromosome 9, Translocation
<u>Chromosome 13</u> 503	<u>2</u>	A	66	1	M		46,XY/47,XY,+13	B	70%/30%
<u>Chromosome 18</u> 143	<u>3</u>	A	107	3 mo.	F	B	47,XX,+18	A	Autopsy specimen
734	3	A	95	F	F		47,XX,+18	A	
1359	3	A	107	1 mo.	M	W	47,XY,+18		
<u>Chromosome 21</u> 2504	<u>7</u>	A	95	1 mo.	M	B	47,XY,+21	A	Formerly GM-258
201*	10	A	30	13	F	W	47,XX,+21,t(1;17)(p32;p13)	A	See Translocation, Chromosome 1
144*	5	A	12	34	F	W	46,XX,der(6)t(6;21)(p25;q11)mat	A	See Translocation, Chromosome 6 and 21; Down's Syndrome

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 10; 6

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Verified	Remarks
<u>TRISOMY/POLYSOMY</u>									
<u>Chromosome 21, continued</u>									
260	10	A	95	11	M	W	46,XY/47,XY,+21		Mosaic; 25% +21
1918	6	B	61	28	M	B	47,XY,+21,inv(9)(p13q21)	B	See Inversion Chromosome 9
1920	2	B	61	23	M	W	47,XY,+21,inv(9)(p13q13)mat	B	See Inversion Chromosome 9
2067	4	H	95	5 mo.	F	M	47,XY,+21	A	
2058	5	B	165	11 da.	F	W	46,XX/46,XX,t(21;21)	B	70%/30% in leukocytes; see Translocation Chromosome 21
<u>Chromosome 22</u>									
2325	4	B	182	11 da.	F	W	47,XX,+22q-		Son of GM-2324, see Lymphoid, Translocation 16
84	5	A	95	1 mo.	M	W	47,XY,+22		
<u>Chromosome X</u>									
254	8	A	75	10 wk.	F	B	47,XXX	A	76% interphase nuclei have 2 sex chromosome bodies; 15% have one; no evidence for mosaicism
1415	3	A	61	27	F	W	48,XXXX	A	4% Polyploidy
326	3	A	125	6	M	W	49,XXXXY	A	Severe retardation; Hypogonadism
1534	10	C	2	26	F	W	49,XXXXX		

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Verified	Remarks
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TRISOMY/POLYSOMY

Chromosome X, continued

157*	11	A	30	28	M	W	49, XXXY, t(4;11) (q35;q23)pat	A	See Translocation, Chromosome 4
324	5	A	125	22	M	W	47, XXY	A	Klinefelter's; homo for G6PD deficient, med. type
325	4	A	125	30	M	W	47, XXY		Klinefelter's; hetero for G6PD def., med. type
Chromosome Y	3	C	106	23	M	B	47, XYY	A	Father of GM-1253 see Inversion Chromosome 9

TRIPLOIDY

1322	3	A	95	2 da.	F	B	69, XXX		Karyotype based on leukocytes; biopsy taken at autopsy
805	6	A	2	15 wk.F	M	I	69, XXY	B	Sib is 47, XY, +21; placental tissue
1672	8	J	88	1 da.	M	W	69, XXY	B	

DELETION/RING/ISOCHROMOSOME

Chromosome 1	6	A	30	2	M	W	46, XY, del(1)(q42)	B	Growth hormone deficiency and hypothyroidism
803	3	C	2	2	M	W	46, XY, del(1)(q24q25)	B	Karyotype based on lymphocytes
2025	4	B	126	2 1/2	M	W	46, XY, del(1)(pter>q2lor22::q25>qter)	A	

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #23

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Veri- fied	Remarks
<u>DELETION/RING/ISOCHROMOSOME</u>									
<u>Chromosome 2</u> 1138*	3	A	95	1	M	B	46,XY,del(2)(pter▶p23)	A	
945	2	C	77	1 1/2	F	O	46,XX,del(2)(qter▶p24:)	A	
<u>Chromosome 3</u> 366	3	A	3	4	M	W	46,XY,rec(3),dup q inv(3)(qter▶q21::p25▶qter)	A	
<u>Chromosome 4</u> 72	15	A	15	11	M	W	46,XY,del(4)(pter▶p14:)	A	
343	3	A	66	3	M		46,XY,del(4)(p)		Wolf-Hirschhorn Syndrome
2003	4	B	95	11	F	W	46,XX,4q-		
<u>Chromosome 5</u> 71*	15	A	15	19	F	W	46,XX,-5,der(5),t(5;?)(p1;?)mat	A	Deletion assumed to be derived rather than a simple deletion; see also Trans. Chromosome 5; ATCC CCL 90
<u>Chromosome 6</u> 109	7	A	134	2	F	W	46,XX,r(6)	B	36% of cells have r(6)
<u>Chromosome 8</u> 1549	8	A	140	7	M		46,XY,del(8)(p)	B	

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 43; 45

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Verified	Remarks
<u>DELETION/RING/ISOCHROMOSOME</u>									
Chromosome 9 870*	2	C	15	22	F	W	46,XX,del(9)(qter>pl3:)	A	
166*	4	A	107	6	M	W	46,XY,r(9)(p2q3)	A	44% of cells have r(9); 46% are 45,XY,-9
1667	7	C	2	13	F	W	46,XX,del(9)(p22)	B	
1893	4	B	140	3	M		46,XY,del(9)(pter>q13::q22>qter)	A	
2356	2	B	140	3	M		46,XY,del(9)(pter>p21:)	B	
Chromosome 11 2008	6	B	69	2 wk.	F	B	46,XX,del(11)(q23)	B	
Chromosome 13 509*	6	A	143	1 1/4	F	B	46,XX,del(13)(pter>q14:)	A	Short term lymphs show r(13); formerly GM-250*
285	3	A	107	4 da.	M	B	46,XY,r(13)	A	
729	8	A	142	14	M	W	46,XY,r(13)	A	
Chromosome 16 2346	2	B	204	3 mo.	M	W	46,XY,del(16)(q22)	B	
Chromosome 18 1118	8	C	150	2 1/2	F	W	46,XX,r(18)	B	

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 40; 21; 20

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Veri- fied	Remarks
<u>DELETION/RING/ISOCHROMOSOME</u>									
<u>Chromosome 18, continued</u>									
1727	13	A	142	45	M	W	46,XY,del(18)(p)		
<u>Chromosome X</u>									
735	17	A	77	17	F	W	46,Xqi,Xdic,inv(2)(p15 q21)	B	See Inversions, Chromosome 2
339	12	A	30	25	F	W	45,X/46,X,i(Xq)		
88*	4	A	12	19	F	W	46,X,i(X)(Xqter>cen>Xqter)	A	Buccal smear= 18% sex chromatin +; Turner Syndrome stigmata In peripheral blood leukocytes
1941	3	B	12	13	F	W	45,X/46,X,del(X)(q11)		
<u>Chromosome Y</u>									
1441	2	A	74	3 1/2	M	W	45,X/46,X,iso(Y)		
1709	4	C	109	11	F	W	46,X,i(Yq)	B	Gonadal dysgenesis
1304	4 IMR	C	56	27	F		<u>TERATOMA</u> 46,XX	B	PGM1=1;PGM3=1;PGD=AB; chromosome polymorphisms: 3=+/+;13=+/+;15=+/+;21=+/+; right ovarian teratoma; same patient
1305	4 IMR	C	56	27	F		46,XX	B	PGM1=1;PGM3=1;PGD=AB; chromosome polymorphisms: 3=+/+;13=+/+;15=+/+;21=+/+; uterine tissue same patient

