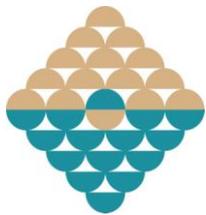


Boyne Research Institute

Annual Report January – December 2013

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Boyne Research Institute

Annual Report, 2013

Mission The joint missions of the Boyne Research Institute comprise research and education. Our research mission is to help understand the causes and consequences of diseases during childhood. Our current projects include studies into the causes of birth defects in families, and the long-term complications of cancer during childhood. Our educational mandate is to provide research experiences for young people from the community, and training for junior scientists.

Objective While retaining our present structure of independence and our community base, we are committed to becoming a world-class research institute. We aim to achieve this by expanding partnerships in Ireland, in Europe and in the United States to complement our capabilities.

Websites For more information on the research approach, newsletters, publications, presentations and results of our studies please visit www.boyneresearch.ie.

Reprints and reports of BRI studies are available from admin@boyneresearch.ie.

Julianne Byrne, CEO



From the Director

The work of the Boyne Research Institute continued to grow and change into its 21st year. We are immensely grateful to all who have provided support in many different ways during these years. We established new relationships in Ireland and Europe that will expand our research collaborations.

Our work with "Irish Families with Neural Tube Defects" saw the publication of a paper into folate-related gene variants.

Our European collaborations continue to grow with the successful application for PanCareLIFE. This consortium will investigate a second series of long-term complications of therapy for cancer during childhood and young adulthood, namely fertility, hearing impairment and quality of life. BRI serves as Research Manager for this 5-year, €6 million project, which will conclude in 2018. Our first EU-funded consortium, PanCareSurFup, now in its 3rd of 5 years, continues on track. PanCare, the pan-European network for care of children after cancer and the umbrella for these two consortia had two successful meetings in 2013. Dissemination activities for PanCareSurFup included the "Race of Brave Bikers", for European survivors, in Italy. This event will continue until 2016. Other activities involving PanCare and PanCareSurFup included a "Cruise toward Life" to raise awareness about the importance of European collaboration in the successful treatment of children and adolescents affected by cancer. More details on these activities within the newsletter:

http://www.boyneresearch.ie/resources/Newsletter_02_December_2013.pdf

I am grateful to the staff, volunteers and board members for their support and good will throughout the year. Support from the local community and from friends and foundations in the United States continues to be crucial. Many blessings on you all.



SUMMARY OF RESEARCH RESULTS FOR 2013

1. STUDIES INTO THE GENETIC ORIGINS OF IRISH FAMILIES WITH NEURAL TUBE DEFECTS

Overall Objective: to uncover evidence of inheritance patterns within families where an individual has been born with a neural tube defect (NTD). Epidemiologic signs of inheritance patterns suggestive of an underlying genetic susceptibility to NTDs could be the occurrence of birth defects of other types and adverse pregnancy outcomes (miscarriages, stillbirths and preterm deliveries). While occurrence of NTDs is more likely in families who already have an affected individual, patterns of inheritance do not conform to well-understood Mendelian patterns. It is widely thought that many genes may be acting together, or that other genetic mechanisms are involved. The emerging understanding of epigenetics may provide an explanation. This is a mechanism whereby the proteins surrounding the DNA in the nucleus are changed as a result of a changing environment or an intracellular process. Here the DNA is not changed, so we do not speak of 'mutations', which specifically refer to DNA. Epigenetic changes can be inherited with the DNA.

Results: Between 1995 and 2002, 79 Irish families with NTDs participated in this research project of the Boyne Research Institute in Drogheda. The first phase consisted of an interview with the nuclear family (parents), covering the health of the proband and family members, including a reproductive history. In subsequent phases we interviewed uncles and aunts, first cousins, the proband and siblings. In 2007, BRI carried out a biospecimen study of folate status among 325 relatives (of all types) in these families. With our colleagues from CDC Atlanta, USA, we examined 5 gene variants involved with the metabolism of folic acid in 322 members of Irish families. In line with our earlier results we found that, compared to paternal relatives, maternal relatives had a higher number of gene variants linked to lower folate metabolism. We had expected that the proportion of gene variants would be similar. But the proportions were different; maternal relatives had more of all risk variants than expected. Possibly we are looking at a situation where an epigenetic mechanism causes decreased folate metabolism resulting in increased birth defects in the maternal line. We will need to have more multi-generational studies to explore this further.

