

# **Autism Speaks Science Strategy 2009-2014**

## **Science in the Service of Autism**



**AUTISM SPEAKS™**  
It's time to listen.

## **Autism Speaks**

UK charity Autism Speaks seeks to use biomedical research to bring benefits to individuals and families affected by autism spectrum disorders. Autism Speaks is dedicated to raising and investing funds to support high-quality peer reviewed research which focuses on determining the causes and biological basis of autism spectrum disorders; improving diagnosis; and advancing and evaluating new treatments and interventions. We are committed to ensuring that increased understanding and new scientific knowledge will improve the quality of life for all those affected.

Autism Speaks has a small but highly committed group of staff to fulfill these aims and since it began work in 2005 the charity has achieved demonstrable results benefiting the autism community by securing new funds for state-of-the art research, leveraging additional research funding from other organisations, and playing a central advisory role on autism research policy through membership of key groups such as the Autism Research Co-ordination Group and direct work with government. The success of the fundraising team in generating new funds means that Autism Speaks is now in a position to consider strategically how it will spend funds not already committed to current projects and its views on the future direction of UK autism research.

This strategy is complemented by Autism Speaks' rolling 3-year business plan, which sets out how the charity will secure the funds needed to underpin the ambitions set out in this document.

Note 1: In this document the term Autism Speaks refers to the UK charity of that name. The US charity of the same name is always described as Autism Speaks Inc.

Note 2: In this document the terms autism and autism spectrum disorder (ASD) both refer to the group of disorders including childhood autism (also known as classical or infantile autism or autistic disorder); Asperger syndrome; pervasive developmental disorder not otherwise specified (PDD-NOS); atypical autism and childhood disintegrative disorder (CDD).

# 1 Executive Summary

Autism is a lifelong disorder that arises from atypical development and functioning of the brain. It is characterised by difficulties in social interaction and unusual patterns of language development, with a tendency to engage in repetitive and/or circumscribed behaviours. Around 1 in 100 people has an autism spectrum disorder yet its causes and nature have until recently been poorly understood. Despite significant advances in funding and discovery over the last five years, led primarily by the United States, research spending is low in comparison to other conditions with similar impact. Many individuals and their families experience significant distress and it has been estimated that the economic impact of autism in the UK is equivalent to £28bn a year.

Autism Speaks' main focus is to fund research that will ultimately bring benefits to the lives of individuals with autism and their families. The UK autism research community is strong and competitive and continues to be progressive in the scientific approaches it takes but Autism Speaks believes that an essential next step is to translate basic medical research findings into meaningful interventions.

Thus in preparing this strategy Autism Speaks has given careful consideration not only to which areas of autism research to fund but also the influence it can bring to bear on the state and direction of autism science. By working closely with international partners such as Autism Speaks Inc and with UK partners such as the Medical Research Council (MRC), the Wellcome Trust, Research Autism and other trusts and charities in the UK, Autism Speaks believes it can stimulate the research community, inspire growth and best practice, encourage researchers to work closely with the stakeholder communities and bring in new expertise.

The current state of autism research; the particular strengths of the UK autism research community; and the published plans of other major funders have been key factors in identifying the ways in which Autism Speaks can contribute strategically to making the UK a vibrant and effective centre of excellence in autism research. These include:

- Developing a sustainable research infrastructure
- Utilising existing and developing new research capacity
- Adding to existing successful research collaborations
- Building on the UK's strengths in neuro- and cognitive science; and
- Consulting with the autism community on the direction of research.

The key scientific questions which Autism Speaks believes must be addressed over the five years of this current strategy are as follows:

- What causes autism and how can we use that knowledge to ease the burden for families?
- What are the specific characteristics of autism that are amenable and relevant to treatment?
- How can we improve early screening and diagnosis, and our understanding of how autism changes over time, in order to achieve timely and appropriate intervention?

Taking each of these in turn, the key goals to be achieved in understanding the causes of autism include continuing identification of the genetic risk markers for autism and how they might be used in diagnosis and treatment; and greater exploration of the role of non-genetic or environmental factors.

In terms of understanding the specific characteristics of autism that are amenable to treatment it will be important to bring together all the relevant biological and behavioral data to identify particular sub-groups; and to explore more thoroughly the medical conditions associated with autism: both of which will be facilitated by increasing the number of biological researchers studying autism.

