

## Autism Spectrum Disorders, Risk Communication and the Problem of Inadvertent Harm

**ABSTRACT:** Autism spectrum disorders (ASDs) are an issue of significant and growing importance to the field of public health. The prevalence of ASDs is rising, and these disorders significantly impact the quality of life of affected persons and their families. Though the etiology of ASDs has long been poorly understood, in recent years, studies are revealing genetic and environmental risk information about ASDs, with much more risk information expected to follow from scientific studies currently underway. The availability of this risk information raises questions about whether and how it should be communicated to individuals, families, and the public at large. One ethical issue of particular concern with ASD risk communication is the possibility that it may cause inadvertent harm to risk message recipients. Here we review the emerging picture of ASD risk, discuss some ways in which it may lead to inadvertent harm, and suggest some future directions for risk communication research and practice that might help to address this issue.

### 1. INTRODUCTION

**A**utism spectrum disorders (ASDs) are an issue of growing public health significance. This set of neurodevelopmental disorders, which includes autistic disorder, Asperger syndrome, and pervasive developmental disorder not otherwise specified (PDD-NOS), is characterized by abnormalities in one or more of the following domains: language use, reciprocal social interactions, and/or a pattern of restricted interests or stereotyped behaviors. Prevalence estimates for ASDs have been increasing over the past few decades, with estimates at ~5/10,000 in the 1960s, and current estimates as high as 1/88 (Newschaffer et al. 2007; CDC 2012).<sup>1</sup> While ASDs encompass a wide range of phenotypes and degrees of severity, the disorders can be difficult or even devastating for affected persons

and their families. Persons severely affected by an ASD may not be able to use language, interact with others, or perform basic daily functions such as grooming or dressing, thus making it impossible for them to live an independent life or to pursue many of the experiences afforded to most persons. In addition, ASDs are associated with intellectual disability in approximately 40% of cases (Volkmar and Pauls 2003; Newschaffer et al. 2007; Levy et al. 2009). Persons less severely affected by ASDs may not experience these specific difficulties, and may even view their ASD as conferring some advantages. In some cases, ASDs may be associated with special skills and/or above-average cognitive ability. Nonetheless, ASDs can present challenges for all affected individuals, particularly as relates to social interactions with others.

ASDs also significantly impact the families of affected persons. On average, parents or other caregivers of persons with ASDs spend 6 times more on healthcare expenses as compared to typically developing children, and many families experience financial hardship in the process (Ganz 2007; Peacock et al. 2012; Sharp and Baker 2007). Similarly, providing care and support for persons with ASDs may require significant time investments from caregivers, as well as the restructuring of caregivers' lives such that they may be required to forego personal interests. ASDs may be viewed as negatively impacting or even "stealing" the relationship that exists between parents and their affected children, and caregivers often report significant stress and anxiety associated with caring for an ASD-affected child (Lee et al. 2008; Webster et al. 2008; Estes et al. 2009; Hines et al. 2012).

Until very recently little information was available regarding the cause(s) of ASDs. Early twin studies (e.g., Bailey 1995) suggested that ASDs were highly heritable, but more specific information on candidate genes awaited the sequencing of the human genome and the further development of genetic screening technologies. An article on genetic counseling for ASDs published in the late 1990s noted that at that time, counseling was limited to the discussion of recurrence risks, since candidate genes and environmental risk factors had yet to be identified (Simonoff 1998). Since the mid-2000s, however, a sizeable and ever-growing number of studies have investigated potential genetic and environmental risk factors for ASDs, the latter assuming increasing importance in light of recent evidence that the environment plays a larger role in the development of ASDs than was previously thought (Hallmayer et al. 2011).

