

# GATEWAY

Volume 10 No 3

Australian Huntington's Disease Association (NSW) Inc

Spring 2007



Australian Huntington's Disease Association (NSW) Inc

## **2007 ANNUAL GENERAL MEETING**

**FRIDAY 23 NOVEMBER 2007**

**7.00PM — 8.30PM**

**21 Chatham Road, West Ryde**

**Refreshments to be served at 7pm**

Non members welcome

**Guest Speaker: Nicolas Patrick**

**National Pro Bono Director, DLA Phillips Fox**

Nicholas will speak about legal issues faced by people with HD  
and how his firm can assist.

Please RSVP (for catering purposes only). Phone 02 9874 9777 or 1800 244 735  
(NSW STD Freecall) or email [maria@ahdansw.asn.au](mailto:maria@ahdansw.asn.au). Please note it is possible to  
attend even if you haven't given an RSVP.

### **AGENDA**

- Apologies
- Minutes of Previous AGM
- Business Arising from Minutes
- Special Guest Speaker
- Annual Report
- Annual Financial Report
- Annual Statement
- Appointment of Auditor
- Election of Office Bearers and Committee Members
- General Business

**Note:** You must be a financial member to be able to vote.

Nomination forms for the Management Committee are enclosed with this newsletter.  
Completed forms should be returned to AHDA(NSW) by Friday 16 November 2007.

# From The Executive Officer

As you would be aware, in 1999, the Association signed a ten year lease for Camellia Cottage at Mayfield, near Newcastle. Its primary aim was "to provide people with accommodation in the community, that meets their needs for supported accommodation and alleviate the isolation that people with chronic disease experience." It was to act as transitional accommodation between community and institutional living, and was not intended to act as an alternative to institutional living.

The property is leased from the NSW Office of Community Housing at a nominal rent and managed by Compass Housing Services. It can accommodate four residents, although occupancy has never been greater than three residents, and sometimes none.

Residents of Camellia Cottage pay rent based on community housing standards but receive additional financial benefits. The Association pays for:

- Gas, electricity and water charges as individual consumption aren't metered.
- Cleaning of common areas and individual bedrooms/bathrooms.
- Grounds maintenance & furnishings for common areas.
- Other ad hoc expenses that would normally be a tenant responsibility, such as extra cleaning, individual smoke alarms

The original intent was that the Cottage would be self-funding, including the cost of restoring the property to an appropriate standard at the end of the lease. Insofar as the Association has provided a financial subsidy to residents, this is an unintended consequence and one that represents an inequity in respect of other potential beneficiaries of the Association's support.

In June 2007, the Association's Management Committee decided to close Camellia Cottage, and surrender its lease of the property. This decision was based on a combination of issues that included:

- Costly maintenance & high utility bills
- Multiple home invasions
- The apparent isolation of tenants, and
- Low occupancy rates

The closure will not happen immediately, as it is a requirement of the lease that six months notice must be given.

I attended a meeting in Newcastle in September, with the Office of Community Housing, Hunter Genetics and Compass Housing Services, to discuss issues relating to the surrender of the lease, the handover of the property, and alternative accommodation options.

Please be assured that we are intent on finding an alternative accommodation solution for this group of residents, and anticipate knowing the outcome of the meeting later this month — we will provide a further update in the next issue of Gateway.

In friendship,

*Jacqueline-Marie Bohm*

# Huntington Happenings

## Holiday Camp Improvements

In the last issue of Gateway we reported on the May holiday camp for residents of Lottie Stewart Hospital. Holiday camps were one of the earliest initiatives of the Association and continue to be a worthwhile use of the Association's resources.

Those with long memories will recall that camps held at "Gilbulla" at Menangle were fully catered for by Gilbulla staff. More recently we have been using Camp Breakaway, which has not been prepared to cater for the smaller numbers we usually have attending. This has been problematic for recent camps – for the last three we have successively used volunteers, a privately-employed cook and a member of staff. For this reason, as well as other staffing issues, we have postponed the camp that was to be held this September. This camp will now be held between the 1<sup>st</sup> and 4<sup>th</sup> of April 2008.

