THE · ASSOCIATION OF · GENETIC · SUPPORT OF · AUSTRALASIA

FUNDED BY THE NSW HEALTH DEPARTMENT

AUGUST 1994 ISSUE 13

ADVANCE NOTICE
ANNUAL GENERAL MEETING

AGSA will be holding its Annual General Meeting on Sunday 27th November 1994 at 10 am at the office in Surry Hills. This will be followed by a Christmas Celebration Lunch. Put this in your diary, it will be a day not to be missed.

MISSION STATEMENT

To facilitate support for those affected directly or indirectly by genetic conditions throughout Australasia.

Registered Charity No.C.C.27702

EDITORIAL

I would like to extend my very sincere thanks to all those AGSA members and friends who rang, following publication of the last Newsletter, offering their congratulations, encouragement and support to the incoming committee and Dianne Petrie. The hiatus AGSA experienced is behind us and with the increase in contacts, together with requests for new groups and information, we can look forward to an assured future.

CHANGES IN GENETIC SERVICES

The future is now secure for families in Queensland following the ground swell of public support and media coverage which led to the Queensland Governments commitment to Genetic Services. AGSA would like to thank those groups, families and professionals who took such a proactive, and at times, personally difficult stance, for the benefit of others.

Dr John Nelson, Clinical Geneticist, Westmead Hospital is returning to Western Australia in mid September to take up a position as Consultant Clinical Geneticist at the King Edward Memorial Hospital for Women and the Princess Margaret Hospital - New South Wales' loss is Western Australia's gain.

I believe that many families in Western Australia will be pleased to see Dr Nelson return. On a personal note I would like to express my appreciation for his support and sense of humour both to myself, the MPS Society and families affected by genetic conditions.

Professor David Sillence, Clinical

ISSN 1033-8624

Geneticist, The Royal Alexandra Hospital for Children, Sydney, heads to the Murdoch Institute, Victoria, in October, for 12 months sabbatical.

Professor Michael Partington, Clinical Geneticist, Newcastle Regional Medical Genetics Unit, retires after establishing services in the Hunter region and bringing with him years of Canadian experience to the development of Outreach Genetic Services in NSW.

Professor Partington will be replaced by Associate Professor Gillian Turner, Clinical Geneticist, from Prince of Wales Children's Hospital, Sydney.

It is our hope that future Newsletters will contain relevant information pertaining to Genetic Services throughout Australasia.

We invite you to read this Newsletter with care as it contains a new column, that of our Support/Information Officer, in which Dianne provides some insight into the diversity of her role.

Please remember this is your Newsletter and we would appreciate knowing of events, change of address and other relevant activities which would benefit from a wide distribution.

Ros Smith

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SUPPORT/ INFORMATION OFFICER REPORT

I was delighted to accept the position and commence duties on 21st March operating out of a small office located in the old Children's Court in Surry Hills. I share the office with WIFT (Women in Film and Television) which always makes for an interesting time.

The last four months for AGSA has been very positive and productive. Enquiries for information, support and/or contact with another family have increased and I would like to thank genetic services and the medical professionals who have fielded my enquiries willingly.

On the 10th June a coffee morning was held for families whose children have Sotos Syndrome and as a result of this meeting a request has been made for a seminar on Sotos and the establishment of a Sotos support group. What started with one family has expanded now to ten families.

On 26th June five families attended a Leigh Disease Seminar and as a consequence there is now a Leigh Disease Support Group. We have received enquiries from the UK and NZ about this group.

On 7th August 30 people, representing 15 families attended the first Klinefelters Meeting. The day was a great success and the participants decided to meet again on 6th November.

On 14th August 12 people, representing 4 families attended the first Rare Chromosomal Disorders Support Group Meeting. An additional 4 families sent their apologies. Another meeting is planned for October.

