

RESEARCH PROJECT

"Molecular Genetics of Alport syndrome"
Granted by Telethon years 1994-1996

"Molecular basis of AMME contiguous gene syndrome (A=Alport phenotype, M=mental retardation, M=midface hypoplasia and E=elliptocytosis) and single gene / phenotype correlation"
Granted by Telethon years 1996-1998 and years 2000-2002

"Autism and Rett syndrome complex: molecular basis of complex disorders"
Granted by Telethon years 2002-2004

"Cell line and DNA bank of Rett syndrome and other X-linked mental retardation"

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Fondata nel 1241

MEDICAL GENETICS

MOLECULAR BIOLOGY DEPARTMENT

SERVICES

GUIDELINES

LIST OF CELL LINES

LIST OF DNA SAMPLES

FORMS

CONTACT INFO

CELL LINE AND DNA BANK OF RETT SYNDROME AND OTHER MENTAL RETARDATION

SERVICES

- Establishment of EBV-transformed lymphoblastoid cell lines from human peripheral blood lymphocytes.
- DNA extraction.
- Storage:
cryopreservation of transformed cell lines at -135°C

storage of DNA at -20°C
- Distribution of cell lines and DNA.

GUIDELINES

SENDING SAMPLES

Users who want to access to the Bank Services for establishing and storing cell lines and/or DNA, should contact the Bank at least 48 hours before sending the specimens. Shipments should be sent early in the week (usually Mondays and Tuesdays) to ensure arrival before the following weekend.

Users must provide relevant clinical data of the patient on a specific form. The form must be sent with the sample and is available electronically or may be obtained by e-mail contacting the Bank. If more than one sample is sent, a form for each sample must be included.

Before sending the specimens, users have to obtain informed consent from the patient or his/her parents. Before signing the consent patients should be informed about the possibility of banking the sample, the possibility of authorising or not the use of the sample for research and the possibility of knowing or not the information that could result from subsequent investigations. Moreover, patients should be informed about the security procedures employed to ensure confidentiality.

To protect confidentiality, the samples will be stripped of personal identifiers and assigned a code. Access to the data linking the code to the patient is restricted to the Bank staff and is protected with a password.

Details of cell lines and DNA source will remain confidential.

Users can limit the distribution their cell lines and DNA samples. In case of limitation we shall request users' permission before any transfer of the sample.

For lymphblastoid cell lines establishment a minimum of 5 ml of blood in sodium heparin tubes is required and should arrive as soon as possible after sampling, preferably within 48 hours.

For DNA extraction a minimum of 5 ml of blood containing 100 microlitres of sterile EDTA 0.5M pH8 is required. Samples should be sent at room temperature immediately after sampling and should arrive within 24 hours. Samples which can not be sent immediately must be shipped frozen.

No payment is required for the service (except for shipping charges).

ORDERING SAMPLES

Cell lines and DNA samples are distributed only to qualified professionals. Samples must be used for research purposes only; no commercial use is allowed. The specimens cannot be distributed to other investigators without previous written permission of the Bank curator. If the samples are quoted in any publication the investigator must include the Bank in the acknowledgements; a reprint of publications will be sent to the laboratory.

No materials are released prior to receipt a signed ordering form with a brief project description and the agreement to accept the above reported terms.

The request form is available electronically or may be obtained by e-mail contacting the Bank. The form must be sent by mail. If an user wants to order more than one sample, a form for each sample must be submitted.

No payment is required for the service (except for shipping charges).