We are pleased to advise however, that following a change of management, Camp Breakaway has now agreed to cater for numbers typical of our camps. As this is an excellent venue in all other respects, this is a very welcome development. In future, we will be working towards making the camps more accessible to those living outside the Sydney – Newcastle area.



## September City Holiday

During September, in place of the September Camp, we held a memorable "short city break," with attendees enjoying a visit to the Sydney Aquarium and Darling Harbour on one day, and lunch at a waterfront restaurant in Drummoyne on the second day.

## Donation Acknowledgements

The Association gratefully acknowledges:

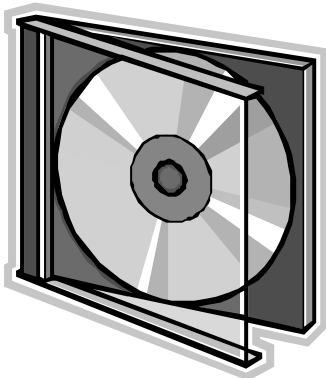
- The Macquarie Bank Foundation, for its generous gift of \$7,000 in June 2007.
- Aon Charitable Foundation (and the Association member whose initiative this was) for a donation of \$500 in July 2007.
- Kelgrove Holdings Pty Ltd (trading as Multi Direct) for financial support to AHDA National to enable the National Chairperson, Ann Jones, to attend the World Congress on Huntington's Disease in Germany in September 2007.

# Huntington Happenings

## 2006 Conference DVD Available

A DVD of presentations from the 2006 AHDA National Conference is now available from AHDA (Vic) for \$25 per copy (plus \$5 postage/handling).

If you would like a copy of the DVD, please ring or email the Association and we'll send you an order form.



## Thank You Members!

A big "thank you" to the 175 members who have applied for, or renewed their membership for 2007/08. Our total membership at the end of August was 321, which means that there are 146 members who have yet to renew.

Maintaining a healthy membership gives us strength when advocating for Huntington's families – if you have not yet renewed, please consider doing so. If you are not sure of your membership status, please ring us on (02) 9874 9777 (or 1800 244 735 from country NSW) and we will advise you.

We are also most grateful to those members who accompanied their membership fee with a donation.

There has also been a gratifying response to the option provided for members to identify their particular status as a person affected in some way by Huntington's Disease. We now have a much better idea of how our membership is constituted, and also have the ability to target mail-outs to particular interest groups.

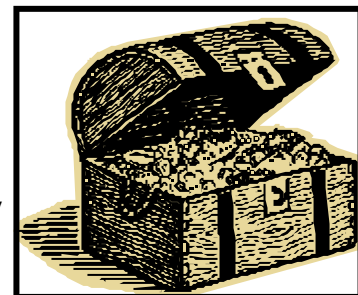
## Community Chest Results

The Mayor of Burwood John Faker, drew the winning tickets on 18th July 2007 at Burwood RSL Club.

The winners are:

- First Prize of a Toyota Corolla: G. Oborn of Morpeth
- Second Prize of \$3,000 jewellery: O. Nidai of The Oaks

Congratulations to the winners, and our thanks to all who helped by purchasing or selling tickets.



# Juvenile HD Research

## Psychiatric and Cognitive Difficulties as Indicators of Juvenile Huntington Disease

HD Lighthouse Contributing Editor's Comment: This retrospective review of the case histories of 29 JHD patients is a very valuable addition to the juvenile Huntington's Disease literature. Out of 1452 HD patients seen over a fifteen year period at the Saltpetriere Hospital in Paris, 2 percent were JHD patients. Their case histories were reviewed for this report.

The average age of onset in the study was 14. The average delay in getting a diagnosis was 9 years, with a range of 0 to 21 years. The delay was caused by the 'nonspecific' features of onset and in some cases, an absence of family history. By nonspecific, they mean symptoms that are not necessarily indicative of JHD but could be caused by other diseases or problems. Traditionally, Huntington's Disease has been diagnosed by characteristic motor symptoms although cognitive and psychiatric symptoms may have been already present.