Don't forget the upcoming Seminar on Support Groups - How to Start One and Keep it Going, to be held on Sunday 11th September at a cost of $6.00. I would like to invite a representative from each member support group of AGSA to attend this meeting. There will be a panel of speakers covering all aspects of

starting and running a group, such as the constitution, tax laws, new charity rules, newsletter production, privacy laws etc. This is a chance to bring your pamphlets and latest information along to up-date our files and to exchange ideas about what's worked for your group and maybe what has not worked. It is intended that the afternoon be reserved for such a forum.

The response, so far, for these meetings has been excellent and I look forward to meeting more families. A bonus of my work. I always enjoyed the AGSA get togethers, hearing what other people were doing and making new friends.

It is the aim of AGSA to hold seminars approximately three times a year, the Support Group meeting being the first. Proposed subjects for future seminars are "Living Grief" and "Behaviour" to be held in early 1995.

Since March AGSA has, in response to requests, facilitated contact between a number of families who sought to communicate with similarly affected families.

If you have not rung the AGSA office to introduce yourself and your group please do so. I would like to hear from you. Don't forget you are able to use the three meeting rooms at 66 Albion Street free of charge, all you need to do is to book them through AGSA. I look forward to either talking to you or meeting with you in the future.

Dianne Petrie

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CONFERENCES

UNIVERSITY OF NEW SOUTH WALES SCHOOL OF TEACHER EDUCATION

SPECIAL EDUCATION SEMINAR NO.4

Date: 13 September 1994
Time: 4.30 pm - 6.30 p.m.
Venue: Clive Monk Theatre

The University of New South Wales,
St. George Campus
Cnr Hurstville Rd & Oatley Ave
OATLEY

Enquiries: Mrs Amy Kolaric
PO Box 88
OATLEY NSW 2223
ph: (02) 570 0919
fax:(02) 570 064

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ARTHROGRYPOSIS

The Australian Arthrogryposis Group (TAAG) Inc is holding its Annual General Meeting on **Saturday 17th September, 1994** at The Royal Alexandra Hospital for Children. Guest speakers will be Dr Jennifer Ault, a Specialist in Rehabilitation Medicine and Ms Wendy Hawker, Physiotherapist.

For more information contact:
Mrs Marie Taylor (02) 938 4726

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TURNER SYNDROME

Turner Syndrome National Conference
30th September 1994 (Fri-Sun)
Santa Sophia College,
University of Sydney.

For more information contact:
Mrs Glenn Fisher
P.O. Box 112
FRENCHS FOREST NSW 2086
(02) 452 4196

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KLINEFELTER SYNDROME AND ASSOCCIATES NATIONAL CONFERENCE

October 1 - 2, 1994
Anaheim, CALIFORNIA

Contact: KS and Associates
P.O. Box 119
Roseville, CA 95678-0119
USA

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GENES IN PRIMARY CARE - WHAT YOU REALLY NEED TO KNOW

October 3 - 5 1994
Cambridge, MA
Co-sponsored by the Cambridge Hospital, Havard Medical School and MIT.

Contact: Alison Harris
The Cambridge Hospital
1493 Cambridge Street
CAMBRIDGE, MA 02139
Tel: 0011 1 617 498 1584

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AUSTRALIAN ASSOCIATION FOR THE WELFARE OF CHILD HEALTH CONFERENCE

Celebrating 21 years of working for children and families in the health care system.

"Facing the Challenge"

Thursday 6 and Friday 7 October 1994.
University of Western Sydney,
158-160 Hawkesbury Road, Westmead
Health Studies Building
P Lecture Theatre.

For details contact:
AWCH National Office
PO Box 113, Westmead, NSW 2145

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FABRY'S SUPPORT GROUP MEETING

Saturday 5th November 1994 1.30 pm

Murdoch Institute
10th Floor
Royal Children's Hospital
Flemington Road
PARKVILLE VICTORIA

For Information Contact:
Margaret Davie (03) 762 3910

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8TH INTERNATIONAL CONFERENCE ON SPINA BIFIDA AND HYDROCEPHALUS

In conjunction with the New South Wales Society for Children and Young Adults with Physical Disabilities.