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #5

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Veri- fied	Remarks
1306	8 IMR	C	56	31	F	W	TERATOMA 46,XX	B	PCM1=1;PCM3=2-1;PGD=A; chromosome polymorphisms: 13=-/-;14=+/-;21=-/-; right ovarian teratoma; same patient
1307	5 IMR	C	56	31	F	W	46,XX	B	PCM1=1;PCM3=2-1;PGD=A; chromosome polymorphisms; 13=+/-;14=+/-;21=+/-; fallopian tube tissue; same patient
48	7	A	95	1	F	W	GONADAL DYSGENESIS 46,XY		Right gonadal tissue
83	7	A	95	1	F	W	46,XY	B	Left gonadal tissue
868	2	C	107	26	F	W	46,XY		Gonadal dysgenesis; type unknown
978	7	C	143	15	F	W	46,XY/45,XO		Gonadal tissue
1491	3	A	95	8	F	W	46,XY		Phenotypic female
1709	4	C	109	11	F	W	46,X,i(Yq)	B	Phenotypic female
1150	4	C	125	23	M	W	BIOCHEMICAL MARKERS	A	Gd(+) ;Xg(a-); Sib
1152	4	C	125	46	M	W		A	Gd(-) ;Xg(a+); Father
1163	4	C	125	15	M	W		A	Gd(-) ;Xg(a+); sib of GM-1150; son of GM-1152

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Veri-fied	Remarks
<u>BIOCHEMICAL MARKERS</u>									
1153	4	C	125	42	M	W		A	Gd(+) ; Xg(a+); Father
1154	4	C	125	37	F	W		A	Phase=Gd Xg/Gd Xg ; Mother
1151	3	C	125	12	M	W		A	Gd(-) ; Xg(a-) ; Proband
1165	2	C	125	36	F	W			Phase=Gd Xg/Gd Xg
1166	3	C	125	39	F	W			Phase=Gd Xg/Gd Xg
1188	3	A	125	53	M	W	46,XY		Gd ; Xg(a+); karyotype based on leukocytes; Proband
1157	5	C	125	42	F	W	46,XX		Gd(+) Xg/Gd Xg(a+); Mother
1192	7	A	125	70	F	W	46,XX	A	Gd(+) /Gd(-) ; Xg(a+); Mother; karyotype based on blood; 100% G6PD(+) in red blood cells and 2 cell lines in hair roots
1193	4	A	125	50	F	W	46,XX	A	Gd(+) /Gd(-) ; Xg(a+); Proband; karyotype based on blood; 100% G6PD(-) in red blood cells, and 2 cell lines in hair roots
1717	3	B	125	39	M	W	46,XY		Xg(a-)
1862	2	B	125	37	M	W	46,XX		Xg(a-); Proband; Phenotypically male
1863	3	B	125	46	M	W	46,XY		Xg(?); Brother
1864	3	B	125	11	M	W	46,XY		Xg(-); Brother
1865	3	B	125	36	M	W	46,XX		Xg(+); Phenotypically male

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Verified	Remarks
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BIOCHEMICAL MARKERS

1869	2	B	125	15	M	W			Xg(-)Gd(-)
1870	2	B	125	12	M	W			Gd(+)Xg(-)
1973	4	B	125	67	F	W	47,XXX		Xg(a+),G6PD mutant; see Trisomy X
1871	3	B	125	47	F	W			Double heterozygote in repulsion; Gd(B)Xg(-)/Gd(Med) Xg(+)
324	5	A	125	22	M	W	47,XXY		Klinefelter's; homo G6PD def.; Med. type
325	4	A	125	30	M	W	47,XXY		Klinefelter's hetero G6PD def.; Med. type
543	12	A	75	7	M	P	46,XY,1qh+	B	Duffy blood group; mental retardation; heterochromatic marker

APPARENTLY NORMAL HUMAN FIBROBLAST CELL CULTURES

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Verified*	Remarks
NON-FETAL TISSUE**								
970	4	C	95	3 da.	M	W		
41	5	A	45	3 mo.	F	W	A	46,XX
302	6	A	95	10 mo.	M			
969	4	C	95	2	F	W	B	46,XX
498	4	A	128	3	M			
497	8	C	128	4	M	W		
408	8	A	128	5	F	W	A	46,XX
409	8	C	128	7	M	W	A	46,XY
499	7	A	128	8	M	W		
38	5	A	45	9	F	B	A	46,XX
500	8	A	128	10	M	W		
1652	7	A	133	11	F	W		
2036	5	J	133	11	F	W		
323	4	A	125	11	M	W		
316	3	A	26	12	M	W		
1651	7	C	133	13	F	W		
2037	8	J	133	13	M	W		
37	4	A	45	18	F	W	A	
726	3	A	26	26	F	O		Brother of GM-326, (XXXXY) See Aging Repository
975	3	C	26	26	F	W		
495	5	A	45	29	M			
23	5	A	95	31	F	W	A	46,XY
24	5	A	95	31	M	W	A	46,XX; mother of Down's child 46,XY; father of Down's child
43	5	A	45	32	F	B	A	46,XX
185	6	A	76	33	F	W	A	46,XX; family history is positive for increased cholesterol
964	2	C	26	33	M	W	A	46,XY

* Verification indicates normal human karyotype by banding

**See also Family Groups listing

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Verified	Remarks
<u>NON-FETAL TISSUE</u>								
2185	6	B	186	36	M	W		Normal spouse of a Huntington's Chorea patient
1650	6	A	133	37	F	W		
1653	8	A	133	37	M	W		
321	3	A	125	40	F	W	A	46,XX; Mother of GM-326 (XXXXY) and GM-323 Father of GM-326 (XXXXY)
322	4	A	125	40	M	W		
275	3	A	26	42	M	W	A	46,XY; see Aging Repository
730	3	A	26	45	F	W	A	46,XX; see Aging Repository
288	4	A	35	64	M	W		
1681	8	A	133	70	M	W		
1680	8	A	133	71	F	W		
1706	5	A	26	82	F	W	A	46,XX; see Aging Repository
731	3	A	135	94	M	W		See Aging Repository
<u>Family Groups**</u>								
41	5	A	45	3 mo.	F	W	A	46,XX; Daughter
37	4	A	45	18	F	W	A	Mother; see GM-637; SV40 trans.]
43	5	A	45	32	F	B	A	46,XX; Mother
38	5	A	45	9	F	B	A	46,XX; Daughter]
323	4	A	125	11	M	W		Son; Brother of GM-326 (XXXXY)
321	3	A	125	40	F	W	A	46,XX; Mother of GM-326 (XXXXY)
322	4	A	125	40	M	W		Father of GM-326 (XXXXY)
1652	7	A	133	11	F	W		Daughter
1651	7	C	133	13	F	W		Daughter
1650	6	A	133	37	F	W		Mother
1653	8	A	133	37	M	W		Father
1681	8	A	133	70	M	W		Paternal grandfather
1680	8	A	133	71	F	W		Paternal grandmother]

**See also preceding page

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Verified	Remarks
<u>FETAL TISSUE (SEE ALSO HUMAN AMNIOTIC FLUID CELL CULTURES)</u>								
9	3	A	26	3 mo.	M		A	46,XY; see Aging Repository
10	2	A	26	3 mo.	M	W	A	See Aging Repository
								46,XY, skin
1380	8	C	26	3 mo.	M	W	A	Lung, 12cm crown-rump, Same fetus
								46,XY
11	2	A	26	2 mo.	M		A	See Aging Repository
468	2	A	26		M			14.5 cm, crown-rump
1379	2	C	26	3 mo.	M			Lung, 10 1/2cm crown-rump
1381	4	C	26	3 mo.	M			Same fetus
1603	2	A	26	3 mo.	M	B		See Aging Repository
1604	2	A	26	3 mo.	M	B		Same fetus
								Lung; see Aging Repository