With this recent rise in ASD risk information comes the challenge of developing ethically defensible risk communication policies in the research,

clinical, and public health settings. Risk and health communication in any context raises ethical issues (Johnson 1999; Guttman 1996, 1997; Guttman and Salmon 2004), but as regards ASDs a number of special issues may apply. Much of the ASD risk information currently available is complex and of uncertain importance, exacerbating bioethical concerns about autonomy, nonmaleficence, and other issues that would otherwise be present but perhaps less difficult to navigate. Furthermore, ASD risk is often communicated to the prospective or actual parents of a child with an ASD, and therefore affects the interests of multiple parties. To date, little has been written about ethical issues in autism risk communication, and many empirical questions remain unresolved. In the present paper we explore some of these ethical issues, with a particular emphasis on the ways in which ASD risk information might lead to inadvertent harm, the ways in which this harm might be mitigated, and key empirical and normative issues for future research. Although this paper avoids determinate conclusions about ASD risk communication policy, given that inadvertent harm is a plausible outcome of ASD risk communication in our view, we do raise concerns about the wisdom of routine communication of all scientifically valid ASD risk information, either to individuals or the public at large, when the potential relevance of this risk information to individual decision-making is unclear.

## 2. ASD RISK COMMUNICATION AND THE QUESTION OF PATHOLOGY

A major issue in the ethics of ASDs concerns the prudential disvalue of ASDs—that is, the impact of ASDs on the quality of life of persons who have them—and, derivatively, the question of whether ASDs should be considered “diseases” or “disorders” (Barnbaum 2008; Pellicano and Stears 2011). The term “risk” generally denotes an undesirable outcome, and some members of the autism community (often self-described as part of the “neurodiversity movement”) might question the entire concept of “ASD risk communication” on the grounds that some or all forms of ASDs are, in their view, inappropriately pathologized.

Whether some or all ASDs should be considered pathologies is an important and complicated question, and one that has received relatively little philosophical attention. In addition to the perspectives of persons in the autism community, a good amount of work remains to be done in prudential value theory in order to adequately come to grips with this issue. Accordingly, we cannot substantially enter this debate in the present analysis, which focuses primarily on the possibility that ASD risk com-

munication could result in inadvertent harm to risk message recipients. We will state our commitment that in their severe forms, ASDs negatively impact affected persons, though we recognize that some persons in the autism community will consider even this judgment to be contestable. In addition, we recognize that as concerns higher-functioning persons with ASDs, judgments about their medicalization are significantly more controversial from a philosophical standpoint, and may even be considered dubious.

Since ASDs can confer benefits as well as present challenges to affected persons, their status as undesirable outcomes might be more equivocal. For example, since early diagnosis of ASDs might lead to better outcomes as concerns such variables as IQ, language use, and social interaction,<sup>2</sup> we might value ASD risk communication if it facilitates early diagnosis and thus helps children to cope with the challenges of their ASD, without necessarily implying that their ASD is an unequivocally bad thing for them; we can acknowledge that ASDs confer benefits without denying that they also present challenges or impose deficits that we are justified in trying to ameliorate. Finally, we emphasize that the prudential impact of ASDs on affected persons is a distinct issue from the moral status of persons affected by ASDs, or the moral permissibility of selective abortion in cases where the future child is likely to have an ASD. Though these issues may sometimes be related in moral analyses (see, e.g., Barnbaum 2008), judgments about prudential value do not entail judgments about these latter two issues.

In a broad way, social views about whether ASDs always constitute pathologies will affect the likelihood of inadvertent harm, since such views seem likely to condition persons' experience of anxiety, guilt, and stigmatization. Nonetheless these two issues, though related, are distinct, and we need not definitively settle questions about medicalization and pathology in order to explore the moral ramifications of inadvertent harm.

### 3. ASD RISK INFORMATION: WHAT IS KNOWN TO DATE

The scientific literature on autism spectrum disorders is already populated with a sizeable number of studies examining ASD risk factors, with many more studies currently underway. The result of all of this scientific activity is a large and growing amount of information that communicators must take into account when discussing risk with research participants, patients, or the general public. While a comprehensive review of known

ASD risk information is not possible here, we highlight some key developments that influence ethical considerations in ASD risk communication.