14th -16th September 1995
The Collaroy Centre, COLLAROY

Information can be obtained from:

S.B.H. Conference Secretariat
GPO Box 128 Sydney NSW 2001
Tel: (02) 262 2277
Fax: (02) 262 2323

Professionals wishing to present a paper or run a workshop should make submissions by 30th September 1994.

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PRELIMINARY ANNOUNCEMENT

MPS 4th International Symposium on the Mucopolysaccharidoses and Related Diseases
Wollongong May 1996

The theme of the Wollongong meeting will be "Genotype to Phenotype and Therapy". Latest developments in clinical management and treatment of patients will be presented.

International experts from various countries will be presenting at this meeting. The success of previous meetings held in Minneapolis, Manchester and Essen, in which families, support group personnel, clinicians and scientists meet and interact, will be maintained. "This unique format has provided an excellent opportunity to share information and experiences of mutual benefit to all delegates."

For further information contact:
The Conference Secretariat
6 Azalea Place
LOFTUS NSW 2232
(02) 521 6785

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GENETICIST SAYS THANKYOU

Mrs R Smith
President
Association of Genetic Support
of Australasia (AGSA)
66 Albion Street
SURRY HILLS NSW 2010

Dear Ros,

I would like to thank you and members of AGSA who rallied to support the need for genetic services for Queensland. I am well aware of the courage it required for a parent to expose his or her child with special needs to the public via the media and I am convinced that it was such actions which convinced Queensland Health of the urgency for a genetics service in that State. I remain hopeful that now that adequate funds and a committed plan are present, that Queensland may be able to attract the appropriate staff to form a proper genetics unit.

Kindest regards,
Dr Jim McGill
Clinical Geneticist
Royal Children's Hospital, Brisbane

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CAN I HELP ?

Support Groups meet many of your needs. However, if you want to talk things through with a psychologist/ genetic counsellor with wide ranging hospital experience, I am now available for private consultations. Individuals or families welcome. For further information please contact:

Bronwyn Butler
(02) 419 6782

CONTACT CORNER

AGSA will publish requests for contact and letters from people searching for families with similar experiences, from those seeking or contributing specific information as well as other resource information.

Anyone who wishes to reply to a request or a letter should write direct to the individual or group concerned where an address is provided. The AGSA office may be contacted for the information to be passed on in the case of anonymous requests. Privacy and anonymity will be ensured if requested.

While AGSA aims to facilitate contacts between families it is unable to assess the suitability of these in individual cases.

It should be remembered that a shared genetic condition does not mean an equally shared value system between families. Different degrees of acceptance and different mechanisms for coping will be encountered and a non-judgmental approach is recommended in establishing contact.

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WERDNIG-HOFFMAN DISEASE

Carolyn Gooch offers peer support to families with a child suffering from Werdnig-Hoffman Disease. She is interested in contacting other families who would like to offer support in other states. If you are interested please contact:
P.O. Box 60
North Beach WA 6020
Telephone: (09) 401 9652

1. **CHORIODEMIA**
2. **INCONTINENTIA PIGMENTOSA NEVUS OF ITO.**

Two families in Western Australia are seeking contact with another family who may have a child with either of these conditions. Please contact AGSA or
The Kalparrin Centre
Children's Hospital Medical Centre,
Roberts Road,
Subiaco WA 6008
Telephone: (09) 340 8094

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COHEN SYNDROME

Lorraine Temple
33 Grieve Street
MACLEOD VIC 3085
is seeking contact with another family with this condition.

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OSTEOPETROSIS

Carl and Kim Allen's son Chad is now 6 months old and they have made contact with two other families who have a child with this condition. The Allens would like to organise a meeting for later in the year and to hopefully make contact with other families. For information please contact the AGSA office.

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TAR SYNDROME

Contact with another family has been requested - phone AGSA for details.

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MYOTUBULAR MYOPATHY

Judy is seeking contact with another family for her nine year old son.