HUMAN AMNIOTIC FLUID CELL CULTURES

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
<u>BIOCHEMICAL MUTANT CONDITIONS</u>									
Cystinosis - 21980									
804	3	C	121	22 wk.F	M	W	--	B	46,XY
Fabry Disease (Diffuse Angiokeratoma) - 30150									
636	8	T	17	18 wk.F	M	W	y-	B	46,XY
Galactosemia (Transferase Deficiency) - 23040									
1743	3	B	77	6 mo.F	M	W	--		See GM-1741 Fibroblast
Homocystinuria (Cystathionine Synthase Deficiency) - 23620									
919	6	C	38	30 wk.F	F		--		46,XX
Hunter Syndrome - 30990									
1584	6	B	77	5 mo.F	M	W	y-	A	46,XY
Lesch-Nyhan Syndrome (HGPRT Deficiency) - 30800									
236	5	B	85	16 wk.F	M		y-	B	46,XY
2338	2 IMR	C	183		M		y-		See GM-2290 & 2291 Fibroblast & GM-2292 Lymphoid
Metachromatic Leukodystrophy - Infantile - 25010									
2095	3 IMR	B	105	18 wk.F	F	W	--		American Indian
2214	4 IMR	B	105	20 wk.F	M	W	--		Probable
<u>CHROMOSOMAL ABERRATIONS</u>									
Translocation									
477	9	C	48	14 wk.F	F	W			t(13;18) with trisomy 13q

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Verified	Remarks
<u>CHROMOSOMAL ABERRATIONS</u>								
<u>Inversions</u>								
2029	5 IMR	A	172	13 wk.F	F			46,XX,inv(9)
2055	7	A	172	17 1/2 wk.F	F			46,XX,inv(9)
<u>Trisomy</u>								
1993	7	A	172	21 wk.F	M	W	A	47,XXY
2269	9	C	109	18 wk.F	M	W		47,XXY
<u>APPARENTLY NORMAL</u>								
472	9	E	58	19 wk.F	M		B	46,XY
473	6	M	58	17 wk.F	M		B	46,XY
474	10	M	58	18 wk.F	M		A	46,XY
956	4	E	68	17 wk.F	F		B	46,XX
957	3	E	68	18 wk.F	F		B	46,XX
1420	11	C	109	18 wk.F	F	B	B	46,XX

HUMAN LYMPHOCYTE CULTURES WITH
BIOCHEMICAL MUTANT CONDITIONS

GM #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
DISORDERS OF AMINO ACID METABOLISM								
Immunoglobulins determined by Immunodiffusion.								
<u>Argininemia - 20780</u>								
2011	E	6	5	F	W	--	B	IgG wk; Kappa+; Lambda wk; IgA+
<u>Citrullinuria (Citrullinemia) - 21570</u>								
235	E	10	31	M	W	--	B	
1204	E	107	8 mo.	M	W	--		Proband; See GM-1044 Fibroblast; (biochemically confirmed) IgG wk Mother; IgG+, Kappa wk Father See GM-1058 Fibroblast; IgM wk Sib; IgG+, IgM+, Lambda+, Kappa wk
1205	E	107	26	F	W	+-(O)		
1206	E	107	27	M	W	+-(O)		
1207	E	107	4 1/2	F	W	+		
1685	E	157	1 da.	M		--		
<u>Cystathionuria - 21950</u>								
1454	E	8	7		O	--	B	Unresponsive to B6
1456	E	8	43		W	+-	B	IgA+, Kappa+
1566	E	8	16		W	--	B	
1562	E	8	42		W	+-	B	IgA wk, IgM wk
1781	E	8	18		W	--	B	Proband; IgG+, IgM wk, Lambda wk; B6 responsive
1461	E	8	39		W	+-	B	Parent; IgA+, IgG+, Kappa+, IgM wk
1807	E	8	Adult	M	W	+-	B	Father
1565	E	8	16	F	W	--	B	Proband, also has PKU, IgW wk, IgM+, IgG wk

GM #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
DISORDERS OF AMINO ACID METABOLISM								
Cystathionuria, continued								
1868	E	8	49		W	+- (O)		
Homocystinuria (Cystathionine Synthase Deficiency) - 23620								
1446	E	8		F	W	+-	B	IgA+, Lambda wk
1447	E	8		F	W	+-	B	Kappa+
1463	E	8	42	F	W	+-	B	Kappa+
1558	E	8		F		+- (O)		Kappa+, IgM+
1559	E	8		M		+- (O)		IgG+, Kappa wk
1560	E	8		M		--	B	Kappa wk
1528	E	8		M		+- (O)		IgA wk
1529	E	8	43	F		+- (O)		IgA+, IgG wk
1532	E	8		M		--		Kappa wk, IgM+
1808	E	8	45	M	W	+-	B	IgG wk, Kappa+
Maple Syrup Urine Disease - (Branched-Chain Ketoaciduria) - 24860								
1366	E	107	7	F	B	--		See GM-1364 Fibroblast; IgG+, Lambda wk
1655	E	107	5	F	W	--		See GM-1654 Fibroblast; IgG+
Phenylketonuria - 26160								
1565	E	8	16	F	W	--		Proband, also has Cystathionuria Father
1807	E	8	Adult	M	W	+- (O)		
DISORDERS OF CARBOHYDRATE METABOLISM								
Fucosidosis - 23000								
1023	E	8	31	M	W	+-	B	Father IgA+, Kappa+ Sib; Proband
1024	E	8	9	M	W	--	B	IgA+, Kappa+ Sib; Proband
1025	E	8	4	M	W	--	B	IgA+, Kappa wk Mother; IgG+
1026	E	8	29	F	W	+-	B	

GM #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks	
DISORDERS OF CARBOHYDRATE METABOLISM									
Galactosemia (Transferase Deficiency) - 23040									
148	E	10	8	M	W	--	B	Proband; IgG+, Lambda+ Father; IgG wk	
149	E	10	47	M	W	+-	B		
2412	E	8	15	M	W	--		Proband; formerly GM-1027 Sib Mother; IgG wk	
1028	E	8	16	F	W	--			
1029	E	8	41	F	W	+- (O)			
Glycogen Storage Disease									
Type II - Pompe Disease - 23230									
1778	E	8	16	F	W	+-	B	Daughter; Kappa+, IgM+ Proband Father	
1464	E	8	46	F	W	--	B		
1568	E	8	71	M	W	+-	B		
Mucopolysaccharidosis									
Type IH - Hurler Syndrome - 25280									
1034	E	8	2 1/2	F	W	--	B		
1867	E	8	5	F	W	--	B		
Type IH/S - Hurler/Scheie									
1032	E	8	15	M	I	--	B	IgA+	
Type IIIA - Sanfilippo A - 25290									
1780	E	8	8	M	W	--	B	Kappa wk, IgM+	
Type VI - Maroteaux-Lamy Syndrome - 25320									
1022	E	8	4	F	W	--	A	IgA+, see GM-519 Fibroblast	
DISORDERS OF LIPID METABOLISM									
Abetalipoproteinemia - 20010									
1453	E	8	18	F	W			IgG wk, Kappa+	
1810	E	8	23	M	W				

GM #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
<u>DISORDERS OF LIPID METABOLISM</u>								
<u>Gaucher Disease</u>								
<u>Types I & 3 (Juvenile & Adult, Cerebral) - 23100</u>								
1019	E	8	35	M	W	+-	B	Father of a Juvenile Gaucher; IgG+, Lambda
1020	E	8	49	F	W	+-	B	Mother of an Adult Gaucher; IgG+, Kappa+
1021	E	8	25	M	W	--	B	Adult Gaucher; IgA+, Kappa wk
1030	E	8	50	F	W	+-	B	Mother of a Juvenile Gaucher; IgA+, Kappa+
1031	E	8	53	M	W	+-	B	Father of a Juvenile Gaucher
<u>Hyperlipidemia - 14425</u>								
1455	E	8	33	M		+-	B	IgG wk, Kappa+
1783	E	8	31	F		+- (0)		Combined
<u>Hyperlipoproteinemia - 14440</u>								
<u>Type II - Familial Hypercholesterolemia</u>								
1448	E	8	18	F	W	+- (0)		IgA+
1766	E	8	23	F	W	+- (0)		IgA+, Kappa wk, formerly GM-1449
1450	E	8	49	M	W	+- (0)		Formerly GM-1452
1767	E	8	19	F		+- (0)		Kappa wk
1458	E	8	30	F		--		IgG+, Lambda wk
1459	E	8		F		--		IgG wk, Kappa+
1460	E	8		F		+- (0)		Kappa wk
1567	E	8	28	M	B	--		
1784	E	8	23	M		--		
<u>Metachromatic Leukodystrophy - 25000</u>								
1017	E	8	14	F	W	--	B	Kappa wk, IgM+
1018	E	8	25	F	P	+-	B	IgA+, Kappa+
1016	E	8	42	M	W	+- (0)		Father of a Juvenile MLD
1785	E	8	14	F	W	--		Juvenile