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LIST OF CELL LINES

data arrivo	codice	malattia	provenienza
25-04-1998	ATS-MR38	ATS-MR sindrome da geni contigui	Siena
11-06-1998	ATS-MR2102	ATS-MR sindrome da geni contigui	Siena
25-09-2001	MR 062	Ritardo Mentale	altri
25-09-2001	MR0125	Ritardo Mentale	altri
25-09-2001	MR1180	Ritardo Mentale	altri
25-09-2001	MR9535	Ritardo Mentale e SPG	altri
28-09-2001	MR 022	Ritardo Mentale	altri
04-10-2001	MR 055	Ritardo Mentale	altri
13-12-2001	MR 046	Ritardo Mentale	altri
16-01-2002	MR118	Ritardo Mentale	altri
11-02-2002	ATS-MR2376	Ritardo Mentale e microematuria	Siena
15-03-2002	ATS-MR2377	Ritardo Mentale e microematuria	Siena
06-02-2002	MR oe	Ritardo Mentale e SPG	altri
09-05-2002	RETT 563	Rett atipica	Siena
09-05-2002	RETT 565	Rett	Siena
22-05-2002	RETT 571	Rett	Siena
22-05-2002	RETT 574	Rett	Siena
23-05-2002	MR97945	Ritardo Mentale	altri
23-05-2002	MR97947	Ritardo Mentale	altri
13-06-2002	RETT 342	Rett	Siena
13-06-2002	RETT592	Rett	Siena
13-06-2002	MR 03	Ritardo Mentale	Siena
08-05-2002	RETT 51	Rett	Siena
10-05-2002	MR 065	Ritardo Mentale	altri
03-07-2002	RETT 10	Rett	Siena
03-07-2002	RETT 304	Rett	Siena
03-07-2002	RETT 603	Rett	Siena
03-07-2002	RETT 608	Rett	Siena
08-07-2002	MR1453	Ritardo mentale	altri
19-06-2002	MR 09	Ritardo mentale	altri
19-06-2002	MR032	Ritardo mentale	altri
19-06-2002	MR050	Ritardo mentale	altri
29-07-2002	MR 065	Ritardo Mentale	altri
29-07-2002	MR 066	Ritardo Mentale	altri
29-07-2002	MR3742	Ritardo Mentale	altri
25-07-2002	RETT 585	Rett	Siena
08-08-2002	RETT 550	Rett	Siena
13-08-2002	RETT 406	Rett	Siena
22-08-2002	RETT 43	Rett	Siena

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LIST OF DNA SAMPLES

29-01-1998	MR 1	Ritardo Mentale	Siena
15-05-1998	MR 1	Ritardo Mentale	Siena
06-06-1998	MR 2	Ritardo Mentale	Siena

9-07-1998	MR 5	Ritardo Mentale	Siena
9/08/1998	MR 6	Ritardo Mentale	Siena
29-01-1999	MR 7	Ritardo Mentale	Siena
29-03-1999	MR 8	Ritardo Mentale	Siena
15-05-1999	MR 9	Ritardo Mentale	Siena
9-07-1999	MR 10	Ritardo Mentale	Siena
28-01-2000	MR 11	Ritardo Mentale	Siena
16-03-2000	MR 19	Ritardo mentale	Siena
14-05-2000	MR 20	Ritardo mentale	Siena
17-06-2001	MR 21	Ritardo mentale	Siena
9-07-2001	MR 22	Ritardo mentale	Siena
06-11-2001	MR 23	Ritardo Mentale	Siena
06-11-2001	MR 24	Ritardo Mentale	Siena
06-11-2001	MR 25	Ritardo Mentale	Siena
06-11-2001	MR 26	Ritardo Mentale	Siena
06-11-2001	MR 27	Ritardo Mentale	Siena
03-01-2002	MR 28	Ritardo Mentale	Siena
08-01-2002	MR 29	RM +microcefalia	Siena
08-01-2002	MR 30	RM +microcefalia	Siena
08-01-2002	MR 31	RM +microcefalia	Siena
08-01-2002	MR 32	RM +microcefalia	Siena
29-01-2002	MR 33	RM +microcefalia	Siena
29-01-2002	MR 34	RM +microcefalia	Siena
01-02-2002	MR 35	RM +microcefalia	Siena
01-02-2002	MR 36	RM +microcefalia	Siena
01-02-2002	MR 37	RM +microcefalia	Siena
18-04-2002	MR 38	Ritardo Mentale	Siena
18-04-2002	MR 39	Ritardo Mentale	Siena
22-04-2002	MR 40	Ritardo Mentale	Siena
22-04-2002	MR 43	Ritardo Mentale	Siena
22-04-2002	MR 44	Ritardo Mentale	Siena
15-05-2002	MR 41	Ritardo Mentale	Siena
15-05-2002	MR 42	Ritardo Mentale	Siena
06-06-2002	MR 45	Ritardo Mentale	Altro
06-06-2002	MR 46	Ritardo Mentale	Altro
06-06-2002	MR 47	Ritardo Mentale	Altro
07-06-2002	MR 48	Ritardo Mentale	Altro
07-06-2002	MR 49	Ritardo Mentale	Altro
07-06-2002	MR 50	Ritardo Mentale	Altro
9-07-2002	MR 51	Ritardo Mentale	Altro
9-07-2002	MR52	Ritardo Mentale	Altro
9-07-2002	MR53	Ritardo Mentale	Altro
9-07-2002	MR 54	Ritardo Mentale	Atro
9-07-2002	MR 55	Ritardo Mentale	Altro
9-07-2002	MR 56	Ritardo Mentale	Altro
9-07-2002	MR 57	Ritardo Mentale	Altro
9-07-2002	MR 58	Ritardo Mentale	Altro
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01/06/2000	RET 330	RTT	Siena
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04/07/2000	RET 357	RTT	Siena
10/07/2000	RET 358	RTT	Siena
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12/07/2000	RET 365	RTT	Siena
12/07/2000	RET 360	RTT	Siena