Judy Yaxley
22 Huntington Drive
Kallangun Qld 4503

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HEREDITARY FRUCTOSE INTOLERANCE

Lydia came to Australia from Germany two years ago and would like contact with another family with the same condition for her son aged 10.
Please contact:-
Lydia Lautenfschlager
17 Duignan Close
Epping NSW 2121
Phone: 868-4463

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BECKWITH-WIEDEMAN

A family in Western Australia is seeking contact with another family. For details contact the AGSA office.

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BEHR'S SYNDROME

Behr's Syndrome is a slowly progressive neurodegenertive disease and a mother of a 17 year old girl has requested contact with another family. Please contact the AGSA office if you can help.

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METHYLMALONIC ACADEMIA

The parents of a 12 year old girl seek contact with another family. Please contact the AGSA office if you can be of assistance.

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WOLF-PARKINSON-WHITE SYNDROME

Family seeking contact, please phone the AGSA office.

SUPPORT GROUPS

FABRY'S DISEASE

In April, 70 people met in Melbourne and formed a Fabry's Support Group. Anyone wishing to be a part of a New South Wales branch of the Fabry's Support Group please contact AGSA.

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LYMPHODEMA

Kerry is currently in contact with six families who hold regular meetings. Kerry would like to established a *support group* and is interested in hearing from other families.

Contact: Kerry McCann
60 Alfred Street
MASCOT NSW 2020
ph: (02) 693 1241

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MULTIPLE EPIPHYSEAL DYSPLASIA SUPPORT GROUP

We have been notified by Jennie Daws President of the above group that support is also available to people with **Spondylo Epiphyseal Dysplasia, Perthes Disease**

and associated conditions. Jennie would be pleased to hear from you or any interested parties.
Contact:
Mrs Jennie Daws
President
Multiple Epiphyseal Dysplasia Support Group,
6 Vivienne Court,
Doncaster East Vic 3109
or contact the AGSA office.

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CONGENITAL ANODONTIA

AGSA has been contacted by a lady from Victoria who would like to set up a *support group* for the above condition. If you are interested please contact the AGSA office for more details.

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KAWASAKI SYNDROME SUPPORT GROUP

Mrs Sue Davison, Coordinator of the K.S.S.G in England, would like to hear from any parents who have a child with Kawasaki Syndrome with the view to sharing experiences and information on treatment.

For Contact:
Mrs Sue Davidson
National Coordinator
Kawasaki Syndrome Support Group
Potters Green
Coventry CV2 2FR
England

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SOTOS SYNDROME

AGSA has been contacted by a number of families who wish to set up a *support group*. Anyone interested please contact the AGSA office.

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SUPPORT GROUP UPDATE

AUSTRALIAN SOCIETY FOR ECTODERMAL DYSPLASIA

15 A'Beckett Avenue ASHFIELD 2131

Secretary - Betty Kozanecki
ph: (02) 799 9783

President - Phil Masters
ph: (045) 711 621

fax:(045) 711 526

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AGSA COMMITTEE UPDATE

Due to the absence of Deborah Redelman at the time of production of the last Newsletter we would like to present a more accurate profile.

Deborah Redelman - Committee Member

Debbie has been the State Co-ordinator of the Tourette Syndrome Association since early 1993. Debbie has a Bachelor of Science with Honours in Human Genetics and Psychology. She was a Research Assistant in the National Perinatal Statistics Unit in 1986-7, which collects and analyses data on birth abnormalities. She is married with three healthy young children.

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PROFILE A - Z GENETIC CONDITIONS

It is the intention of AGSA to profile, in each issue, a particular Support Group/Disorder, thus increasing awareness within our membership of the range of genetic conditions. Also it hopes that where overlaps occur in conditions, Support Groups may liaise with each other and thus gain a broader understanding of facilities, aids, etc. that may be of value to your individual membership.

Please ensure that all support group information is recent and reliable. It is of paramount importance that you let us know your group is 'Alive and Well' and happy to take referrals.

For your information we profile.....