GM #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM								
HGPRT Deficiency								
467	E	83		F				Induced with ethylmethane sulfonate, 6-thioguanine resistant
Inosine Triphosphate Pyrophosphohydrolase (ITPase) Deficiency								
1619	E	177	29	F	W			Kappa+, IgM+; See GM-1617 Fibroblast
Lesch-Nyhan Syndrome (HGPRT Deficiency) - 30800								
1899	E	66	10	M	W	y-		Proband Mother; Kappa+ See GM-2290-2291 Fibroblasts; see GM-2338 Amniotic; fetal tissue
1900	E	66	Adult	F	W	+-(0)		
2292	E	183	F	M		y-		
Xeroderma Pigmentosum - 27870								
1646	E	133	21	F	W	--		See GM-1389 Fibroblast, Kappa+, IgM+
OTHER DISORDERS OF KNOWN BIOCHEMISTRY								
Menkes Syndrome (Kinky Hair Disease) - 30940								
1245	E	107	5 da.	M	W	y-		Sib of GM-220; See GM-1057 Fibroblast; Kappa+, IgM+
1982	E	107	2	M	W	y-		Proband; see GM-1981 Fibroblast; Kappa wk, IgM+
1984	E	107	26	F	W	+-(0)		Mother; see GM-1983 Fibroblast; IgG+, Kappa+, IgM wk
Porphyria								
Acute Intermittent Porphyria - 17600								
1363	E	112		F	W	+-	A	
2133	E	112	65	M	W	+-	A	Father, see GM-941 Fibroblast, Kappa+, IgM+
2134	E	112	63	F	W	++	A	Mother, see GM-940 Fibroblast, IgG+, Kappa+
2135	E	112	25	F	W	+-	A	Proband; see GM-933 Fibroblast

GM #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
OTHER DISORDERS OF KNOWN BIOCHEMISTRY								
Porphyria								
Acute Intermittent Porphyria, continued								
2229	E	112	23	F	W	+-	B	
2230	E	112	58	F	W	+-	B	
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY								
Adrenal Hyperplasia (Adrenogenital Syndrome) Type III - 20191								
2242	E	107	9 mos.	F	W	--		See GM-2241 Fibroblast
Basal Cell Nevus Syndrome - 10940								
1553	E	104	27	M	W	+- (0)		IgA+, IgG+, Kappa+, Lambda wk
1576	E	104	31	F	W	+- (0)		IgG+, Lambda wk
1578	E	104	39	M	W	+- (0)		IgA wk, Lambda+, IgM+
1656	E	104	53	M	W	+- (0)		See GM-1657; GM-1658 Fibroblast; IgA wk, Kappa+, IgM+
1726	E	104	58	F	W	+- (0)		See GM-1725, Fibroblast; IgA+, IgG wk;
2139	E	104		F	W	+- (0)		See GM-2138 Fibroblast
2099	E	104	31	M	W	+- (0)		See GM-2098 Fibroblast; IgA+
Cockayne Syndrome - 21640								
1712	E	6	21	M	W	--		
Cystic Fibrosis (Mucoviscidosis) - 21970								
504	E	25	19	M	W	--	B*	Probant; Lambda+, IgM+
505	E	25	Adult		W	+-	B*	Parent; IgG wk, Kappa+
506	E	25	8	F	W	--	B*	IgA+
608	E	25	21	M	W	--	B*	IgG+, Kappa+
897	M	89	12 1/2	M	W	--		IgG+, Lambda wk

*Production of ciliary dyskinesia factor

GM #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY								
Cystic Fibrosis, continued								
1442	E	8	31	M	W	+-	B*	Father
1443	E	8	28	F	W	+-	B*	Mother; IgA+, Lambda+
1445	E	8		F	W	--	B*	Proband; Kappa+
1530	E	8	5	M		--		Lambda+, IgM+
1531	E	8	23	M	W	--		IgG+, Kappa wk
1444	E	8	32	F		+-	B*	IgG+, Kappa wk
Diabetes Mellitus - 22210								
1241	E	34	37	M	W			See GM-1486 Fibroblast; maturity onset diabetes; IgG+
1817	E	137	65	M	W			MODY diabetes; primary affective disorder, biopolar; Lambda wk, IgM+
1905	E	34	32	F	W			MODY diabetes; Kappa+, IgM+
Family #1, Maturity Onset Diabetes - 22210								
1240	E	34	33	F	W			Sib; see GM-1122 Fibroblast; IgA+, Kappa+, Lambda+, IgM+
1246	E	34	46	M	W			Sib; see GM-1219 Fibroblast; IgG+, Kappa+, IgM+
1247	E	34	30	M	W			Sib; see GM-1237 Fibroblast; IgG+, Kappa+, IgM wk
1243	E	34	35	M	W			Sib; see GM-1430 Fibroblast; IgG+, Kappa+
1956	E	34	44	M	W			Sib; see GM-1955 Fibroblast, Kappa+, IgM+
1498	E	34		F	W			Sib; see GM-1497 Fibroblast; IgA wk, IgG+, Kappa+. IgM wk
1242	E	34	22	M	W			See GM-1435 Fibroblast; son of GM-1498; Lambda+, IgM+
1244	E	34	20	M	W			See GM-1496 Fibroblast; son of GM-1498; IgG+, Kappa+

*Production of ciliary dyskinesia factor

GM #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
<u>DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY</u>								
<u>Diabetes Mellitus</u>								
<u>Family #1, Maturity Onset Diabetes, continued</u>								
1410	E	34	15	F	W			See GM-1409 Fibroblast; mat. cousin of GM-1242-1244; only juvenile diabetic in family
1838	E	34	22	F	W			IgG+, Kappa+; pat second cousin of the 6 sibs; MODY type II; see GM-1837 Fibroblast
<u>Family #2, Juvenile with Optic Atrophy - 22230</u>								
1795	E	189	13	F	W			Optic atrophy; sister; see GM-1610 Fibroblast; Kappa+, IgM+
1796	E	189	16	M	W			Normal brother; IgG wk, Kappa+
1797	E	189	42	M	W			Normal father; see GM-1701 Fibroblast; Kappa+, IgM+
1799	E	189	18	F	W			Optic atrophy sister; see GM-1609 Fibroblast; Kappa+, IgM+
1800	E	189	6 1/2	F	W			Healthy sister, twin; IgG+
1801	E	189	6 1/2	F	W			Healthy sister, twin; Kappa+, IgM+, Lambda wk
1802	E	189	39	F	W			Normal mother; Kappa wk, Lambda+, IgM+
1803	E	189	11	F	W			Normal sister; Lambda+, IgM+
<u>Dysautonomia (Riley-Day Syndrome) - 22390</u>								
599	E	24		F	W	--		IgA+, Kappa+
600	E	24		M	W	+- (O)		IgM wk
601	E	24	10	M	W	--		Kappa+, IgM+
602	E	24		M	W	+- (O)		Kappa+, IgM+
603	E	24		F	W	+- (O)		IgG+
1465	E	8	21	F	W	--		IgM wk