The questions of improving diagnosis and how autism manifests itself over time need to be addressed by ensuring that diagnosis is based on the latest knowledge and techniques, and that there is improved understanding of autism in adolescents and adults.

Autism Speaks can help in the achievement of these goals in a number of ways including scientific meetings and fora, calls for proposals, fellowships, creating and supporting shared research resources, partnership working, lobbying and the conduct of feasibility studies.

The first draft of this strategy was the subject of consultation with a wide range of stakeholders including researchers, families affected by autism, and research funders. Their comments and suggestions have been incorporated into this document and we gratefully acknowledge their contribution.

It is hoped that this science strategy will provide a guide to families, funders and the research community to the areas and direction of Autism Speaks' research funding for the period 2009-14, and by measuring progress annually against the key goals, a way to gauge progress in scientific development over the next 5 years.

## **2 Setting the context**

### **2.1 Autism Spectrum Disorder**

Approximately 1 in 100 individuals in the UK has an autism spectrum disorder (ASD), ranging from individuals with severe learning difficulties and no language through to individuals with IQs in the average range able to hold down a job or start a family. Along with the well characterised difficulties in social and communication skills, and a tendency to focus on repetitive and stereotyped behaviors, individuals with ASD also have higher rates of intellectual impairment, epilepsy, sleep difficulties, gut problems and abnormal sensory responses. Approximately a quarter of individuals with ASD undergo a period of development during the first few years of life when they regress (i.e. there is a period when their development noticeably declines) but what causes this regression remains unknown. As more adolescents and adults with ASD are being diagnosed it is becoming increasingly apparent that they initially present clinically with a mood or anxiety disorder for which they are often prescribed medication.

Children with ASD can be diagnosed reliably from about three years old but the study of infants at increased risk of developing ASD because of a family history (i.e. they have an older brother or sister already diagnosed with ASD) is helping us to understand that there are behavioral characteristics (e.g. failure to respond to name) and biomarkers (e.g. unusual pattern of head growth) that may characterise this at risk population in early development. Under international diagnostic criteria ASD is defined as a pervasive developmental disorder, meaning it may change over time but it does not go away, and there are many adults in the UK with ASD, a significant proportion of which are either wrongly or undiagnosed. The use of standardised diagnostic assessments (e.g. the Autism Diagnostic Interview and the Autism Diagnostic Observation Schedule) in recent years has been a great advantage to the autism clinical and research community, allowing for more cases to be diagnosed, a greater consensus on clinical diagnosis and for researchers to compare their findings by setting agreed inclusion criteria.

ASD was shown to be a strongly inherited disorder by the study of identical and non-identical twins in the 1970s and 1990s. In identical twin pairs (who share 100% of their genes) if one twin had ASD the chance of the other having ASD was somewhere between 60-90%. In non-identical twin pairs (who share 50% of their genes) if one twin had ASD the chance of the second twin having ASD was between 0-10%. Based on the evidence of these twin studies whole genome scans on large scale collections of DNA from autism families are now being carried out. Studies have looked for linkages (such as chromosomal regions most commonly shared by autism families), identified candidate genes and most recently explored the significance of copy number variations (sub-microscopic regions of DNA that are "replicated" or "deleted"), which together have led to significant advances in genetics research and identification of the underlying genetic factors in up to 40% of ASD cases. Of particular interest is the identification of genes involved in ASD that influence how synapses in the brain develop and function. Technological advances in genetic analysis now allow scientists to conduct research in a fraction of the time it would have previously taken and the wealth of current research is leading scientists to a greater understanding of the multiple causal routes to developing ASD.

Despite the importance of genetics research, and the potential it holds to identify the causes for a possible range of 'autisms', it is becoming increasingly apparent that non-genetic and environmental factors also play a causal role. Indeed, genetically identical autism twin pairs sometimes appear very different in their behavioral and clinical profile, suggesting a causal role for genetic and non-genetic factors in these cases. The environmental factors in ASD (i.e. those factors that might affect the development of the brain both pre- and post-natally) are beginning to be explored vigorously, particularly in the US, and recent findings of increased parental age and maternal immune dysfunction as risk factors for ASD are evidence of this research bearing fruit.

