Wales Gene Park Education & Engagement

Highlights of 2015
Wales Gene Park Education & Engagement

Established in 2002, the Wales Gene Park continues to operate in a field of fast developing technologies and rapidly emerging opportunities for their application. Going forward into the second decade since the first release of the human genome sequence, we want to ensure that researchers in Wales are in a position to meet the challenges and opportunities presented by human genetics and genomics and that our health practitioners can use new genetic and genomic knowledge to prevent, better diagnose and better treat human illness. To do this the Wales Gene Park provides technology and expertise, trains and supports researchers and engages with and educates professionals and the public. This broad portfolio of activity is undertaken by an able and enthusiastic team without which the high standard of genetic research and education in Wales would be unsustainable.

Education & Engagement Programme

The Wales Gene Park delivers an innovative and continually developing annual programme of Education & Engagement events.

Continuing professional development is provided for health care professionals through a range of conferences, seminars and workshops on all aspects of genetics. These events provide information about the latest advances in genetics to further the education and training of these professionals and keep them up to date with this rapidly changing field.

For teachers, we provide continuing professional development on the social and ethical issues surrounding gene technology and research-based topics. We also have a Teachers’ Genetics Network with over 380 members who receive a termly newsletter containing genetics-related news and information. For students we hold a 6th Form Conference and a Genetics Roadshow on alternate years as well as organising one off events such the interactive dramas ‘Boy Genius’ and ‘Meet the Mighty Gene Machine’ and also consultation sessions and discussions throughout the year.

One of our key objectives is to raise public awareness of genetics and this is being achieved through a range of innovative projects, often in partnership with Techniquest. Past projects include a Citizens’ Jury for young people on the issue of ‘Designer Babies’, a mock trial about the National DNA Database, a hands-on exhibition called ‘Diversity, History and Inheritance in Cardiff Bay’. Regular public discussion events on topical genetics issues and illuminating public lectures on subjects such as DNA and crime detection are also organised.

By funding a Development Officer for Wales, first with the Genetic Interest Group and then with its successor organisation, Genetic Alliance UK, the Wales Gene Park has supported patients and families affected by genetic disorders and over 160 patient organisations and groups that represent them. The Wales Gene Park has brought benefits to people with genetic and rare conditions by increasing knowledge through research and its translation into NHS services. The patient and family perspective is at the centre of policy and decision-making. The Wales Gene Park ensures that this view is core to all activities undertaken within research, NHS and policy settings and inputs this to health and social policy work in Wales. This has been achieved by the development of reports and submission of oral and written evidence to the Welsh Government.
Highlights of 2015

Health Professionals

Living with Genetic Conditions

This educational session begins with a genetic counsellor, or other health professional, giving a talk on genetics and inheritance. This include a brief refresher covering some basic genetics concepts, along with more specific information about the genetic condition on which the session will focus e.g. cystic fibrosis.

Someone affected by a particular genetic condition, either an individual with the condition, a family member, or carer, then presents a personal narrative. This gives an insight into the impact a genetic condition can have on an individual or family, and provides a powerful account of how it affects them on a daily basis. Speakers often describe how their condition was diagnosed, their medical regime, treatment and management, and how it can affect them on a social or psychological level. Educationally it can help to link theory and practice, and highlight the relevance of genetics to healthcare.

Following the talks, there is a question and answer session, which usually elicits a range of interesting questions from the audience. Genetic conditions that have been the subject of these sessions include cystic fibrosis, tuberous sclerosis complex, and Huntington’s disease.

Originally intended for, and delivered to, Year 12 and 13 school pupils, the sessions have recently been extended to undergraduate students on nursing degree courses at Welsh universities including Cardiff University and University of South Wales. These undergraduate sessions have been positively received by both the students and educators. They have evaluated extremely well, with the vast majority of students reporting that they felt more informed about genetics and wanted to know more about the subject. Anecdotal feedback from the speakers indicates that this is also a valuable opportunity for them to enhance their teaching experience and highlight the role of genetics services, for the patients and family members to have a voice and to improve knowledge and understanding amongst future health professionals and others of what it is like to live with a genetic condition.