GM #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY								
Dysautonomia (Riley-Day Syndrome), continued								
1466	E	8	11	M	W	--		Kappa+, IgM+
1777	E	8	22	F	W	--		Kappa+, IgM+
Dyskeratosis Congenita - 30500								
1775	E	107	6 1/2	M	W	y-		See GM-1774 Fibroblast; Kappa wk
Dystonia Musculorum Deformans - 12810; 22450								
2217	E	184	30	M	W	+- (0)		Age of onset 24 yrs.; IgA+, IgG+, Kappa+, Lambda wk; See GM-2215 Fibroblast
2256	E	184	14	F		--		Recessive form, age of onset 12 yrs.; See GM-2255 Fibroblast
2264	E	184	14 1/2	F	W	+- (0)		IgG+; Jewish background, age of onset 8 yrs.
2305	E	184	16	F	W	+- (0)		Age of onset 7 yrs.; see GM-2304 Fibroblast
2307	E	184	13	M	W	+- (0)		Age of onset 10 yrs.; Jewish background, see GM-2306 Fibroblast
2347	E	184	21	F	W	+- (0)		Swedish background, age of onset 5 yrs.
Huntington Chorea - 14310								
2078	E	136	25	F	W	+		At risk; daughter; see GM-2077 Fibroblast; IgA+
2080	E	136	48	F	W	+-		Proband; see GM-2079 Fibroblast; IgG+, Kappa wk
2146	E	186	54	M	W	+-		Proband; see GM-2147 Fibroblast
2148	E	186	54	F	W	++		Normal wife; see GM-2149 Fibroblast
2150	E	186	28	F	W	+		Daughter; see GM-2151 Fibroblast

GM #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY								
Huntington Chorea, continued								
2152	E	186	40	F	W	++		Normal widow of deceased proband; see GM-2153 Fibroblast
2154	E	186	20	F	W	+		Daughter; see GM-2155 Fibroblast
2156	E	186	16	F	W	+		Daughter; see GM-2157 Fibroblast
2158	E	186	19	F	W	+		Daughter; see GM-2159 Fibroblast
2160	E	186	21	M	W	+		Son; see GM-2161 Fibroblast
2162	E	186	12	M	W	+		Son; see GM-2163 Fibroblast
2166	E	186	58	F	W	+- (0)		Affected sister of GM-2165, Fibroblast Kappa+, IgG+
2168	E	186	52	M	W			Normal spouse of GM-2166, see GM-2169, Fibroblast
2178	E	186	38	F	W	+		Maternal niece of GM-2165, 2166, daughter of GM-2186
2180	E	186	52	F	W	++		Normal spouse of GM-2165
2182	E	186	21	F	W	+		Daughter of GM-2165, see GM-2183 Fibroblast
2186	E	186	60	F	W	+		Sister of GM-2165, 2166; IgA+, Kappa+, Lambda+; see GM-2187 Fibroblast
2176	E	186	26	M	W	+		Son of GM-2165, see GM-2177 Fibroblast
2188	E	186	63	M	W	++		Normal spouse of GM-2186, see GM-2189 Fibroblast
2170	E	186	22	F	W	+		Daughter; see GM-2171 Fibroblast
2172	E	186	52	F	W	+- (0)		Proband; see GM-2173 Fibroblast
2174	E	186	55	M	W	++		Spouse; see GM-2175 Fibroblast
Hyper IgM								
390	E	10	3	F	W			

GM #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY								
Neurofibromatosis (Von Recklinghausen Disease) - 16220								
1634	E	104	61	M	W	+- (0)		Kappa wk, IgM wk; see GM-1633 Fibroblast; Atypical
1861	E	104	41	M	W	+- (0)		Kappa+, IgM+; see GM-1860 Fibroblast
1641	E	104	19	F	B	+- (0)		See GM-1639 Fibroblast; IgG+, Kappa+, IgM+
Persistence of Fetal Hemoglobin (Hemoglobin F) - 14170								
2064	E	192	53	M	B	+- (0)		Heterozygous for Elliptocytosis; Kappa+, IgM+
Schizophrenia and Psychiatric Disorders - 18150								
1487	E	137	53	F	W			Normal Mother Proband Schizophrenic; Father]
1488	E	137	23	M	W			
1489	E	137	55	M	W			
1793	E	137	26	M	W			Affected son; see GM-1792 Fibroblast] Proband; see GM-1833; Atypical Psychosis Fibroblast; Lambda+, IgM+ Affected daughter; see GM-1835 Fibroblast; IgA+, IgG+, IgM+, Kappa+ Normal daughter; see GM-1882 Fibroblast; IgG+, Kappa+, Lambda+, IgM+
1834	E	137	56	M	W			
1836	E	137	27	F	W			
1883	E	137	25	F	W			

GM #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY								
Schizophrenia and Psychiatric Disorders, continued								
1825	E	137	56	M	W			"Carrier" for schizo., father; see GM-1824 Fibroblast; IgA+, Kappa+, IgM+, Lambda wk Proband; Schizophrenia
1827	E	137	24	F	W			IgA wk, Kappa+, IgM+, IgG+
1847	E	137	20	M	W			Normal first cousin; see GM-1846 Fibroblast; son of GM-1845; IgG wk, Kappa +
1885	E	137	31	M	W			Normal brother; Lambda+, IgM+
1845	E	137	55	M	W			Affected uncle, father of GM-1847; See GM-1844 Fibroblast
Sea-Blue Histiocyte Disease - 26960								
1913	E	163	24	F	W	--		See GM-1912 Fibroblast; IgG+, Kappa wk
Tuberous Sclerosis - 19110								
1636	E	104	17	M	B	+- (0)		See GM-1635 Fibroblast; IgA wk, Kappa wk
1638	E	104	20	F	B	+- (0)		Kappa+, IgM+; see GM-1644 Fibroblast
Waldenstrom Macroglobulinemia - 15360								
1501	E	10	69	M		+- (0)		

HUMAN LYMPHOCYTE CULTURES WITH
CHROMOSOMAL ABERRATIONS

GM #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
<u>SYNDROME WITH INCREASED CHROMOSOME BREAKAGE</u>								
<u>Ataxia-Telangiectasia - 20890</u>								
717	E	66	11	M	W	--		
718	E	66	9	M	W	--		Sib; Kappa+
719	E	66	13	F	W	--		Proband; Lambda+
736	E	66	Adult	M	W	+(O)		Father; Kappa+
781	E	66	4	F	W	--		Sib; Kappa wk
1525	E	54	19	F	W	--		Sib; Lambda+, IgM+; 46,XX,t(14;14)
1526	E	54	28	F	W	--		clone in short term lymphocytes Proband; IgG wk, Kappa wk, IgM+; 46,XX,t(X;14) clone in short term lymphocytes
<u>TRANSLOCATIONS</u>								
1203	E	66	30	F	W			46,XX,t(4;12); IgG wk, Kappa+, IgM+
1063	E	67	12 1/2	F	B			46,XX,t(4;13)(q21;q14); IgG wk
633	E	89	8	M	W			46,XY,t(7;10)(q2;q11);Kappa+, IgM+
1388	E	61	14	F	W			46,XX,t(9;13)(q22;q12)mat; IgA+, Kappa+
1561	E	8	36	F	W			46,XX,t(4;11)(q25;q13);IgA+, IgM wk
1261	E	107	13	F	W			45,XX,-13,-18,+t(13p;18p); IgA+, Kappa wk, Lambda+
2324	E	182	27	F	W			46,XX,t(16;22)(p13;q22); Mother of GM-2325 Fibroblast, see Trisomy 22; Kappa+
<u>TRISOMY/POLYSOMY</u>								
1416	E	61	27	F	W			48,XXXX; IgM wk

GM #	Culture Media	Submitter Code	Age	Sex	Race	Verified	Remarks
1202	E	66	7	M	W		49XXXXY; Kappa+, IgM+
1201	E	144	1	F	W		45,XX,-21,+t(21;21) derived from a Down's Syndrome patient with 46,XX,t(21;21)
1919	E	61	28	M	B		47,XY,+21,inv(9)(p13q21); see GM-1918 Fibroblast
1921	E	61	23	M	W		46,XY,+21,inv(9)(p13q13)mat; see GM-1920 Fibroblast; Kappa+, IgM+