A small number of twin studies (e.g., Bailey 1995) performed in the 1970s–1990s suggested that a high degree of genetic liability existed for ASDs, with estimates at about 60% heritability for autistic disorder and 90% for the broader autism phenotype. However, these estimates have recently been revised downward, with a recent and large twin study conducted in the United States using current diagnostic standards estimating heritability for both strict and broad autism phenotypes at less than 50%, suggesting a potentially greater role for environmental risk factors. The authors of this study speculate that a shared environment amongst many twins inflated estimates of heritability in previous twin studies (Hallmayer et al. 2011). The fact that dizygotic twin concordance in this study was substantially larger than non-twin sibling recurrence risk as documented in another large study completed under current diagnostic standards (Ozonoff et al. 2011) suggests that the prenatal period may be particularly important with respect to environmental influences.

Research on identifying specific genetic risk factors for ASDs is making steady progress, but at present is characterized by some degree of uncertainty regarding the number of genes involved, which specific genes are involved, the causal effect sizes that individual genes exert, and whether these effects rely on gene–environment interactions. Early estimates for the number of genes implicated in ASDs were as low as 2–4 (Simonoff 1998), but the number of genes or chromosomal regions associated with ASDs that have been identified to date now numbers well over 100 (see, e.g., Rosenfeld et al. 2010; Scherer and Dawson 2011). Furthermore, many of the genes identified to date are rare and not fully penetrant. Even when fully penetrant, too few persons carry these genes to account for the number of persons diagnosed with ASDs; hence there is still much “missing heritability” regarding genetic risk factors for autism (Pinto et al. 2010; Rutter 2011).<sup>3</sup>

Replication is another concern when considering ASD genomic risk factors. For example, a recent review of the role of common genetic variants in ASDs noted a lack of replication between different studies. This could be because an initial finding is spurious, but it could also be due to differing screening methodologies (e.g., different DNA probes) or the fact that replication studies usually require a larger sample size than the initial studies they are trying to confirm (Devlin et al. 2011). Studies of

rare variants also face considerable challenges with respect to replicability.

For some time it has been known that approximately 5–15% of ASD cases are “syndromic” cases in which a known genetic cause exists. Examples of such syndromic causes of ASDs or ASD-like conditions include Fragile-X syndrome, Rett syndrome, and Prader-Willi/Angelman syndrome (Caglayan 2010). In all instances only a portion of individuals with these syndromic conditions will develop an ASD. Therefore, though the ASD risk associated with these conditions is often substantial, none of these causes appear to be necessary and sufficient. More recent research has identified a number of additional genetic mutations that appear to be strongly causative of ASDs, insofar as such mutations are rarely or never found in non-affected controls. For example, mutations in the SHANK3 gene were found in a small number of individuals with ASD and not in any normal controls; nor were these mutations seen in non-affected parents (Betancour et al. 2006). Similarly, duplications at chromosome 15q11-13 are associated with ASD in approximately 85% of individuals who have them (Cook and Scherer 2008). However, these highly penetrant mutations account for only about 1% of non-syndromic ASD cases (Rutter 2011). In many cases genetic abnormalities associated with ASDs are found in non-affected individuals, and furthermore the effect size of identified ASD risk factors is frequently small. Instead of constituting sufficient causes of ASDs, such risk factors merely confer *susceptibility*, which in some cases may not be more than a few percentage points of risk (Berkel et al. 2010; Devlin et al. 2011; Pinto et al. 2010; Rosenfeld et al. 2010; Rutter 2011).<sup>4</sup> Hence in most cases the causation of ASDs conforms to a “threshold” model whereby multiple genetic and/or environmental risk factors exert small to moderate causal effects.

An additional complicating factor when interpreting genetic risk factors for ASDs is that candidate genes are often not specific for an ASD phenotype, also being implicated in intellectual disability with or without ASD, schizophrenia, attention-deficit and hyperactivity disorder/hyperkinetic disorder, and epilepsy (Rosenfeld et al. 2010; Rutter 2011).