“Through sharing our personal experiences of family life and living with a rare genetic disorder, I have been empowered.....sharing my experiences with students and others who will provide care for families like mine is so important; knowledge is key to improve future outcomes for my son and others who live with a genetic disorder”

Speaker, Living with Genetic Conditions

“It’s a fantastic and rare opportunity for trainees to really hear about and understand the experiences of families. This leads to better care for these families as trainees provide better support to patients and know how to link in with genetic services”

Genetic Counsellor
Cardiff International Cardiovascular Conference

The Wales Gene Park has worked, for several years, in collaboration with Dr Dhavendra Kumar of the All Wales Medical Genetics Service to deliver a bi-annual conference on Cardiovascular Genetics. After four successful events, it was decided to combine this year’s event with the annual conferences of the following organisations: The British Congenital Cardiac Association (BCCA), Paediatricians with Expertise in Cardiology Special Interest Group (PECSIG), the British Adult Congenital Cardiac Nurses Association (BACCNA) and the Congenital Nurses Association (CCNA).

This International conference provided an excellent forum for information sharing, discussion and debate on recent advances, new clinical applications and contentious issues related to common and rare inherited cardiovascular conditions. The event focussed on early and young onset diseases but also included many adult and late onset Mendelian and complex cardiovascular disorders. The faculty included distinguished speakers from the UK, Europe and North America.

The event attracted just under 500 people to Cardiff from to listen to world-renowned speakers give updates in the field and to network.

Association for Inherited Cardiovascular Conditions (AICC)

Management of inherited cardiac conditions is currently one of the most rapidly evolving areas in cardiology. The last decade has seen a major change in our recognition of these conditions from apparently rare isolated disorders, to a group which collectively are common and which may be associated with the tragedy of unnecessary sudden death. The AICC aims to provide consistent, top quality education and training, advice on management and best practice, as well as acting as a forum for data collection, audit and collaborative research. Membership is open to clinicians, nurses, counsellors, scientists and professions allied to medicine, as well as to persons from organisations and charities involved in support of such families.

The AICC held its first Annual Meeting & AGM in Cardiff in 2011. It was organised to coincide with the established Cardiff Cardiovascular Genetics meeting. This year was the 3rd time that the meeting had been held in Cardiff, being held in London on alternate years. Over 130 delegates from across the UK attended the meeting to find out about the latest advances and to network and share their expertise.

Hospital Saturday Fund Awards Evening

The Hospital Saturday Fund is a registered charity whose aims are to provide assistance through its charitable funds for:

- Individuals with medical conditions or disability who would benefit from assistance with the purchase of specialised equipment or from practical forms of treatment
- Registered health charities such as hospitals, hospices and medical organisations who are in need of grants for medical projects, care, research or support of medical training

In November, the Wales Gene Park hosted the Awards Evening for local organisations & individuals who obtained funding from the charity. The event was attended by over 90 people and during the evening, Professor Julian Sampson received a grant toward the cost of continued research into the genetic condition Tuberous Sclerosis.
Schools & Colleges

Schools’ Genetics Roadshow

After holding the Sixth Form Genetics Conference for a few years, it became clear that not all schools in Wales were able to travel to Cardiff or Wrexham to participate. In response, the Wales Gene Park set about organising a roadshow that would take genetics into schools all across Wales & the bordering counties. Each event on the roadshow offers up to three genetics experts who talk to a minimum audience of 100 students. Schools with fewer students join with other local schools to make up the numbers. This initiative was a success from the start and the 2014-15 Roadshow reached nearly 3,600 students from 57 schools/colleges. Over 70 speakers from academia, the NHS and industry gave over 100 genetics-related talks and many schools have already requested a repeat visit from the next roadshow.

Other Educational Events

In addition to our regular programme for schools and colleges, the Education and Engagement team also took part in several other activities, which included:

- A Careers Fayre at Bassaleg School; pupils from years 9 to 13 had a chance to visit Wales Gene Park’s stand to find out more about courses and careers related genetics.