UNCLASSIFIED HUMAN LYMPHOCYTE CULTURES

GM #	Culture Media	Submitter Code	Age	Sex	Race	Verified	Remarks
<u>Allergic Asthmatic</u>							
604	E	25	10	M	W		Kappa+
<u>Leukemia</u>							
463	E	10	60	M	W		Chronic Lymphocytic Leukemia
<u>Multiple Myeloma</u>							
1311	E	51	70	M	B		Bone marrow culture Kappa wk; Same patient IgG myeloma; IgG+, Lambda+
1312	E	51	70	M	B		
1500	E	10		M			

APPARENTLY NORMAL HUMAN LYMPHOCYTE CULTURES

GM #	Culture Media	Submitter Code	Age	Sex	Race	Verified	Remarks
892	E	24	12	F	W		IgM+
1953	E	89	22	F	W		
946	E	24	22	F	W		Kappa+, IgM+
1079	E	89	22	F	W		
131	E	10	23	F	W		IgG+, Kappa+
333	E	10	23	F	W		IgG+, Kappa+
546	E	89	23	F	W		
607	E	25	23	F	W		Kappa+, IgM+
922	E	24	23	F	W		IgG wk, IgM+, Kappa+
924	E	24	23	F	W		
923	E	24	24	M	W		IgA+, Lambda+
605	E	25	24	F	W		IgG+, Kappa+
130	E	10	25	M	W		IgG+, Kappa+
558	E	89	26	M	W		IgM+
1805	E	89	26	F	W		
536	E	89	27	M	W		
621	E	89	28	M	W		IgG+
1078	E	89	30	F	W		IgG+
893	E	24	32	F	W		Lambda+
894	E	24	32	F	W		
1806	E	89	33	M	W		Formerly GM-1074
1989	E	89	33	M	W		Formerly GM-1076
2184	E	186	36	M	W		Normal spouse of HD patient, see GM-2185 Fibroblast
1814	E	89	37	F	W		Formerly GM-1072
1815	E	89	42	M	W		Formerly GM-1075
1954	E	89	44	F	W		Formerly GM-1080
1990	E	89	51	F	W		Formerly GM-1077
1310	E	89	51	M	W		IgA wk, IgG+, Lambda+, IgM wk
1056	E	26	65	M	W		IgA+, Lambda+
606	E	24	Adult		W		Kappa+
891	E	24	Adult		W		

SV40 VIRUS TRANSFORMED CELL CULTURES

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
<u>Galactosemia (Transferase Deficiency) - 23040</u>									
638	48	C	51	3 mo.	M		--	A	SV40 transformed GM-54 culture, Gal-1-P uridyl transferase not present after crisis; G6PD-B, T-antigen positive; see GM-54 Fibroblast
639		C	51	8	M	B	--	A	SV40 transformed GM-52 culture, Gal-1-P uridyl transferase not present after crisis; G6PD-A; See GM-52 Fibroblast
<u>Lesch-Nyhan Syndrome (HGPRT Deficiency) - 30800</u>									
847		B	51	5 1/2	M	B	y-	A	SV40 transformed GM-177 culture; deficient for HGPRT, G6PD type A; see GM-2063 Fibroblast
<u>Apparently Normal</u>									
637	40	C	51	18	F	W		A	SV40 transformed GM-37 normal culture; T-antigen positive; See GM-37 Fibroblast

ANIMAL CELL CULTURES

GM #	Passage #	Culture Media	Submitter Code	Verified	Remarks
<u>Mouse</u> 86					
		K	40	A	Clone #745, Friend DMSO hemoglobin inducible secreting cell line
979		Q	158	A	Murine erythroleukemic line derived from a splenic focus in DDD mouse infected with Friend Leukemia virus; aneuploid and hypotetraploid; inducible for erythroid differentiation by DMSO
346		J	83	A	A9 cell line; deficient in HGPRT and selectable with 8-azaguanine
347		J	83	A	B82 cell line; deficient in thymidine kinase and selectable with 5-bromodeoxyuridine
<u>Syrian Baby Hamster Kidney</u> 345		<u>J</u>	83	A	TG-2 cell line; deficient in HGPRT and selectable with 6-thioguanine
348		J	83	A	B1 cell line; deficient in thymidine kinase and selectable with 5-bromodeoxyuridine
511		J	83	A	A5 cell line; deficient in thymidine kinase but with increased folate reductase

GM #	Passage #	Culture Media	Submitter Code	Verified	Remarks
<u>Chinese Hamster</u>					
215		R	22	A	V79 young adult male cell culture established by C. Ford and G. Yerganian and used in somatic cell genetics to isolate drug resistant mutants (lung tissue)
458		J	53	A	CHW normal male Chinese hamster cell culture established by C.C. Lin, U. of Calagary from skin taken from the ear
459		J	53	A	CHW-1102 cell culture derived from CHW normal culture after treatment with methylmethanesulphonate and selected with 8-azaguanine; culture is deficient in HGPRT

AGING CELL REPOSITORY CULTURES

Repos- itory #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Remarks
Alzheimer Disease of Brain - 10430								
GM 364	2	A	139	53	M	W	+-	See GM Repository
Bloom Syndrome (See GM Repository) - 21090								
GM 1620	22	C	44	8 mo.	F	W	--	
Cowden Disease - 15835								
AG 1964	3	B	195	64	M	W	+-	Affected
AG 1966	3	B	195	58	F	W	+-	Affected
AG 1965	3	B	195	60	F	W	++	Normal
AG 1967	3	B	195		M	W	++	Normal
AG 1968	3	B	195	49	F	W	++	Normal
AG 1969	2	B	195		F	W	++	Normal
Lowe Oculocerebrorenal Syndrome - 30900								
AG 1756	3	B	94	2 1/2	M		y-	See GM Repository
Mulibrey Nanism - 25325								
AG 2122	7	A	4	11	F		--	
Progeria (See GM Repository) - 26410								
GM 917	13	C	49	17	F		--	Atypical; cachectic dwarfism
AG 989	11	A	55	20	M			Atypical
AG 990	9	C	55		M			Classical
AG 991	7	A	55	4	M		--	
GM 1177	8	C	49	9	M		--	
GM 1178	18	C	49	34	M		--	
AG 1972	8	B	91	14	F	W	--	Atypical; progeria-like
AG 1710	5	B	197		M			

Repos-itory #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Remarks
Thanatophoric Dwarfism - 27365								
AG 711	2	A	143	1 da.	M	W		Cartilage Spleen Lung Skin Same patient
AG 712	2	A	143	1 da.	M	W		
AG 713	2	A	143	1 da.	M	W		
AG 714	3	A	143	1 da.	M	W		
Translocation								
AG 1839	7	B	31	10	F	W		45,XX,t(7q;13q); left side Hemihypertrophy; right side Same patient
AG 1840	7	B	31	10	F	W		
Xeroderma Pigmentosum (See GM Repository) - 27870								
AG 1951	12	C	201	40	M	W	--	From USSR From USSR; DeSanctis Cacchione type
AG 1952	12	C	201	7	M	W	--	
TUMOR PATIENTS								
Chronic Myelogenous Leukemia								
AG 1731	3	B	94	19	F			Irradiated Non-irradiated; lymphocytes show Phila. marker chromosome; patient had Wilm's tumor previously Same patient
AG 1732	3	B	94	19	F			