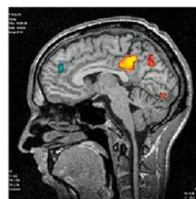
Research of the biological basis of ASD has mainly focused on neurobiology. Advances in neuroimaging (techniques of visualising the structure and working human brain) along with the collection of post-mortem human brain tissue in repositories have allowed scientists to model structural and functional differences in the brains of individuals with ASD. Neuropathological research (the study of post-mortem brain tissue) is the most effective method for scientists to examine the microscopic detail of how cells within the brain are organised and structured, and of any differences in brain chemistry. It is also a way for scientists to show how specific genes underlying ASD may be expressed in a person's brain tissue. Magnetic Resonance Imaging technology (MRI - see picture below) helps scientists to form clear pictures of the structures of the brain, how the brain functions whilst a person completes a task (functional or fMRI) and the connections between different regions of the brain (diffusion tensor imaging or DTI). Neuroimaging research suggests that individuals with ASD have regions of the brain that are connected atypically, in particular the areas of the brain underlying social behavior, and it is this abnormal connectivity that manifests in unusual patterns of thinking and behaving.



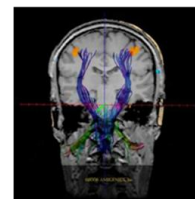
Magnetic Resonance Imaging



MRI



fMRI



DTI

#### Techniques using magnetic resonance imaging

There are currently no accepted behavioral or pharmaceutical interventions that will ameliorate all symptoms of ASD and the only US FDA (Food and Drug Administration) approved medication (risperidone) is to treat irritability associated with autistic disorder. The core deficits of autism (social, communication, repetitive/stereotyped behavior) are not yet treatable using medication. ABA (Applied Behavioral Analysis), TEACCH (Treatment and Education of Autistic and related Communication-handicapped Children) and PECS (Picture Exchange Communication System) are amongst the small proportion of interventions for ASD that have been shown to hold some but not overwhelming efficacy.

One of the greatest challenges for treatment researchers is to understand why some interventions work well with some children, but not all. The factors that influence the effectiveness of an intervention could be genetic, environmental or biological and an important aim of future research will be to identify these factors so that individualised treatments can be designed.

There have been recent reports of the efficacy of parent mediated interventions to improve social and communication skills, raising the possibility that a parent may become a more formal educator of their child. There are also a number of "home-treatments" that are used by many parents in an attempt to ameliorate their child's difficulties (e.g. special diets; vitamin supplements) but there has been little or no reliable research of whether these treatments are efficacious.

## **2.2. Autism Research in the UK**

Autism researchers in the UK have traditionally been internationally competitive and produced science that has moved the field forward at pace, particularly in researching genetics, neuroscience, cognitive models, the development of standardised diagnostic tools, medical conditions associated with ASD and more recently issues around the ageing population. To understand the causes of ASD, UK genetics research has given us some of the first replicable findings of which chromosomes autism families may share. Studies of post-mortem brain tissue have informed us about the subtle differences in the brains of individuals with ASD and when during development these changes may occur, increasing the chances of scientists identifying pharmacological treatments. The design of diagnostic and screening tools has allowed clinicians and researchers to reach a consensus on definitions of ASD and has led to the UK having the means to develop accurate prevalence data.

There are a number of senior clinical researchers currently working in the UK who have a strong background and training in ASD (a number of whom received training from Professor Sir Michael Rutter in the 1980s). Cognitive psychology (how a person thinks and feels) also has a strong history in the UK arising from Professors Beate Hermelin's and Neil O'Connor's research in the 1970s, and the important cognitive models of Professors Uta Frith and Simon Baron-Cohen in the 1980s and 90s. There are currently a number of small to medium sized autism research centres around the UK, which conduct a broad range of research.

Although there are advances being made in terms of our understanding of the biological basis of autism, funding organisations and clinical researchers need to think more strategically, and beyond the typical two or three year grant period, to identify opportunities and collaborations to translate their research findings into treatments or diagnostics. The Mapping Autism Research report prepared by the Institute of Child Health in 2004 showed clearly the gap between what was being researched (primarily symptoms) and what families wanted (primarily causes and interventions) and although progress has been made this can be accelerated.

The main funders of autism research in the UK are Autism Speaks, Autism Speaks Inc and the MRC. Following the 2001/2002 review of autism research, "ring fenced" money for autism was provided by the MRC and a number of studies were funded, including an Autism Imaging Multicentre Study (AIMS) and a Pre-school Autism Communication Trial (PACT). These MRC grants were to encourage the best science and collaboration amongst partners.