- A DNA and forensic science workshop at Rhymney Comprehensive School; year 10 pupils took part in an interactive workshop about DNA and found out how DNA fingerprinting is used in forensic science.
• Teachers’ focus group and Continuing Professional Development session, an event was held in collaboration with Techniquest entitled ‘Cutting Edge Science – What’s new in Genetics?’ The day comprised a series of talks showcasing the latest developments in genetics and genomics, a bioinformatics taster session, and a focus group to ascertain teachers’ requirements in terms of genetics education. The event was sponsored by the National Learning Centre.

• A Film Screening of Jurassic World and genetics talk; over 130 year 8 and 9 pupils from schools across south Wales attended a special screening of Jurassic World hosted in collaboration with Into Film as part of their annual schools’ film festival. After the film screening, Dr Rhys Jones gave a talk on genetic engineering and cloning.

• A Pathology Sixth Form Day run by Cardiff University School of Medicine; year 12 and 13 pupils from schools across south Wales attended an event to explore the specialism of pathology, and visited Wales Gene Park’s interactive stand to find out more about genetics.
A Living with Genetic Conditions Summer School; this session was held as part of Cardiff University's Step Up Plus Widening Access Summer School for year 12 pupils with the ability to succeed at higher education. Genetic Counselor Dr Nicki Taverner began with a talk on inherited conditions, which was followed by a talk from Beth Clarke, who spoke about the impact cystic fibrosis has on her life. Pupils also had the opportunity to put questions to the speakers.

Public Groups

Community Group Talks

A number of genetics-focused talks were delivered to community organisations including Women’s Institute, Probus, and University of the Third Age groups throughout Wales. Topics covered ranged from genetics and genomics in everyday life, next generation sequencing to using DNA technology in forensic science. Groups participating in these talks included Flintshire U3A, Shire Newton WI, Monmouth U3A, Brecon U3A, Abergavenny Probus, Pontllanfraith Probus and The Vale Probus.

Public Talks

Wales Gene Park organises at least three talks each year, which are aimed at engaging the general public in topical genetics subjects. Audiences range from school pupils and medical students through to retired members of the public.

Three public talks were held during 2015.

- Clues in your poo: How screening is helping the fight against bowel cancer

Professor Julian Sampson (Wales Gene Park and Cardiff University), Hayley Heard (Bowel Screening Wales) and Dr Sunil Dolwani (Cardiff and Vale University Health Board) discussed inherited bowel cancer, the national screening programme, and clinical advances in the field. The audience also heard a personal account from someone affected by inherited bowel cancer, which was followed by a question and answer session with the expert panel.
• Richard III: From Car Park to Cathedral

Dr Turi King and Carl Vivian (University of Leicester) spoke to an audience of over 400 members of the public about the discovery of King Richard III, and how DNA was used to identify the remains of the king in the car park.

“Great delivery of a very complex subject”

“Made the science easy to understand – excellent”

“Will remember this for a long time – thank you”

“Brilliant entertainment and educators”

• Will this treatment work for me? How genomics is helping to personalise medicine

Dr Rachel Butler (All Wales Medical Genetics Service), Professor Phil Routledge (Cardiff University) and Dr Andrew Owen (Liverpool University) spoke on genomic medicine and the Cancer Research UK Stratified Medicine Programme and advances in pharmacogenomics.

“Good and informative evening with easy to understand speakers”
Patient Groups

Policy work

UK Strategy for Rare Diseases & Welsh Implementation Plan for Rare Diseases

Rare Disease UK was a campaign established by Genetic Alliance UK calling on the UK governments to implement the EU Directive for all Member States to develop a Plan for rare diseases by the end of 2013. The Development Officer for Genetic Alliance UK in Wales was a member of the UK Rare Disease Forum who were responsible for developing the UK response: https://www.gov.uk/government/uploads/system/uploads/attachment_data/file/260562/UK_Strategy_for_Rare_Diseases.pdf

The Development Officer was also a member of the group that was tasked with drafting the Welsh Government response to the UK Strategy for Rare Diseases, providing support in drafting the patient empowerment section of the Welsh Plan. Engagement events were facilitated by the Development Officer between Welsh Government and patients and patient group representatives to feed their views into Welsh Government regarding development of the plan.