Ten children first presented with motor disturbances, nine with psychiatric problems, and ten with cognitive decline. In six of those, the decline in school performance was rapid.

Of those presenting with motor symptoms, none started with rigidity although 21 patients had dystonia at some point and 18 had chorea. The first signs in those presenting with motor symptoms were chorea (3 children), myoclonus (involuntary twitching affecting 3 children), falling (2), handwriting difficulties (1), and twitching of the shoulders (1).

**"The average age of onset in the study was 14. The average delay in getting a diagnosis was 9 years, with a range of 0 to 21 years. The delay was caused by the 'nonspecific' features of onset and in some cases, an absence of family history. "**

Of those presenting with psychiatric symptoms, three had serious depression, three had substance abuse problems, one had fugue states (doing things without being cognitively aware) and suicide attempts, one was psychotic, and one had a number of behavioral changes. In the group overall, psychiatric problems were serious with seven children attempting suicide.

Although maternal transmission of JHD is usually referred to as rare, one-quarter of the patients had an affected mother. Although the literature usually states that JHD is associated with CAG counts of 60 or higher, nearly half of the patients in the study had between 45 and 58 repeats.

Clearly then, in JHD as with adult HD, there is a substantial variation in onset and clinical progression of the disease. Individuals don't necessarily follow the recognized pattern of symptoms for JHD - motor onset, known family history, paternal transmission, and high CAG repeats.

Biglan and Shoulson provided commentary in the same edition of the journal. They recognize the variability of early symptoms of those with HD and point out that the problem of determining onset is shared with adult Huntington's Disease as well. "Onset of HD in juveniles and adults, while a discrete outcome of intrinsic value to families and care providers, remains a probabilistic judgment that has yet to be validated prospectively."

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# Research News

Continued from Page 5

With therapies on the horizon, getting data from longitudinal, prospective studies is critical. They describe two ongoing studies for adults, PHAROS and Predict-HD, as well as the newest study COHORT which also includes children.

## References

Kevin Biglan and Ira Shoulson, "Juvenile-Onset Huntington Disease: A Matter of Perspective," Archives of Neurology 2007;64:783-784.

-- Marsha L. Miller, Ph.D.

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## The Molecular Zip Code Research Yields a Drug Target

HD Lighthouse Contributing Editor's Comment:

This is breakthrough research!

The HD Lighthouse has been following the molecular zipcode research (see <http://www.hdlighthouse.org/research/brain/updates/1365protein.php> ) so it is exciting to see how it has been advancing, yielding a new drug target for an upstream pathology as well as new insights into the normal function of the Huntingtin protein.

The research team of Dr. Ray Truant has found that the Huntingtin protein is associated with the endoplasmic reticulum (ER). The ER is a structure within the neuron (and other cells too) where proteins and lipids are synthesized and processed for use inside and outside the cells. It's also where protein quality control is maintained. If a protein isn't folding right, then the ER will try to refold it properly so it can be used or degrade it so it won't do any damage.

The Huntingtin protein associates with the ER through its first 18 amino terminals. They reversibly target the ER as well as vesicles in the cell and the process is dependent on ATP, cellular energy.

They also found that the normal function of the Huntingtin protein is to respond to ER stress by communicating with the nucleus, the control center of the cell. The Huntingtin protein shuttles back and forth between the ER and the cell nucleus. Previously, it was thought that the normal Huntingtin protein was a cytoplasmic protein which did not go into the nucleus of the cell, but through the use of their live cell model technology, the researchers could see that it does. It's just that it enters and exits very quickly.

However, the HD version of the protein goes into the nucleus of the cell and has trouble exiting. It builds up and is toxic to the cell.

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# Research News

The association of the Huntingtin protein with the ER sheds some light on some earlier research results. A team of Japanese researchers found that using RNA interference to knock down the normal mouse Huntingtin protein led to malformation of the ER -- but not other organelles and structures in the cell.