12/07/2000	RET 361	RTT	Siena
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18/07/2000	RET 368	RTT	Siena
19/07/2000	RET 369	RTT	Siena
26/07/2000	RET 370	RTT	Siena
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04/08/2000	RET 379	RTT	Siena
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10/08/2000	RET 382	RTT	Siena
10/08/2000	RET 383	RTT	Siena
10/08/2000	RET 384	RTT	Siena
11/08/2000	RET 385	RTT	Siena
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01/09/2000	RET 387	RTT	Siena
11/09/2000	RET 388	RTT	Siena
11/09/2000	RET 389	RTT	Siena
11/09/2000	RET 390	RTT	Siena
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14/05/2001	RET 496	RTT	Siena
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26-06-2001	RET 502	RTT	Siena
27-06-2001	RET 503	RTT	Siena
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25/7/2001	RET 509	RTT	Siena
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6-09-2001	RET 511	RTT	Siena

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16-10-2001	RET 519	RTT	Siena
16-10-2001	RET 520	RTT	Siena
17-10-2001	RET 521	RTT	Siena
19-10-2001	RET 522	RTT	Siena
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28/11/2001	RET 531	RTT	Siena
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03/12/2001	RET534	RTT	Siena
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19-12-2001	RET 538	RTT	Siena
19-12-2001	RET 539	RTT	Siena
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21/12/2001	RET 541	RTT	Siena
21/12/2001	RET 542	RTT	Siena
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21/01/2002	RET 529	RTT	Siena
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19-02-2002	RET 548	RTT	Siena
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5-03-2002	RET 550	RTT	Siena
5-03-2002	RET 551	RTT	Siena
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8/03/2002	RET 553	RTT	Siena
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7-02-2002	RET 560	RTT	Siena

15-03-2002	RET 561	RTT	Siena
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06-06-2002	RET 610	RTT	Siena
06-06-2002	RET 611	RTT	Siena

07-06-2002	RET 612	RTT	Siena
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2-07-2002	RET 615	RTT	Siena
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9-07-2002	RET 617	RTT	Siena
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16-07-2002	RET 625	RTT	Siena
16-07-2002	RET 626	RTT	Siena
26/07/2002	RET 627	RTT	Siena
26/07/2002	RET 628	RTT	Siena
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13-08-2002	RET 630	RTT	Siena
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20-08-2002	RET 633	RTT	Siena
21-08-2002	RET 369	RTT	Siena
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27-08-2002	RET 635	RTT	Siena
27-08-2002	RET 636	RTT	Siena
28-08-2002	RET 637	RTT	Siena
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02-09-2002	RET 639	RTT	Siena
02-09-2002	RET 640	RTT	Siena
3-09-2002	RET 641	RTT	Siena

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FORMS

Please read guidelines carefully before filling in and signing forms.

INFORMED CONSENT



AZIENDA OSPEDALIERA SENESE

U.O. GENETICA MEDICA

CONSENSO INFORMATO PER LA CONSERVAZIONE DI MATERIALE BIOLOGICO "BANCA DI LINEE CELLULARI E DNA DI PAZIENTI CON SINDROME DI RETT E ALTRE FORME DI RITARDO MENTALE X-LEGATO"

Siena, _____

Il sottoscritto.....

nato/a a il

residente
in..... Via.....CAP.....

in sede di consulenza genetica presso
..... il

informato dal dott.che:

Il proprio DNA e/o linee cellulari derivate da proprie cellule potranno essere conservati presso la "banca di linee cellulari e dna di pazienti con sindrome di Rett e altre forme di ritardo mentale X-legato".