The MRC also held a forward look for autism in January 2009, to review the progress of its previously funded projects and assess what the next research steps should be. Autism Speaks Inc (and its predecessors Cure Autism Now and the National Alliance for Autism Research) have always accepted applications for programmes, projects and fellowships in the UK on a competitive basis. During 2006-2008 twenty projects were funded by Autism Speaks Inc in the UK, totaling \$3m.

## **2.3. Autism Speaks Funded Research**

The projects currently funded by Autism Speaks broadly fall into genetics or neuroscience and have been related to key strategic infrastructure, ground breaking research activities or to capacity building (i.e. Mentor Based Fellowships)

- In 2005 Autism Speaks began working with Professor Johnson at Birkbeck College to scope the potential range, innovation and benefit of a major UK network and research study of children at increased risk for developing autism (i.e. they have an older brother or sister with a diagnosis of ASD). Autism Speaks took the lead in creating a funding consortium and since 2007 the programme has been operational as the British Autism Study of Infant Siblings (BASIS). This study design allows scientists to investigate the very early development of children some of whom will develop typically, and others who will later go on to develop

ASD or a milder form of the condition. The quality and care that the BASIS research team take means that families choose to stay in close contact, increasing the chances of these children participating in future research. Autism Speaks has been instrumental in funding the core infrastructure and research team, giving BASIS the ability to recruit, retain and support participating families and allowing much wider research activities than would otherwise have been possible. This has been significant in leveraging funding from other bodies such as the £1.5m grant made by the MRC to Professor Mark Johnson for the study of cognition in typically and atypically developing babies and toddlers.

- Autism Speaks has been the lead co-ordinating and funding partner on the first Chair of Autism Research at the University of Cardiff. The new Chair (Professor Sue Leekam, April 2009) acts as a catalyst for a new programme of scientific research that will complement the work of the Welsh Assembly Government's ASD Strategic Action Plan, and stimulate growth of autism research in Wales, as well as providing stakeholders and families with additional regional expertise. This new resource is positioned within a University setting that has an established and strong track record in developmental psychology, extensive state of the art brain imaging facilities and reputation for advancing the field of mental health genetics.
- The funding contributed to Professor Monaco at the University of Oxford for Phase II of the international Autism Genome Project is helping to identify those genes underlying susceptibility to ASD. This research will be vital in the quest to subtype groups of individuals within the autism spectrum and may also have value in detecting increased risk for autism as part of clinical management programmes.
- Autism Speaks also funds the Brain Bank for Autism and Related Developmental Research at the University of Oxford. The brain bank is a high value resource since it works in concert with the larger Autism Tissue Program based in the US and is accessible by researchers worldwide (based on favourable peer review). Post mortem brain research is essential in the identification of subtle differences in the brains of individuals with ASD and it is only by the microscopic study of post mortem tissue that scientists will be able to associate genetic and neuroscientific findings. Considering the expense and the need for highly co-ordinated activities, a centralised Brain Bank for Autism means that post-mortem brain research can move forward at a much higher and rigorous level than would occur if scientists obtained brain material through their own individual efforts.
- The Autism Speaks funded functional neuroimaging (fMRI) study at the Institute of Psychiatry in London continues the UK's strong history of twin research and investigates how genetic and environmental factors affect brain development in identical and non-identical twin pairs. This study will provide scientists with information about why specific regions of the brain may have developed atypically, which will be valuable in designing treatments and identifying the brain regions these should be directed to.
- Autism Speaks has also supported the next generation of researchers with a pre-doctoral fellowship in Cambridge to investigate a rarely researched area of the field - why children with ASD are abnormally sensitive to certain sights and sounds – a topic that has potential clinical and educational applications.

## 2.4. Where is the potential for expanding effective autism research in the UK?

Based on the strengths of the UK autism research community and projects/initiatives that are currently funded there are a number of opportunities for Autism Speaks to contribute strategically to making the UK a vibrant and effective centre of excellence in autism research.