LEIGH DISEASE

kindly edited by Dr John Christodoulou, Biochemical Geneticist, Royal Alexandra Hospital for Children, Camperdown.

Symptoms

Leigh Disease is caused by genetic defects involving the metabolic pathways of energy (ATP) production. It is characterised by damage to the brain, spinal cord, optic nerve and in some cases, an enlarged heart or abnormalities of kidney function. The disorder is usually first diagnosed during infancy but may begin later.

The clinical features of Leigh Disease are:

- onset usually in infancy or early childhood
- progressive neurological deterioration
- motor delay
- intellectual disability
- respiratory abnormalities
- visual abnormalities
- loss of coordination (ataxia)
- involuntary movements
- slurring of speech
- epilepsy
- slow growth, poor weight gain
- characteristic brain pathology

This disorder can be very variable and not all characteristics of the disorder may be present in any one affected individual.

Incidence

80% of known cases of Leigh Disease affect infants. The remaining 20% show symptoms as late as adulthood. Both sexes are affected by Leigh Disease.

Diagnosis

There are three levels of investigations used in the diagnosis of Leigh Disease using the following tests:-

i) - blood & CSF lactate/pyruvate/ alanine
   - CT scan/MRI of brain

ii) - muscle biopsy (and sometimes liver) for pathology and biochemical studies

iii) - DNA testing

Treatment

The treatment of Leigh Disease is almost always very disappointing.

Attempts have been made with vitamin

cocktails such as vitamin C, K, riboflavin and thiamine. In addition, coenzyme Q and dichloracetate have been tried.

Inheritance of Leigh Disease

There are three known mechanisms for the inheritance of Leigh Disease:-

1. Autosomal recessive
   - 1 in 4 risk
     (boys risk equal to girls)

2. X-linked
   - 1 in 4 risk
     (usually boys only although girls may be more mildly affected)

3. Mitochondrial
   - risk up to 100% (boys or girls)

For more information on Leigh Disease, please contact:

The Leigh Disease Support Group
c/- Mrs Robyn Moody
14 Argyle Street
PENSHURST NSW 2222

National Leigh Disease Foundation Inc.
613 Childs St
CORINTH MS 38834-4810 USA
(6010) 287-8069

Lactic Acidosis Support Group
P.O. Box 480282
DENVER CO 80248 USA
(303) 287-4953

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FAMILY STORY

LEIGH'S DISEASE - "A Mothers' Story"

My name is Robyn and I would like to tell you about my children and their family. I have been married for 12 years to my husband Trevor and have been blessed with three children, Cameron , 9 years, Byron, 6 years and Erin, 2 years.

One year before my own birth, my parents suffered the loss of their second son, Kenneth, at the ageof 2 years 4 months, to what they were le dto believe was Pneumonia. That was in February 1959. Little did they realise, that his passing would later aid in the formulation of a diagnosis that would affect the very soul of our family.

My first pregnancy (Cameron) was uneventful followed by a natural birth. During his first twelve months he reached his normal milestones climaxing with his first steps taken just after his first birthday. It was about this time that he became unwell with a "virus", that I noticed he had difficulty in maintaining his energy with his walking, he went back to crawling for a number of months, also he wasn't as active as others youngsters of his age, i.e. climbing etc. From then on his development was perceived to be "normal" until he was approximately 3 years of age when I became concerned about his language, he hada noticeable stutter and unable to string correct words together.

It was at this time he was assessed by several clinicians, with one diagnosis being Minimal Cerebral Dysfunction,

with no gross abnormal neurological signs. By the time Cameron reached 7 years, he had a tendency to pick up colds which would develop into asthma, viruses, very easilly, which at every time would exacerbate symptoms such as, falling down, ungainly gait, stuttering, dribbling and lethargy. Two months following his seventh birthday Cameron contracted another cold which layed him very low, necessitating admission to hospital. This was symptomised by his inability to work, loss of reflexes and general run down condition, eg virtually unconscious at the time of admission. Following several tests and investigations, my husband and I were asked by the Consultant Neurologist for information on family medical history, illnesses or maybe unexplained deaths etc. It was then that I related to him the story of my brother Kenny. It was by chance that the doctor asked if there was an autopsy performed and if so, where? Fortunately for us all, it was carried out at the same Children's Hospital that Cameron was admitted to. Resulting in his search through hospital records we were informed that Kenny's death, in fact was due to Leigh's Disease. After a few more tests, Cameron was also diagnosed with Leigh's.