Activities in 2013: A report on the associations between molecular markers and maternal transmission was published: Green RF, Byrne J, Crider KS, Gallagher M, Koontz D, Berry RJ. Folate-related gene variants in Irish families affected by neural tube defects. *Frontiers in Genetics* (2013) 10.3389/fgene.2013.00223. This publication is available for download at:

http://boyneresearch.ie/resources/Green_Byrne_Crider_Folate%20related%20gene%20variants_Frontiers%20in%20Genetics_November%202013.pdf

2. LATE EFFECTS AFTER CHILDHOOD CANCER

Overall Objective: To carry out research studies into the long-term consequences of cancer during childhood to help survivors and their families make informed choices to improve and maintain their health.

Background: Treatment for childhood cancer has improved greatly in the last 60 years, and now approximately 80% of children will reach the 5-year survival point, most being considered cured of their original disease. However, the therapies that produced cures are toxic to healthy tissues, and can have long-term complications, especially in a growing child. This new group of patients is increasing; estimates suggest that Europe has between 300,000 and 500,000 survivors of childhood cancer. Not all require long-term care, but some do, and at different levels of surveillance. As treatments evolve and improve, the numbers of survivors in the population will also increase. Continued follow-up of existing and new cohorts of survivors is needed to provide accurate and timely information for survivors and the research community in order to prevent and remediate where possible the long-term consequences of cancer and its treatment.



2013 Activities

1. **Survival after childhood cancer in Ireland:** BRI's website is regularly updated to provide information about childhood cancer survival in Ireland and BRI's collaboration with its European counterparts into the late effects of treatment after cancer during childhood and adolescence. It provides useful links to many support groups in Ireland and Europe.
2. **PanCare** (www.pancare.eu):



Founded in Lund, Sweden, in 2007, PanCare is a multidisciplinary pan-European network of professionals, survivors and their families that aims to reduce the frequency, severity and impact of late side-effects of the treatment of children and adolescents with cancer. The long-term strategic aim of PanCare is to ensure that every European survivor of childhood and adolescent cancer receives optimal long-term care. PanCare has given rise to two pan-European consortia – PanCareSurFup and PanCareLIFE – to implement this strategy. The Boyne Research Institute is a founder member of PanCare. Dr. Byrne continues to serve as a member of the board of PanCare, and as Hon. Treasurer.

3. **PanCareSurFup (PanCare Childhood and Adolescent Cancer Survivor Care and Follow-Up Studies, www.pancaresurfup.eu):**



PanCareSurFup is a €6 million EU-funded, 5 year project that started in 2011 and will conclude at the end of January 2016. PanCareSurFup evaluates the risks from cancer treatment on the long-term health of cured survivors of cancer diagnosed during childhood or adolescence: specifically on second malignancies, on cardiac disease and on late mortality. Fifteen institutions from 10 European countries are pooling their data to create a very large data set of more than 90,000 survivors of childhood and adolescent cancer. Now past the half-way mark, the PCSF participants are actively engaged in data acquisition and planning the first publications. The guidelines group has joined forces with colleagues from the US and Canada to create harmonized guidelines for follow-up care based on the best global information to help provide every European survivor and their families with better access to care and better long-term health.

Dr Byrne is Research Liaison in this consortium, serves on the Publications Committee and is a member of the Executive Board. In addition, she is deputy leader of WP7 – the dissemination and training work package. As such, the Boyne Research Institute is actively engaged in PCSF with dissemination of methods and results, communication with stakeholders and reviews of procedures. In 2013 WP7 organised the “The Race of Brave Bikers” in Marostica, Italy, and the “Cruise toward Life” in Genoa.

Boyne Research Institute continued its involvement of coordinating the translation of the shortened version of the English PanCareSurFup brochure into different European languages. A brochure in Slovenian was printed this year and a version in Czech is in the process of being produced. To view/download these brochures visit our website at:

<http://www.boyneresearch.ie/brochures.html>.



4. **PanCareLIFE (PanCare Studies in Fertility and Ototoxicity to Improve Quality of Life after Cancer during Childhood, Adolescence and Young Adulthood www.pancarelife.eu):**



PanCareLIFE is a €6 million EU-funded, five-year project that started in November 2013, to investigate three serious late effects, namely, loss of fertility, loss of hearing and impaired quality of life. A group of 15 institutions from 8 European countries will collect data from more than 12,000 survivors to evaluate the risks to their long-term health, to develop guidelines for care, and to disseminate results to the general public. This innovative project will look at the ways that genetic make-up can modify risks of late effects. Stratifying survivors into different risk groups so that long-term care can be delivered more efficiently will save survivors and the health-care system time, worry and money. The Boyne Research Institute serves as Research Manager for the PanCareLIFE consortium and looks forward to our active engagement in all aspects of this project.