- 1) **Developing a sustainable research infrastructure:** This could include: the funding of research networks (such as BASIS) to promote data sharing, best use of resources and collaboration; increasing the number of researchers in the field via fellowships; a centralised means of recruiting research participants to make that process simpler for researchers and families; centralised data repositories to increase the amount of data available for researchers; meetings or conferences to promote networking (always valuable in promoting translation), sharing of data and discussion of research direction. Autism Speaks is strongly positioned to be the catalyst for much of this infrastructure development that may not happen without its encouragement or funding support.
- 2) **Utilising existing and developing new research capacity:** Attracting the brightest and best into autism research and then supporting their careers is vital to energy and creativity. There is a need therefore to support the next generation of researchers and for them to be mentored by the current senior research staff. Autism Speaks is dedicated to increasing the number of fellowships for junior and senior researchers, which is evident for example in both prior financial commitments and the current fund raising support for a new chair in autism research at UCL to build on its existing excellence in cognitive neuroscience.
- 3) **Adding to successful existing collaborative studies:** BASIS (British Autism Study of Infant Siblings), the Brain Bank for Autism, and the PACT and AIMS studies mentioned above have proven to be successful collaborative efforts. Autism Speaks can fund components of these projects to add value or encourage the translation of their findings into clinically meaningful tools (e.g. a larger NHS funded study of PACT). Autism Speaks can both fund directly as appropriate and also look to lever by working together with other funding organisations.
- 4) **Building on the UK's strength in neuroscience and cognitive science:** The upcoming findings from the MRC AIMS study will be extremely informative as to the structural and functional differences in the brains of adults with ASD. The Brain Bank for Autism at Oxford will also stimulate research activity in the field of neuropathology and ASD.

However, there is still a need for neuroscience and cognitive studies of under researched groups (such as individuals with ASD and learning difficulties; children) and the strength of the UK community could allow it to take a lead in the methodology and practice of this research. New neuroscience departments, such as the Wellcome/Sainsbury centre at UCL within close proximity to the existing autism research centres in London, have the potential to increase the capacity for autism/developmental related neuroscience in the UK. It will also be important to build on UK achievements in other areas of developmental neurobiology including stem cell research and to apply this expertise and knowledge to ASD. Autism Speaks will continue to work with other funders to encourage co-operative and collaborative activity.

- 5) **Consulting with the autism community on the direction of research:** The UK has individuals with ASD and families who can provide excellent advice as to the content and direction of autism science. There is also an excellent network of autism advocacy groups and stakeholders, with whom Autism Speaks has good working relationships. Consultation, planning and dissemination of findings to families and stakeholders should become a more centralised theme within grant applications and not simply an add-on.

### **3 Science Objectives**

In drawing up this strategy Autism Speaks has been able to take into account the strategies of major funders in the United States who have done much to advance the field of autism research. These include Autism Speaks Inc, who in 2008 conducted a detailed and extensive analysis of past performance and achievement in the research it historically funded and also looked to the future in terms of identifying what new scientific knowledge is required to move the field towards delivering tangible benefits to families. This analysis and that of the US National Institutes of Health have been highly informative and Autism Speaks has been able to draw on and adopt some of the core objectives identified whilst considering how to target new science and facilitate research efforts specifically in the UK. Thus set out below are what the charity believes are important and achievable scientific goals for UK autism research over the next 5 years and how Autism Speaks can help to achieve them.

Each sub-section is in the form of a brief discussion of a key scientific question, followed by our view (in priority order) of what needs to happen over the next 5 years to address the question, and a prioritised list of specific actions Autism Speaks can take to facilitate this.

Autism Speaks will work with other funders and collaborators in the UK to progress the activities outlined, as one of our key strengths has proven to be our ability to bring together organisations, people and funders. Other high value scientific opportunities may also be considered.

#### **3.1. What causes ASD and how can we use that knowledge to ease the burden for families?**

There are likely to be numerous causes for ASD and it will be a major scientific venture to discover them and determine their impact. It will be vital for scientists to understand about the range of causes and so genetic, environmental and gene x environmental models will need to be developed. By understanding what causes ASD, clinicians will be able to inform families about their risks and provide them with advice and guidance (e.g. through genetic counseling). Learning more about why one child has a different type of ASD compared to another child may well help clinicians in the future to inform families about outcomes for their children and the best interventions can be designed. The recent animal research of syndromes that produce similar behavioral characteristics to ASD (e.g. Fragile X and Rett syndromes) suggests that with specific gene therapies or pharmacological intervention some of the aberrant behavioral characteristics can be reversed.