Tali campioni verranno utilizzati solo ed esclusivamente per scopi di ricerca.

Verranno garantiti l'anonimato e la riservatezza sulla provenienza del campione.

Autorizza la conservazione del suddetto materiale biologico presso la "banca di linee cellulari e DNA di pazienti con sindrome di Rett e altre forme di ritardo mentale X-legato", secondo le modalità tecniche che questo laboratorio ritiene opportune.

Letto e controfirmato
Il Medico consulente

Letto e controfirmato
Il consultando

per i minori:

Letto e controfirmato
Il genitore

Autorizza/Non autorizza l'uso del suddetto materiale biologico a fini di ricerca scientifica

Letto e controfirmato

Letto e controfirmato

Il Medico consulente

Il consultando

per i minori:

Letto e controfirmato
Il genitore

Richiede che tutte le informazioni rilevanti per la salute mia e della mia famiglia che si rendessero disponibili:

Mi siano comunicate nel rispetto del diritto alla riservatezza dei dati personali. Nel caso i risultati riguardassero altri membri della famiglia o minori o soggetti non in grado di intendere e volere, e soprattutto in caso di diagnosi presintomatica di patologie genetiche ad esordio tardivo, le informazioni saranno date nel rispetto del diritto di scelta dell'interessato o di chi ne fa le veci.

Non mi siano comunicate.

Letto e controfirmato
Il Medico consulente

Letto e controfirmato
Il consultando

per i minori:

Letto e controfirmato
Il genitore

FORM FOR SENDING SAMPLES



AZIENDA OSPEDALIERA SENESE

U.O. GENETICA MEDICA

Policlinico "Le Scotte" Viale Bracci, 53100 Siena. Tel. (0577) 233303 - 233324 - 585316 .
Fax: 233325, E-mail: geneticamed@unisi.it

Paziente:

Cognome:
Nome:
Nato a:
Il:

Richiedente:

Cognome:
Nome:

Istituzione:

Indirizzo:

Telefono:

Fax:

e-mail:

Campione inviato il:

Per: Linea Cellulare
 DNA

Limitazione della distribuzione del DNA/ linea cellulare ad altri ricercatori a scopo di ricerca:

SI

NO

DATI CLINICI

Fenitipo: Affetto
 Sano
 Non conosciuto

Diagnosi:

Dettagli clinici
rilevanti:

Luogo

Data

Firma del richiedente

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FORM FOR REQUESTING SAMPLES



AZIENDA OSPEDALIERA SENESE

U.O. GENETICA MEDICA

Policlinico "Le Scotte" Viale Bracci, 53100 Siena. Tel. (0577) 233303 - 233324 - 585316 .
Fax: 233325, E-mail: geneticamed@unisi.it

Richiedente:

Cognome:

Nome:

Istituzione:

Indirizzo:

Telefono:

Fax:

e-mail:

Materiale richiesto: Linea Cellulare
 DNA

Codice:

Descrizione del Progetto:

Io sottoscritto dichiaro di accettare le seguenti condizioni:

Il campione inviato può essere usato esclusivamente per scopi di ricerca

Il campione non può essere distribuito ad altri laboratori senza previo permesso scritto da parte della Banca.

In caso di pubblicazione di risultati ottenuti facendo uso del campione ricevuto la Banca verrà inclusa nei ringraziamenti ed un reprint del lavoro verrà ad essa inviato. Le spese di spedizione sono a mio carico.

Luogo

Data

Firma del richiedente

-----interruzione pagina-----

CONTACT INFO

To send and request samples please contact:

Prof. Alesandra Renieri
Dr. Ilaria Meloni

Address: Dipartimento di Biologia Molecolare
Unità Operativa di Genetica Medica
Policlinico Le Scotte
Viale Bracci 22
53100 Siena, Italy

Telephone: 39 0577 233303
39 0577 233259

FAX: 39 0577 233325

e-mail: geneticamed@unisi.it