Repos-itory #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Remarks
Multiple Myeloma								
AG 1311	3	E	156	55	M	B		Bone marrow Lymphocyte Skin Skin Same patient
AG 1312	3	E	156	55	M	B		
AG 1360	3	A	156	55	M	B		
AG 1485	3	A	156	55	M	B		
Neuroblastoma								
AG 2202	3	B	94	9	F			Non-irradiated skin Irradiated skin Same patient
AG 2203	3	B	94	9	F			
AG 2272	3	B	94					
Osteogenic Sarcoma								
AG 2086	2	B	94	20	M			Skin
Retinoblastoma - 18020								
GM 913	3	C	65	2	M	W	++	See GM Repository; skin, does not grow well
GM 914	3	C	65	2	M	W	++	Conjunctiva; sporadic
AG 1142	4	C	94	2 1/4	F	W		46,XX,del(13)(q14q22) Same patient
AG 1484	3	E	94	2 1/4	F	W		
AG 1231	3	C	65	2	F	W	++	Lymphoid culture
AG 1232	3	E	65	2	F	W	++	Conjunctiva; sporadic
AG 1979	4	J	198	2	M		++	Tumor
AG 1946	6	J	198	1 1/2	M		++	Conjunctiva; sporadic
AG 1947	6	J	198	2	M			Conjunctiva; sporadic
AG 1123	3	A	94	9 mo.	F	B	+-	Conjunctiva; ident. twin hereditary
AG 1131	3	C	94	9 mo.	F	B	+-	Conjunctiva; ident. twin itary
AG 1223	8	C	161		F	B	+-	Conjunctiva; same patient as GM-1131
AG 1408	4	A	65	8 mo.	F	W	+-	Conjunctiva; hereditary
AG 1978	4	J	198	2	M		+-	Conjunctiva; hereditary
AG 1980	4	J	198	1	F		+-	Conjunctiva; hereditary
AG 1262	4	C	198	7 wk.	F	W	+-	Conjunctiva; hereditary

Repos- itory #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
<u>Wilms Tumor</u>									
AG 1615	3	B	160	9	M	B/W			Non-irradiated skin] Same patient
AG 1616	3	B	160	9	M	B/W			Irradiated skin
AG 2089	6	B	94	19	F				
AG 2129	2	B	199	10	M	B			Fibroblast culture] Same patient
AG 2130	2	E	199	10	M	B			Lymphoid line
AG 1894	2	B	94	12	M	B			Irradiated skin] Same patient
AG 1895	2	B	94	12	M	B			Non-irradiated skin
<u>APPARENTLY NORMAL CULTURES</u>									
GM 11	2	A	26	2 mo.F	M			A	See GM Repository, 46,XY
GM 1379	4	C	26	3 mo.F	M	W		A	Lung, Puerto Rican, 46,XY] Same
GM 1381	4	C	26	3 mo.F	M	W		A	See GM Repository, 46,XY] fetus
GM 9	3	A	26	3 mo.F	M			A	See GM Repository, 46XY
GM 10	2	A	26	3 mo.F	M	W		A	See GM Repository, 46XY] Same
GM 1380	8	C	26	3 mo.F	M	W		A	See GM Repository, 46XY] fetus
GM 1603	2	A	152	3 mo.F	M			A	Lung, 46XY
GM 1604	2	A	152	3 mo.F	M			A	See GM Repository;] Same fetus
AG 1437	2	A	152	3 da.	M	W			Lung
AG 1439	3	A	152	3 da.	M	B			Foreskin
AG 1440	4	A	152	3 da.	M	B			Foreskin
AG 1518	2	A	152	3 da.	M			A	Foreskin
AG 1519	2	A	152	3 da.	M			A	Foreskin: 46,XY
AG 1520	2	A	152	3 da.	M			A	Foreskin: 46,XY
AG 1521	2	A	152	3 da.	M				Foreskin
AG 1522	2	A	152	3 da.	M				Foreskin
AG 1523	2	A	152	3 da.	M			A	Foreskin
GM 316	3	A	26	12	M	W		A	Foreskin; 46,XY
AG 2220	3	B	200	22	M	W		A	See GM Repository
GM 275	3	A	26	42	M	W		A	See GM Repository
GM 730	3	A	26	45	F	W		A	See GM Repository

Repos- itory #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Verified	Remarks
APPARENTLY NORMAL CULTURES								
AG 2257	1	B	200	46	F	W		Skin Same Lung patient Skin fibroblasts See GM Repository; formerly GM-237 See GM Repository
AG 2258	4	B	200	46	F	W		
AG 2222	2	B	200	49	M	W		
GM 1706	4	A	26	82	F	W	A	
GM 731	3	A	26	95	M	W	A	
SPECIALLY CHARACTERIZED DIPLOID CELL CULTURES*								
IMR 90	1	A	26	16 wk.F	F	W	A	Lung; see guidelines for distribution of these cells, Science 196:60-63 (1977)
IMR 91	1	A	26	14 wk.F	M	W	A	Lung Same
IMR 91	1	A	26	14 wk.F	M	W	A	Skin fetus

*Large quantity of characterized cells frozen at early passage

CODE FOR CULTURE MEDIA

For reference see: Morton, Helen C., A survey of Commercially Available Tissue Culture Media. In Vitro 6:89-108, 1970.

- A - McCoy's 5A with 20% fetal bovine serum (FBS) not inactivated (Iwakata and Grace modification)
- B - Ham Fl2 with 20% FBS not inactivated
- C - Minimum Essential Medium Eagle in Earle's BSS with 20% FBS not inactivated (with 2x concentration of essential and non-essential amino acids and vitamins)
- D - Roswell Park Memorial Institute 1640 with 10% FBS inactivated 60°C for 1/2 hour
- E - Roswell Park Memorial Institute 1640 with 20% FBS inactivated 60°C for 1/2 hour
- F - Minimum Essential Medium Eagle in Earle's BSS with 30% FBS not inactivated (with 2x concentration of essential and non-essential amino acids and vitamins)
- G - Ham Fl2 with 10% FBS not inactivated
- H - McCoy's 5A with 10% FBS not inactivated (Iwakata and Grace modification)
- I - Roswell Park Memorial Institute 1640 with 20% FBS not inactivated
- J - Minimum Essential Medium Eagle in Earle's BSS with 10% FBS not inactivated (with 2x concentration of essential and non-essential amino acids and vitamins)

- K - Minimum Essential Medium Eagle in Earle's BSS with 15% FBS
not inactivated (with 2x concentration of essential and
non-essential amino acids and vitamins)
- L - Minimum Essential Medium Eagle in Earle's BSS with 12% FBS
not inactivated (with 2x concentration of essential and
non-essential amino acids and vitamins)
- M - Roswell Park Memorial Institute 1640 with 30% FBS inactivated
60°C for 1/2 hour
- N - Minimum Essential Medium Eagle in Earle's BSS with 10% FBS
inactivated 60°C for 1/2 hour (with 2x concentration of
essential and nonessential amino acids and vitamins)
- O - Roswell Park Memorial Institute 1640 with 30% FBS not inac-
tivated
- P - Ham Fl2 with 10% FBS inactivated 60°C for 1/2 hour
- Q - Ham Fl2 with 20% FBS inactivated 60°C for 1/2 hour
- R - Minimum Essential Medium Eagle in Earle's BSS with 5% FBS
not inactivated (with 2x concentration of essential and non-
essential amino acids and vitamins)
- S - Minimum Essential Medium in Hanks' BSS with 10% FBS not
inactivated (with 2x concentration of essential and non-
essential amino acids and vitamins)
- T - Minimum Essential Medium in Hanks' BSS with 20% FBS not
inactivated (with 2x concentration of essential and non-
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- U - Ham Fl2 with 30% FBS not inactivated

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2. An (X;14) translocation, unbalanced, 47 chromosomes. Repository identification No. GM-74. J. Opitz, P.D. Pallister. Cytogenet. Cell Genet. 12:291-292 (1973)
3. A (14;22) Robertsonian translocation, 45, chromosomes, Repository identification No. GM-5. H.A. Lubs, F.H. Ruddle. Cytogenet. Cell Genet. 12:368-369 (1973)
4. A (15;18) translocation, unbalanced, 45 chromosomes. Repository identification No. GM-17. D.S. Borgaonkar. Cytogenet. Cell Genet. 12:370-371 (1973)
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18. A (1;2) translocation, balanced, 46 chromosomes. Repository identification No. GM-257. O.S. Alfi, R.C. Miller, A.E. Greene, L.L. Coriell. Cytogenet. Cell Genet. 14:154-155 (1975)