As a first step, researchers will need to understand how certain causal risk factors affect the body (e.g. does a particular abnormal gene alter the development of a brain region that could ultimately be treated with a specific medication). There may also be a number of protective factors that influence a child's development and these will be vital to the design of effective treatments. The UK is strong in autism genetics research and therefore the overarching aim should be to build on this and look for opportunities to develop strengths in environmental research.

### **Goals**

- 1) Leading UK scientists continue the identification of genetic risk markers for ASD
- 2) The research community enlarges sample collection of genetic and other bio samples, to increase the amount of data available and allow scientists to look more carefully at the range of causal factors
- 3) Researchers include non-genetic or environmental factors (e.g. analysis of maternal antibodies; obstetric reports) as variables in causal research or as mediators in treatment trials
- 4) Clinicians and geneticists work together to explore the feasibility of using genetic findings as diagnostics through clinical settings
- 5) For those genetic risk markers that are clearly identified as causal, scientists develop animal models and treatments are explored. This will provide a model of the effect of abnormal genes on brain development and how to treat this

### ***How Autism Speaks can help achieve these goals***

- 1) Continued funding support of the International Autism Genome Project or similar large-scale genetic collections in the UK
- 2) Fund a feasibility study of how to collect genetic and biological data via collaborative NHS clinical networks. Disseminate findings of the methodological and ethical issues around this type of research
- 3) Fund a workshop on environmental sciences and follow up with a call for proposals on environmental research in ASD
- 4) Look to support or lever funds for a prospective study – perhaps in high-risk samples – that includes measures in pregnancy, neo- and post-natal periods
- 5) Encourage autism researchers to explore environmental factors in their research and environmental science experts from outside the field to conduct autism research
- 6) Once a genetic finding is published and replicated, fund an animal model of how this gene may affect brain development

### **3.2. What are the specific characteristics of ASD that are amenable and relevant to treatment?**

One of the conundrums of autism is that although individuals within the autism spectrum share common social and communication difficulties, they are also diverse and unique. It remains unclear as to what causes a constellation of features (behavioral, cognitive, biological) to come together to form subgroups, or how treatments can be best directed to individuals or subgroups rather than the whole ASD population. A number of questions remain unanswered, including:

- what makes an individual with ASD different from typically developing children (e.g. what are the structural differences in the brains of children with ASD compared to typical children)
- within the autism spectrum what subgroups exist and why (e.g. are there a group of individuals with increased head size and epilepsy)
- what are the genetic and biological differences between individuals who have ASD + LD (learning difficulty) compared to individuals with ASD and average range IQ
- do individuals with ASD and average or above average IQ form a subgroup that have a more distinct pattern of structural brain abnormalities
- what are the genetic, biological and behavioural differences between males and females with ASD?

The overarching theme is to more clearly define what ASD is in terms of biology, clinical features and behavior, so that treatments can be directed to the needs of individuals. The UK's expertise in neurosciences, clinical medicine and diagnosis puts researchers in a strong position to meet these objectives. There should also be a more sophisticated statistical analysis of subgroups within the autism spectrum and the chance to relate these groups to putative genetic and environmental risk factors.

### **Goals**

- 1) A centralised database is established and run by researchers for storing biological, diagnostic, cognitive and behavioral data
- 2) Statisticians begin data mining UK and US data repositories (e.g. National Database for Autism Research) to look for sub groups and phenotype/genotype correlations
- 3) The amount of research into medical conditions associated with ASD (epilepsy, sleep, gut problems, glue ear, anxiety) is increased and treatments developed
- 4) The brain bank for autism in Oxford continues to increase the number of donations, collect rich phenotypic information and stimulate UK neuropathological autism research
- 5) Researchers form a network of neuroscientists, collaborating and meeting once a year
- 6) The amount of research on individuals with ASD and learning difficulties is increased
- 7) More non-neuroscience biological researchers enter the autism field