The good news for HD families is in the press release and not in the article itself. First, there is a kinase (an enzyme which works by transferring phosphate groups from high energy donors such as ATP) which directs the Huntingtin protein into the nucleus of the cell. Keeping the HD protein out of the nucleus would save the cell. They are working on developing a drug to inhibit this kinase and have already determined the structure of the kinase, a necessary step in the process.

Second, even if the kinase inhibitor does not prove to be a treatment, other drugs can be tested in this live cell system and the researchers will be able to tell what they are doing in the cell in real time.

Basic research into HD pathology has been following several valuable lines of investigation and plans for drug development and clinical trials focus on different HD pathologies. Because so much has been going on, it can be hard for HD families to see just how research findings have been building on each other and how much progress has been made toward understanding and treating the disease.

If you visit the Huntington Society of Canada's website through the link below, you can see Dr. Truant's original proposal in 2001 to investigate this line of research. <http://www.huntingtonsociety.ca/english/truant.htm>

You can follow Dr. Truant's team's progress through research articles by doing a search on Pub Med. Go here <http://www.ncbi.nlm.nih.gov/sites/entrez?db=PubMed> and search under 'Huntington's Disease Truant R'. Funding for the research is now paying off in terms of understanding the function of the Huntingtin protein and suggesting a promising new drug target.

Whether or not a kinase inhibitor proves to be a treatment, this study represents a major advance in our knowledge of the disease and will certainly contribute to the development of a drug to treat Huntington's Disease.

## References

Omi K, Hachiya N, Tokunaga K, and Kaneko K, "siRNA-mediated inhibition of endogenous Huntington disease gene expression induces an aberrant configuration of the ER network in vitro." *Biochemical and Biophysical Research Communications* 2005 December;338(2):1229-1235

Wu JC, Liang ZQ, and Quin ZH, "Quality control system of the endoplasmic reticulum and related diseases." *Acta biochimica et biophysica Sinica* (Shanghai) 2006 Apr;38(4):219-26.

-- Marsha L. Miller, Ph.D.

Posted to the HDL: 21 Aug 2007

Source: Human Molecular Genetics Advance Access published on August 18, 2007

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# Pro Bono Legal Services

In October 2006, in partnership with the Association, DLA Phillips Fox launched a pro bono legal service for people with Huntington's Disease and their families.

This Partnership allows the Association to refer directly to DLA Phillips Fox, people who require legal assistance. The assistance is not subject to a means test, and provides direct access to a lawyer without having to satisfy the criteria of traditional pro bono legal services.

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**"Since the inception of the Partnership, the Association has made a number of referrals to DLA Phillips Fox, with positive outcomes."**

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DLA Phillips Fox is one of the largest law firms in Australia and this assistance is provided under their National Pro Bono Program. They have an extensive national network with offices in Sydney, Canberra, Adelaide, Brisbane, Melbourne and Perth.

If there are instances where DLA Phillips Fox is unable to assist due to lack of expertise or resources, they will liaise with other firms doing pro bono work in an attempt to find a firm that can assist.

Since the inception of the Partnership, the Association has made a number of referrals to DLA Phillips Fox, with positive outcomes, including:

- The provision of a barrister at late notice, for a member with a court hearing in Western Sydney.
- Referral to a regional firm, for a Sydney based member with an upcoming regional court hearing.

The commitment, empathy, resourcefulness and professionalism demonstrated by DLA Phillips Fox, continues to impress the AHDANSW team, and members can be assured of a speedy, caring response to their requests for legal assistance under this partnership.

Members can access this resource by contacting the Association directly and simply requesting a referral. You will not need to divulge the details of your legal matter to the Association - we simply arrange a letter of introduction to our partner, DLA Phillips Fox, and advise them of your impending request.





# Carer News



## Coming to a Town Near You... Carer Support Program Country Meetings

Over the next three months we are planning Carer and Family Support Meetings in the following areas.

**Central West:** Blue Mountains, Bathurst, Mudgee, Gilgandra, Dubbo, Parkes and Orange.

**South West:** Cowra, West Wyalong, Hay, Griffith, Narrandera, Wagga Wagga, Yass and Goulburn.

**North West:** Merriwa, Gunnedah, Coonabarabran, Narrabri, Moree, Glenn Innes, Armidale, Tamworth and Singleton.

We want to meet as many of our Carers as possible and make sure the voices of our country Carers and Families are heard.