The Development Officer sits as a member of the Implementation Group for the Rare Disease Plan, which has oversight of the implementation of the plan’s recommendations in Wales.

The plan is available to view here: http://gov.wales/docs/dhss/publications/150220rarediseaseplanen.pdf

The Rare Disease UK campaign was shortlisted for ‘Best Campaign in Wales’ at the 2015 Public Affairs Awards.

Genomics & Stratified Medicine

A key priority in 2015 was awareness raising in the National Assembly for Wales regarding access to specialised services and therapies including genetic testing. Genetic Alliance UK launched a report at Ty Hywel in January 2015 with key recommendations outlining improvements required across the area of access to specialised services and therapies following consultation and engagement with a variety of stakeholders. The report is available to view online: http://www.geneticalliance.org.uk/docs/accesswales.pdf.

Meetings with the then Deputy Minister for Health, Vaughan Gething to emphasise the importance of keeping pace with developments within the field of stratified medicine followed.

We collaborated with Cancer Research UK and the Association of British Pharmaceutical Industry (ABPI) in September 2015 to provide a series of education events across the Assembly to explain the role of stratified medicines and their important contribution to improving and sustainable healthcare.
Improving Access to Orphan and Ultra-orphan Medicines in NHS Wales

Written and oral evidence was presented to the consultation on the appraisal process for orphan and ultra-orphan medicines in Wales. Working with Public Health Wales and Welsh Government, the Development Officer helped to set up an event to consult with patients and patient groups.

The Development Officer for Genetic Alliance UK played a key role in the reviews of the orphan and ultra-orphan appraisal process and Individual Patient Funding Request (IPFR) process. This led to a £1 million investment by the Minister in a new service provided by the All Wales Therapeutics and Toxicology Committee (AWTTC) to improve the appraisal of and patients’ access to new and innovative medicines, including orphan and ultra-orphan drugs.

The All Wales Medicines Strategy Group (AWMSG) will now provide expert advice on the use of off-label medicines. Health Boards and WHSSC have developed a single, all-Wales decision for each patient cohort supported by advice from AWTTC. Our Development Officer in Wales has inputted to the work of AWTTC to develop these new processes. The Development Officer has been a patient representative for the first two appraisals to go through the new orphan and ultra-orphan appraisal process, Clinical and Patient Involvement Group (CAPIG).

All advice following the appraisal of a new medicine must go to the Cabinet Secretary for Health for ratification.

Individual Patient Funding Request Process (IPFR) Review

As a member of the group that conducted a review of the IPFR process, the Development Officer represented the patient perspective alongside the Cystic Fibrosis Trust on the group. This led to a report, which made 10 recommendations on how to strengthen the IPFR process in Wales– they were all accepted by the former Minister for Health and Social Services, Mark Drakeford. Work to implement the recommendations has been undertaken and the process will be strengthened further by a new role for the All Wales Therapeutics and Toxicology Centre (AWTTC).

Other work has included Input into a roundtable discussion with Assembly Members and patient groups around access to medicines for rare diseases speaking specifically about the IPFR process and exceptionality criteria underpinning this.

Access to Cross-border Healthcare Arrangements Between England and Wales

The Welsh Affairs Committee published their report on cross-border healthcare arrangements in 2015. Genetic Alliance UK provided written and oral evidence to the inquiry and attended a workshop led by Committee members. Genetic Alliance UK is quoted in the report which makes a number of recommendations about improvements needed to ensure that Welsh patients with rare diseases receive timely and equitable access to specialised services in England - the report is available: [http://www.publications.parliament.uk/pa/cm201415/cmselect/cmwelaf/404/404.pdf](http://www.publications.parliament.uk/pa/cm201415/cmselect/cmwelaf/404/404.pdf)

Other policy work:

- Submitted a response to the Welsh Government consultation regarding proposed changes to the Independent Living Fund
- Attended workshop and submitted consultation response regarding changes to the Human Tissue Authority Codes of Practice and Standards
Invited to be a member of the Task and Finish Group looking at Genetic Testing for Stratified Medicine to develop commissioning structures for making genetic tests available to Welsh patients and to make decisions on what tests are funded. The group is led by Rachel Butler, Head of the NHS Laboratories at the All Wales Medical Genetics Service (AWMGS) and the Welsh Health Specialised Services Committee (WHSSC).