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31. A (Y;15) translocation, 46 chromosomes. Repository identification No. GM-118. N. Hahnemann, R.C. Miller, A.E. Greene, L.L. Coriell. Cytogenet. Cell Genet. 15:408-409 (1975)
32. Beta-Glucuronidase deficiency mucopolysaccharidosis. Repository identification No. GM-121. W.S. Sly, A.E. Greene, L.L. Coriell. Cytogenet. Cell Genet. 15:410-412 (1975)
33. A (14;15) Robertsonian translocation, 45 chromosomes. Repository identification No. GM-479. M.S. Golbus, C.J. Epstein, M.M. Aronson, A.E. Greene, L.L. Coriell. Cytogenet. Cell Genet. 17:61 (1976).
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47. A (4;7) translocation, 46 chromosomes. Repository identification No. GM-773. L.G. Jackson, B. Bozarth, M.M. Aronson, A.E. Greene, L.L. Coriell. In Press.
48. Duarte variant-galactosemia heterozygote. Repository identification No. GM-1996. G.N. Donnell, W.G. Ng, O.S. Alfi, A.E. Greene, L.L. Coriell. In Press.
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HL-A antigens of human lymphocyte cell cultures listed on pages 107 through 122. The Human Genetic Repository is indebted to Dr. Roger Kennett, Department of Human Genetics, University of Pennsylvania Medical School, Philadelphia, Pennsylvania.

For further data showing reactions with a selected set of NIH antisera, contact Dr. Kennett.

GM #	HL-A	GM #	HL-A
235	(Aw31,33)A9,Bw15,B12	1366	A2(A9),B8,B12
558	A3,A11,Bw15,Bw22	1410	A2,Aw24,Bw35,B13(B12)
892	A2(A9),B12,B18	1446	A2,3,Bw35,Bw17
1027	A2-B12/A1-B8	1447	A1,A10,B7,(Bw16)
1028	A1/A2-B12	1454	A28,A2,Bw40,Bw21
1029	A2-B12/A28-Bw35	1455	A11(A3),Bw22,(Bw40)
1032	A9,A10,B13	1456	A1,B13,Bw27
1056	A9,29,B12,Bw15	1461	A2,A3,Bw22,Bw40
1063	A1,B12,Bw17	1463	A2,A11,B27,Bw22
1204	A2-B12/A28-Bw35	1500	A2(A3),B12,Bw15
1205	A28-Bw35/A1-B13	1501	A3,A9,Bw22,(B12)
1206	A11-Bw15/A2-B12	1525	A28,(Aw31,33),B12
1207	A11-Bw15/A28-Bw35	1526	A9,A10,Bw17,B13(Bw40)
1241	A3,A10,B13,B12	1528	A10,(A3),B18,Bw22
1245	A9,A10,B13,B18	1529	A10,Aw23,B8,B18
1261	A1,A2,Bw15(B18)	1530	A2,A11,Bw17,Bw35
1311	A10,A11(A3),B12	1531	A2,(A9),B7,(Bw40)
1312	A10,A11(A3),B12	1532	A2,B7,Bw22,(B18)

GM #	HL-A	GM #	HL-A
1539	A3,Aw30,(B12),Bw15	1814	(A1),A2,A11,B8,Bw40
1553	A28,A3,Bw22,B13(B12)	1815	A2,A9,B7,B13(B12)
1556	A1,B13,B27	1817	A10-Bw35/A2-Bw21
1558	A10,Aw24,B7,Bw15	1819	A10-Bw35/Aw24-B13
1559	A3,A10,Bw15,B27	1821	A2-Bw21/A1-Bw22
1560	A2,3,B7,B18	1823	A10-Bw35/A28-B14
1561	A10,Aw24,Bw35,(B12)	1825	A2,A3,Bw22
1562	A3,A10,B13,B18	1827	A11-B12/A2(A28)-Bw16
1565	A28,Aw24,B7,Bw35	1836	A28-Bw16/A3-Bw35
1566	A1,A28,B13,Bw35	1838	A28(A2),Aw30,B12(B13),Bw35
1655	A1,A2,B13,Bw17(B23) (Bw40)	1845	Aw32-Bw22/A2(A28)-Bw16
1685	A1,Aw32,B18,B27	1847	A11-B12/Aw32-Bw22
1712	Aw32,A10,Bw15,B14,(A3)	1853	Aw32,w24,Bw40,B12
1715	A2,A9,Bw40,(B7)	1855	Aw32(A1),B12,B27
1716	A10,Bw15	1857	A9,A11,B7,Bw15
1726	A28,Aw30,B5,Bw16	1861	A3,A2;B27,Bw22(Bw40)
1775	Aw30,(Bw40),B13,B5	1867	A2,Aw30,B12,Bw22
1779	A2,10,(B18)(Bw35)	1868	A28,A10,Bw15,Bw16
1785	A2,A29,B12,B16	1883	A28,Bw35
1793	Aw24,B18,B13	1884	A29,Aw31,33,B12,Bw15
1805	A1,A9,B18,(B13)	1885	A2,A3,Bw35,Bw22
1806	A1,A2,Bw17,B18,(Bw22)	1899	A2,A11,B18,B5(Bw35)
1807	A1,A2,Bw35,B13	1900	A2,A28,B18,B5(Bw35)
1808	A2,A10,B12,B27	1901	A1(A9),A11,B8,Bw35
1810	A1,A10,B7,B13	1902	A2,A11,B13

GM #	HL-A	GM #	HL-A
1905	A11, Bw35		
1913	A2, Aw31, B18, B13		
1930	A2, A29, B12, Bw35		

APPENDIX E

NSF CELL CULTURE CENTERS

The Cell Culture Centers at the Massachusetts Institute of Technology and the University of Alabama at Birmingham are accepting applications for large scale cell and virus production required for highly meritorious research projects. These centers established by the National Science Foundation are intended to serve as a facility and research resource for scientists throughout the country.

The MIT Center is headed by Dr. Phillips W. Robbins of MIT and Dr. Richard L. Davidson of Harvard Medical School. This facility is designed for large scale monolayer and suspension cultures and has experience with a large number of different cell lines and viruses and also with diploid human fibroblasts. The University of Alabama center is headed by J. Claude Bennett of the Department of Microbiology and Ronald T. Acton of the Diabetes Research and Training Center. This facility is designed for large scale suspension cultures with a capability of producing one kilogram or more of cells per week. Their experience to date is primarily with lymphoblastoid cell lines. The purpose of these centers is to produce cells and viruses on a large scale in order to allow scientists to conduct novel and important experiments in basic biological research that could not be accomplished with the materials and resources in the investigators' own laboratory. Approval of applications is on

a competitive basis, based on the merit of the research project as evaluated by Centers' Steering Committees.

Examples of recent activities at the Centers are:

1. Production of 100 mg Sindbis Virus propagated in 200 roller bottles of cells for use in x-ray structural studies.
2. Production of 750 roller bottles of SV40 transformed BALB 3T3 cells for purification and characterization of T antigen.
3. Growth of 300 liters of mouse leukemia cells for isolation and structural analysis of a specific lysine transfer RNA implicated in the control of cell division.
4. Growth of 3.1×10^{12} BW 5147 cells for the isolation and structural analysis of H-2k and Thy-1 alloantigens.

Following approval of an application, the investigator sends a stock of cells or virus to the center. The stock is then grown to the requested amount, under conditions of careful handling and rigorous screening for contaminants (including mycoplasma), and prepared according to the needs of the investigator. Investigators will be asked to pay minimal costs to cover media and other supplies.

The procedure for applying involves submitting a letter specifying the amount of cell or virus required along with a brief description of the relevant research project and available supporting material. Applications or inquiries should be addressed to:

Mr. Donald Giard
Cell Culture Center
E17-321
Massachusetts Institute of Technology
77 Massachusetts Avenue
Cambridge, MA 02139

OR

Dr. Ronald T. Acton
Diabetes Research and Education Hospital
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