### ***How Autism Speaks can help achieve these goals***

- 1) Explore the feasibility/hold a meeting on how to centralise phenotypic data collection
- 2) Fund a fellowship/study of statistical modeling and data mining of data
- 3) Consider a future call for proposals on research into medical conditions associated with ASD and how to treat them
- 4) Work closely with the brain bank at Oxford to facilitate research and collaboration with the US Autism Tissue Program; support initiatives such as MRC strategy to access increased numbers of paediatric and control brains; fund neuropathological imaging and genetic studies
- 5) Fund feasibility study of scanning rarely researched groups; look for opportunities to build on the AIMS collaborative neuroimaging network; fund a meeting for the first UK "Autism Neuroscience Network"
- 6) Encourage/engage with scientists from other areas of biological research with the potential to identify biomarkers to enter the autism field

### **3.3. How can we improve early screening and diagnosis, and our understanding of how ASD changes over time, in order to achieve timely and effective intervention?**

Families often have to struggle hard to have their children diagnosed with ASD and typically in the UK it is after some years of visiting different professionals that they get a final answer. The research of high risk infants and the "red flags" for autism that are recommended for use by US paediatricians to screen for ASD at 18 months have great potential clinical value.

The goals should be for diagnosis to be made accurately and early in development so that treatments can be introduced to improve outcomes. It is possible, however, that there are groups who may be more likely to receive a diagnosis when they are older (e.g. secondary school age adolescents with Asperger syndrome), so screening or diagnosis may need to be targeted at different ages. Finally, there is an increasing interest in the UK adult population with ASD and the lack of services they are receiving. Overall, more research is needed into how children and adolescents with ASD change across development (longitudinal studies) and into adulthood, so treatments can be

made age-appropriate and targeted at specific stages of development (e.g. the transition from childhood to adolescence).

The UK has a number of clinicians who have led the field in the development of screening and diagnostic instruments and so they are in a prime position to move the field forward.

### **Goals**

- 1) Researchers bring together the latest thinking on screening for and diagnosing ASD
- 2) Longitudinal research is conducted on the BASIS sample and the factors that affect developmental trajectory and response to treatment are identified
- 3) Clinicians and researchers develop protocols for how early markers and screening can be introduced into NHS clinics
- 4) Scientists identify the genetic, environmental and biological mechanisms underlying why individuals follow different developmental trajectories
- 5) Outcomes for adolescents and adults are investigated and risk and protective factors are identified
- 6) Clinicians are using genetic and biomarkers as part of the diagnostic process

### ***How Autism Speaks can help achieve these goals***

- 1) Fund a meeting on the future of diagnosing/screening ASD
- 2) Continue support for BASIS and "bolt-on" projects such as iBASIS (intervention in high risk infants)
- 3) Arrange meetings with NHS/Dept of Health staff to discuss how screening tools could be developed that meet the criteria for use within clinical settings
- 4) Stage a call for proposals or lever funds from MRC for the revision of existing or development of new diagnostic protocols
- 5) Fund studies that allow researchers to conduct longitudinal research or revisit existing autism cohorts
- 6) In light of the growing UK interest in older adolescents and adults with ASD, fund research of the risk and protective factors for these individuals and appropriate diagnostic assessments (e.g. self report measures)
- 7) Encourage other organisations to develop training protocols for clinical staff (GPs, health visitors, paediatricians) on how to recognise ASD.

## **4 The Future**

### **4.1. How will Autism Speaks measure the success of the grants it funds?**

This document sets out the strategic context for future calls for proposals and selection of projects for funding by Autism Speaks. It will be just as important however for Autism Speaks to set up measures to assess the value of the research it has funded. This value can be measured in many ways and as the projects now being funded are completed researchers will be expected to help us by filling out a report briefly describing for example their findings, what changes had to be made to their study design and why, what the potential long-term benefits are for people with ASD and their families, the number of presentations/publications (scientific and lay), collaborative meetings that were either run or attended as part of the project, who was employed on the grant and will they continue in autism research, what the study team's future plans are and how Autism Speaks could further assist them in meeting their targets. Such a report could be descriptive and has the potential for Autism Speaks or an independent organisation to rate each project in the context of the priorities set out in the science strategy. Researchers could also be contacted again after a period of time has elapsed to assess the long-term benefits of Autism Speaks funding.

### **4.2. Future versions of the science strategy**

Autism Speaks will update this 5-year science strategy annually and reflect on particular advances in the field or significant shifts in the direction of the science. In 2012 the Trustee Board of Autism Speaks will consider to what extent scientific development sponsored by Autism Speaks has met its objectives, with recommendations to be made for the science strategy spanning the period 2014-2019.

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