Please contact us on Freecall 1800 244 735 (country NSW only), if you would like to meet up with other Carers and local service providers, or simply have a "cuppa" with Maria Mackell, the Carer Support worker.

## Carers Week

Did you know that National Carers Week takes place from 14-20 October 2007? Carer's Week is **the** national week for Carers - and a time to celebrate the amazing job Carers do every day of the year!

This year's theme 'Anyone, Anytime' recognises that becoming a Carer is often unexpected, and can happen to anyone at any stage of life. Many councils and community organisations will have activities or events, so keep an eye on your local paper around this time.

AHDANSW Carer Support Program is planning a mix of events in October. We realize many people who work can't attend weekday events, so we will also be including some weekend and evening events.

Please contact us if you are interested in attending any of the following:

- Oral History Project Launch
- Carer's Week Morning Tea Party
- HD Family Carer's Garden Club Day
- Carer's Photo Voice Exhibition



## Family Relationships Service For Carers

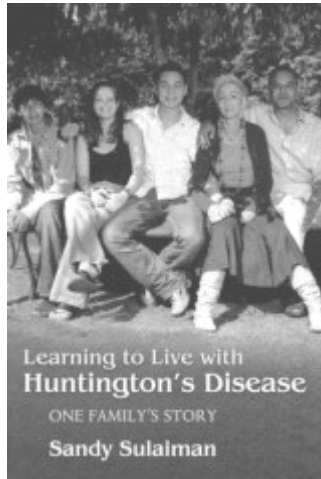


Supporting a person with disability or chronic illness can require families to make complex and difficult decisions. Achieving agreement between family members on outcomes that are in the interests of the whole family can be difficult.

A Family Relationship's Service for Carers, funded by FaCSIA, has been established in each State and Territory to assist parents and other family members to reach agreement on important issues.

Call the Association, or Interrelate Family Centres on (02) 9745 5544.

# From One Carer To Another



## Book Review: "Learning to live with Huntington's Disease-One Family's Story"

Written by Sandy Sulaiman, published by Jessica Kingsley Publishers, London, 2007.

This book is inspiring and uplifting.

Sandy Sulaiman was diagnosed with HD in 1999, and the book includes her story as well as that of members of her family.

The endorsements on the back cover from Richard Branson and Jane Paulsen were glowing, but I wasn't sure how well it would "read". However, I was truly impressed and moved by the courage, humour and wisdom of the family members as each of them describe the impact the disease on their lives. The cheerful family photo on the front cover really reflects the beauty of the family - both ordinary and extraordinary at the same time.

Sandy's story is moving and courageous; the account by her husband is frank, honest but loving. But it is the sensitive and perceptive accounts from Sandy's two sons that I found most impressive. They spoke directly and openly about getting on with their own lives, while dealing with the realities of their family's battle with HD, and the possibilities ahead for them.

Each person had their say with a fresh, unaffected air revealing their journeys, ups and downs and sometimes different recollections. And they all acknowledged the invaluable role played by some dedicated family members who did not contribute. The dedicated, hardworking mother, who felt she could not contribute to the book, was clearly an important support for the whole family, across generations. The respectful acknowledgement of her by all the contributors, should warm the hearts of Carers everywhere. The book concludes with a chapter on Tips for Living with HD. All in all, a very positive and inspiring read.

Note: If you can't locate a copy at your library, we have a copy available for loan, donated to AHDA(NSW) by gleebooks.