**Engagement with Patients and Patient Groups**

**The Helping Welsh Patients Project**

Funding of £15,000 was secured from John Ellerman Foundation, Waterloo Foundation and Oakdale Trust to support this project which aims to develop peer-to-peer support networks for rare and genetic conditions where none is available locally.

- Restless Legs Syndrome: Set up a support group in Wales linked to RLS UK, held an event to bring together patients, health professionals and researchers in late 2014.
- Tuberous Sclerosis Complex: Worked with Welsh volunteer and local advisor to set up a Welsh support group and recruit more families to the network. Funding from Waterloo Foundation to undertake activities to support carers and families.
- Rare Inherited Eye Disease: Working with patients and families to develop a patient group in South Wales. The first meeting of the group was held on Rare Disease Day 2015 (28th February). A Facebook group has been set up for patients and families to interact.

**Rare Disease Day 2015**

The Welsh Rare Disease Day reception was held in the Senedd on 3rd March. Rare Disease Day is an annual event, which provides the rare disease community across the world with an opportunity to increase awareness of rare diseases and highlight rare diseases as a public health priority to government and health departments.

The Deputy Minister for Health spoke at the event to launch the Welsh Implementation Plan for Rare Diseases. The plan is now available online: [http://gov.wales/topics/health/nhswna/plans/rare/?lan=et](http://gov.wales/topics/health/nhswna/plans/rare/?lan=et).

The launch of the plan was covered by the media: [http://www.walesonline.co.uk/news/health/new-plan-launched-tackle-rare-8754398](http://www.walesonline.co.uk/news/health/new-plan-launched-tackle-rare-8754398)

Dr Rachel Butler, Head of Laboratories at the All Wales Medical Genetics Service spoke about new developments in genomics and stratified medicine and Rachael Humphreys who has the rare condition Behcets Syndrome spoke about her experiences of living with a rare disease and some of the difficulties for patients in accessing specialised services. There was cross party support for the event with representation from the Welsh Government, Vaughan Deputy, the then Deputy Minister for Health, Darren Millar AM who was Shadow Health Minister and Kirsty Williams AM who was Leader of the Welsh Liberal Democrats co-sponsored the event. Darren closed the event, which was attended by over 100 stakeholders including representatives from Welsh Government, the Medical Director for Cardiff and Vale Health Board, commissioners from the Welsh Health Specialised Services Committee as well as researchers, patient groups, patients and families affected by rare diseases.
**Selection of Patient Focused Events held in 2015**

**Rare Inherited Eye Disease Inaugural Patient Day**

This meeting brought together over 50 people affected by rare inherited eye disease, especially those located in South Wales. The meeting brought together families, patient groups and health professionals who had an opportunity to learn about services and support available as well as hear about research opportunities. The Optometric Adviser to the Welsh Government, Dr Barbara Ryan, spoke about the Wales Eye Care Plan and patients with inherited eye disease, and there was an information session about the Welsh Rare Disease Implementation Plan. A key element of the day was an opportunity for patients to meet with others with the same rare condition. There was also a patient panel and Q&A session with the audience.

**Launch of Tuberous Sclerosis Complex Clinic**

Wales Gene Park & Genetic Alliance UK supported the organisation of a launch event for the first specialist clinic for tuberous sclerosis in Wales. Over 70 patients, carers and health professionals gathered on Friday 8th May for the launch of the new specialist clinic for the rare condition, tuberous sclerosis complex (TSC) to be hosted by Cardiff and Vale University Health Board. The number of carers attending the event was 31. The clinic will enable patients with this rare condition to receive more comprehensive expert management and treatment.

Genetic Alliance UK hosted an event in Cardiff on 8th May to raise awareness of the clinic amongst patients and families which will be held at the University Hospital of Wales. Dr Graham Shortland, Medical Director of Cardiff and Vale University Health Board officially launched the clinic and there were presentations from clinicians and researchers, Jayne Spink, the Chief Executive Officer of the Tuberous Sclerosis Association and a young patient.