My second pregnancy (Byron) was eventful requiring my hospitalisation for the last trimester. He was delivered by Caesarian section at 36.5 weeks. He was placed in the Neonatal Intensive Care Ward due to immature lungs. His development was similar to that of Cameron's , although not as noticeable until the age of approximately 4 years. It was at this time that he was admittde to the Children's Hospital for a kidney infection. I then noticed Byron was having problems maintaining his balance whilst walking, eg falling over constantly with no automatic reflex response. Thus followed the same testing as Cameron and Kenny; Leigh's Disease.

After continual physiotherapy and occupational therapy programs, the boys attend our local state school. Naturally they experience some difficulties, eg keeping up with their classmates, both in the classroom and playground, however they are presently coping with the aid of a provided integration teacher. Both my husband and I believe that they maybe aware that they are slightly different to other children, however it can be said with pride, that they have not lost their sense of humour, their love of sport. They both play in the local soccer competition, and have received encouragement and sportsmanship awards.

At this time, after the devastation of this news had settled, my husband, myself and our immediate family naturally reamin hopeful for a cure, however we take one day at a time and relsih in teh wonderful light that these two boys have brought into our lives.

P.S. Erin our daughter and third child, survived a normal pregnancy and birth, but due to the above circumstances concerning bothe Cameron and Byron, she has also been tested for Leigh's. We are still waiting the results.....

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RESOURCES AND AIDS

CARER CLOTHING PTY LTD.

Katrina Alfred is a successful Clothing Designer who, for some years, has been looking after a husband with MS. Finding no availability of suitable clothing, and after discussions with other Carers, she has developed a range of garments which goes a long way towards meeting the needs of both Carers and their Charges.

No Carer wishes to cause pain or discomfort. A prime design target of the CARER CLOTHING range is that the garments can be fitted or removed with a minimum of discomfort for the wearer who may, for example, be in a wheel chair, or unable to lift their arms above shoulder level. As another example, pants styles are available which allow fitting without the need to remove shoes. If your would like to view the Carer Clothing Range contact:

Peter Fitzpatrick,
Sales Manager,
Shop 8,
25 Hudson Parade,
Clareville NSW 2107
Phone: (02) 973 1400
Fax: (02) 973 1498

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WHAT'S LIFE WORTH?

LIFE+CARD is a credit card sized, laminated, patented microfilm. Simply hold the card up to any light, day or night, and all your personal medical data and details are instantly readable with the naked eye by doctors, ambulance and emergency personnel - all 34 questions enabling immediate treatment with a minimum of time loss. Comprehensive protection - 24 hours a day, 7 days a week. Ambulance and Hospital staff are being advised to look for the LIFE+CARD on all emergency patients prior to examination. Simply carry LIFE+CARD with you and avoid delays in treatment that may cause loss of life.

Contact:
LIFE+CARD
P.O. Box 437
Pymble NSW 2073
Phone: (02) 867 5638
FAX: (02) 983 1232

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FARM COTTAGE WITH DISABILITY ACCESS

Kabungarra, at Freshwater Creek near Geelong in Victoria, is perfect for families with a disabled child looking for a holiday.

Alison and Charlie Corke have designed and built a cottage where children with disabilities can experience all the fun of the farm.

They have built an all-terrain rickshaw which provides access to all parts of the farm.

Brochures are available from the AGSA office or by contacting the farm direct either by writing to:

Kabungarra
275 Bogans Lane
FRESHWATER CREEK VIC 3216

or phoning Alison on: (052) 64 5280.