## The Beautiful Side of Caring at Aquila Carer's Holiday House

As Carers we often feel we can't take a break from our commitments, but, we can! I recently had a short break for Carers at Hawks Nest. It was one of the few smart things I've done lately. We were pampered in every way by Maria and Karen from AHDA, as well as volunteers Karen and Steve Hall, and masseur Richard Holland. Massage, foot spas, and pedicures were all on offer. Our meals were 5-star quality including a gourmet barbeque. A short stroll from the end of the street was a beautiful unspoiled beach. We took a river cruise up the Myall River and watched the dolphins and birdlife. Steve took some of us fishing. (please don't ask about the ones that got away). There was good talk and great company. I urge all Carers to take time out and come back to their caring roles... refreshed! **June Mitchell (02 September 2007)**

# Rural and Regional Update

## **WAGGA**

### **News Flash!**

*A new HD Family Support Group has started in Wagga!*

The first meeting was held in September, and the next meeting will be in November.

Families and Carers from Wagga and the surrounding areas are invited to join this new group, and will be made very welcome at the November meeting.

For further information, please contact Lisa on 02 6926 4772.

## **ACT AND SOUTHERN NSW**

*HD Family Support Group (ACT & Southern NSW)*

A reminder for Families & Carers in the ACT and Southern NSW area — the next meeting will be held on Saturday 3rd November, 2007.

For further information, please contact Felicity on 02 6287 1552

## **MID NORTH COAST**

*HD Benefit Concert at Sherwood Winery*

10am—4pm Saturday 3rd November 2007

Artists include: Turnbull Sessions, Hickey Sisters, Jo Robinson, Woodbees, Belly Dancers and the Drumming Sheilas.

Family activities include: BBQ, Lucky door prizes, creative arts/Stalls, Face painting, Clown/fairies, games and competitions. For further information, please contact Michelle O'Brien on 02 6566 0164, or email [tickets@dayinthevines.com](mailto:tickets@dayinthevines.com). You can also visit their website at [www.dayinthevines.com](http://www.dayinthevines.com)

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## Gateway

'Gateway' is the newsletter of the Australian Huntington's Disease Association (NSW) Inc.

## AHDA (NSW) Inc

The Australian Huntington's Disease Association (NSW) Inc is a not-for-profit organisation established in 1975.

## Our Mission

The energies and resources of the Australian Huntington's Disease Association (NSW) Inc are directed towards satisfying the needs of people with or at risk for Huntington's Disease and their families in NSW and the ACT by providing and/or facilitating delivery of a range of quality services.

## Our Philosophy

People with Huntington's Disease and their families are individuals with equal value to all other members of Australian society, with the right to treatment and care by knowledgeable professionals and care givers, the right to appropriate support services and the right to have the best quality of life possible.

## Our Services

These include education and information; advocacy; counselling and referral; holiday programs; family support; rural outreach and client services.

## Committee 06/07

President: Mark Bevan  
Vice President: Anne Low  
Secretary: Keith Dingeldei  
Treasurer: Richard Bobbit  
Members: Jim Finn

## Australian Huntington's Disease Association (NSW) Inc

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Facsimile: (02) 9874 9177  
STD Free Call: 1800 244 735  
Web Site: [www.ahdansw.asn.au](http://www.ahdansw.asn.au)

## Association and Other Useful Contacts

### AHDA(NSW)

**Jacqueline Bohm**  
Executive Officer

**Robert Curran**  
Administration Officer

**Lily Ma**  
Administration Assistant

**Maria Mackell**  
Carer Support Coordinator

**Karen Bevan**  
Activities Coordinator

**Huntington Disease Service**  
**Dr Elizabeth McCusker**  
Director  
Westmead Hospital  
(02) 9845 6793 (leave message)  
Lottie Stewart Hospital  
(02) 9804 5803  
(Tuesday afternoon)

**HD Clinic Appointments**  
Outpatients Department  
Westmead Hospital  
(02) 9845 6544

**Outreach Service**  
**Suzie Docherty**  
Social Worker,  
Westmead Hospital  
(02) 9845 6699

**Virginia Munro**  
Social Worker,  
Westmead Hospital  
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### Predictive Testing

**Fiona Richards**  
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**Angela Lownie**  
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**Jeanette Moxon**  
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**Huntington's Lodge**  
Lottie Stewart Hospital  
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**Nursing Staff**  
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**Hunter HD Service**  
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Hunter Genetics,  
Western Suburbs Hospital  
(02) 4985 3100