Marie James is the parent of a young adult with the condition and is also a trustee of the Tuberous Sclerosis Association.

Marie commented:

"I welcome with great excitement the launch of a TSC clinic in Wales. Professor Julian Sampson and his team at the Institute of Medical Genetics at Cardiff University has a long standing association with exciting and groundbreaking progress in the field of tuberous sclerosis. The clinic will be ideally located at the University Hospital of Wales linking with established clinical and research expertise. The development of this clinic will undoubtedly facilitate improved access to specialist care, treatment and support for TSC patients and their families here in Wales."

**Behçet’s in a Day Event**

Behçet’s in a Day is an educational event and the first of its kind in Wales to bring together patients and health professionals to find out more about this rare condition, its management and treatment options.

Genetic Alliance UK supported one of the Welsh trustees of the Behçet’s Syndrome Society to organise the event which attracted over 90 delegates including secondary care health professionals, commissioners and patients and families. Specialists from the three Centres of Excellence in England presented at the event on areas including treatment of Behçet’s, oral care in Behçet’s, and Behçet’s
and the eye. It also provided an opportunity to highlight issues that patients with Behçet’s have been experiencing in trying to gain access to a specialist centre in England.

Rachael Humphreys, who is a patient and trustee of the Behçet’s Syndrome Society, spoke at the event and gave her perspective of living with the condition. She will be driving forward her campaign to ensure that patients in Wales receive equitable access to the Centres of Excellence in England.

Rachael also spoke to Wales online to raise awareness of the condition and improvements needed to support patients in Wales: [http://www.walesonline.co.uk/news/health/rachael-humphreys-painful-ulcers-over-q203884](http://www.walesonline.co.uk/news/health/rachael-humphreys-painful-ulcers-over-q203884).

Rachel commented:

“Since attending Rare Diseases Day back in 2013 I have seen first-hand the support, commitment and passion Genetic Alliance UK has in representing patients with rare diseases, and their organisations. They work closely with patients, promoting rare diseases, highlighting inequality of care, and fighting for change. They offer networking opportunities, not only to meet other patients, but also members of the Welsh Assembly Government, as well as health care representatives from specialised services. Recently, Genetic Alliance UK and the Wales Gene Park helped me organise a one-day conference designed to educate patients and healthcare professionals on Behcet’s Disease so that standards of care and understanding of the disease could be improved. This could not have happened without their experience and commitment to improve specialist services for patients with rare diseases in Wales.”

**Rare Disease Patient Network Launch**

The Rare Disease Patient Network was launched in October 2015 and the inaugural meeting was attended by over 80 patients, families, researchers and healthcare professionals.

The rare disease network was formed to engage patients, families and patient organisation representatives in the work that the Wales Gene Park is undertaking in Wales, in particular to support the implementation of the Welsh Rare Disease Plan and improve engagement in research opportunities.
Other Events Attended

The Wales Gene Park also had its interactive stand at numerous other events throughout the year including the Cancer Research UK Open Day, Wales Cancer Research Centre launch, and the Involving People Network meeting.
## Attendance at Education & Engagement Events in 2015

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<td>CRUK Open Day</td>
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<td>Stratified Medicine Meeting @ The Senedd</td>
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<td></td>
<td>Rare Disease Network Launch</td>
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<td></td>
<td>Public Talk – Richard III</td>
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<td>November</td>
<td>Cardiff International Cardiovascular Conference</td>
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<td>Schools’ Genetics Roadshow</td>
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<td>Association for Inherited Cardiac Conditions</td>
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<td>HSF Awards Evening</td>
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<td>Genetics Talks Old Castle Primary School Bridgend</td>
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<td>Trainee Meeting</td>
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<td>Schools’ Film Festival – Jurassic World</td>
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<td>December</td>
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<td>Public Talk – Bowel Cancer</td>
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<td>Totals</td>
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A total of **6565** people attended WGP Education & Engagement events in 2015
Attendees at WGP Events 2003-2015

Total Attendees at WGP Events 2003 - 2015