PanCareLIFE Kick-off Meeting Mainz November 2013

RACE of BRAVE BIKERS, April 2013



'The Race of Brave Bikers', now in its second year, took place on the 28th/29th April 2013 in the mediaeval town of Marostica, Italy. The event was organised by PanCareSurFup as part of its dissemination activities to raise awareness of the increasing number of paediatric cancer survivors in Europe. Funding for the bicycles and cycling gear (*see photos*) was provided by the NEAR Foundation and the Barilla Food Group, and was run in association with the professional bicycle race – GranFondo Fisik. Dr Byrne travelled to Marostica and cycled with the cancer survivors who came from all over Europe.

The week-end also included a roundtable featuring experts and doctors from leading European hospitals and institutions, whose message was that every European childhood cancer survivor deserves access to the best possible care and better long-term health. The event demonstrates the importance of optimum quality-of-life for the survivors and the need for research studies into the late effects of their complex paediatric cancer treatment. This wonderful event will take place annually until 2016.





CRUISE TOWARD LIFE, October 2013



PanCare and PanCareSurFup were delighted to be part of the fundraising event - "Cruise toward Life; Mettiamoci all'Opera" which was held in Genoa, Italy on 24th/25th October 2013. The event was run by MSC Cruises with the support of other Italian sponsors. The aim was to inform people, not directly involved in childhood cancer, about the success in Europe of treating children and adolescents affected by cancer, the importance of the cooperative work of a European group like PanCare, and the goal of achieving a complete cure in childhood cancer, which is today a reality, as well as the task of caring about the health status of long-term survivors (role of PanCareSurFup).

Dr Byrne attended the evening aboard *MSC Opera* which included a cocktail reception, dinner, speeches, entertainment and an auction. The evening was a great success with 1,200 guests in attendance. Boyne Research Institute was involved in producing a banner and postcard for the event to raise awareness of PanCare. A press conference and round table was arranged for the following day. It involved scientific experts from Italy (AIEOP), Europe (PanCare), and the U.S., who discussed results, problems, and projects on childhood cancer.



BRI DONATIONS 2013

Ita Deveney, a member of the Toner family who are long-time supporters of the Boyne Research Institute, kindly donated €305 on behalf of the Sequoia Ladies Golfing Society of Armagh, Northern Ireland to continue the Institute's work into neural tube defects. Dr Byrne, CEO of the Boyne Research Institute, expressed her sincere thanks to Mrs Deveney and the Sequoia Ladies for their generosity and continued support. Dr Byrne thanked all our donors for their support of our forthcoming Summer Student Programme 2014.

STAFF & VOLUNTEERS

Staff in 2013 consisted of the CEO, Dr. Julianne Byrne, assisted by research administrator Rebecca Lawler. Rosaire Kelly continues to provide valuable support for the Boyne Research Institute.

MEETINGS & PRESENTATIONS

1. **PanCare** Meetings in 2013: Spring meeting in Genoa, Italy, 17-19 April; Autumn meeting in Amsterdam, The Netherlands, 1-2 October.
2. **PanCareSurFup**: Spring meeting in Genoa, Italy, 16-17 April; Autumn meeting in Amsterdam, 2-3 October.
3. **PanCareLIFE**: Kickoff meeting in Mainz, Germany, November 2013.



COLLABORATIONS

For a number of years, the Boyne Research Institute has participated in national and international collaborations, in areas covering developmental problems resulting in birth defects and cancer during childhood. At present, we are involved in collaborative research with research teams in Ireland, Europe and the United States of America.

- ❖ Dr Paul Walsh, Dr. Harry Comber, National Cancer Registry of Ireland, Cork, Ireland
- ❖ Dr RJ Berry, National Center on Birth Defects & Developmental Disabilities, Centers for Disease Control & Prevention, Atlanta, GA, USA,
- ❖ Dr. Lars Hjorth, University of Lund, Sweden (chairman, PanCare & coordinator, PanCareSurFup)
- ❖ Dr Peter Kaatsch, University of Mainz, Germany (coordinator, PanCareLIFE)

GOVERNANCE OF THE BOYNE RESEARCH INSTITUTE, 2013



We are grateful to those members of our Board of Trustees who give freely of their time and energy to help advance the mission of the Boyne Research Institute. The Board of Trustees meets quarterly.