For a number of decades the fact that environmental exposures could cause autism has been accepted, through evidence developed related to the study of congenital exposure to rubella and cytomegalovirus, as well as exposure to known powerful teratogens (see Yamashita et al. 2003; Dufour-Rainfray et al. 2011). However, progress in identifying additional specific environmental risk factors where exposure is still prevalent today has lagged somewhat behind progress regarding genetic risk factors. In

part this was due to the initial emphasis placed on gene discovery following the early twin studies where heritability seemed extremely high, coupled with the advances in molecular biology that have made full genome scans routine. Discovery of environmental risk factors also shares the common challenge faced by genomic research in that individual risk factors, environmental and genomic, are likely components of multiplex causal mechanisms. Recently epidemiologic studies have replicated the finding that autism risk increases with advancing parental age and that autism risk is higher among children born preterm and/or at low birth weight (Durkin et al. 2008; Lampi et al. 2012). The extent to which these findings are related to environmental, genetic, or epigenetic mechanisms remains to be determined. Recent studies have provided some initial evidence around new environmental factors including organophosphates, acetaminophen, selective serotonin reuptake inhibitors (SSRIs), and air pollution, for example (Herbert 2010; Landrigan 2010; Croen et al. 2011; Volk et al. 2011). As with many ASD-implicated genes, the effect sizes of environmental risk factors may be modest.<sup>5</sup>

Complicating matters in the search for environmental risk factors is that environmental effects are likely influenced by genetic susceptibility. For example, maternal prenatal vitamin use may confer protection against a child's subsequent development of ASD, but the level of protection has been shown to vary depending on genotypes associated with one-carbon metabolism (Schmidt et al. 2011). In addition, specific variations in the *reelin* gene are positively associated with ASD in areas where organophosphate (OP) pesticide use is high, but not in areas where OP use is low (D'Amelio et al. 2005). The study of gene-environment interactions is extremely challenging, as it requires large sample sizes with high-quality genetic and environmental data.

Overall, the present situation with regard to our etiologic knowledge about ASDs is promising, but also complex, uncertain, and rapidly changing. This complexity and uncertainty are likely to exacerbate concerns about inadvertent harm that are present in many risk communication contexts, but which are particularly difficult to navigate as concerns ASDs. Furthermore, the complexity and uncertainty of ASD risk information may in some circumstances call into question the fundamental value of risk information that motivates risk communication in the first place. We discuss these issues in the following section.

## 4. RISK COMMUNICATION AND INADVERTENT HARM

ASD risk communication may occur in any number of contexts, for example doctor–patient interactions, mass public health communications, scientific journal publications, academic meetings, and popular news media and Internet articles. Risk communication may also serve a number of ethically important purposes, including the substantive improvement of decisions, fostering autonomous choice and showing respect for persons, providing explanation, and building trust and community. However, these positive aspects of risk communication should be considered against some possible negative effects, one of the most prominent being the possibility of inadvertent harm to the individuals and populations with whom risk communicators engage.

Numerous ethical treatments of risk and health communication have discussed its ability to cause harm, regarding such topics as breastfeeding, contraception, food and alcohol consumption, smoking, and others (Guttman 1996, 1997; Guttman and Salmon 2004; Bayer 2008). These harms may include (1) arousing fear, worry or anxiety amongst message recipients; (2) blaming or stigmatizing message recipients, or causing them to feel guilty (outcomes associated with moral culpability); and (3) inadvertently leading message recipients to take actions that are not in their interests, for example by acting upon misunderstood risk information. All of these adverse outcomes are relevant to ASD risk communication, and in addition we recognize a fourth category of adverse outcome that is relevant to ASD risk communication in particular: (4) the alteration of individuals' identity (in their own view or in the view of others) in ways that are detrimental to them (e.g., by restricting their ability to self-determine).

Moral concern about harm arising from risk communication can be subsumed under a principle of nonmaleficence, which holds that we ought not to harm others. Nonmaleficence might also be specified to hold that we should prevent harm from occurring or otherwise try to mitigate harm; as well as that we should not impose risk upon others. While nonmaleficence is a core moral principle that is accepted by virtually all normative ethical theories, disagreements about its scope and degree of stringency can arise. As concerns risk communication, it might be argued that transient or very minor anxiety or guilt, for example, ought not to be considered morally salient harms—either because they are too minor to really be considered a setback to one's interests, or because it is natural and appropriate for people to experience these emotions in response to receiving risk information. Furthermore, it might even be argued that these emotions, when

properly managed, will make it more likely that people will engage in appropriate (however this is defined) risk-responsive action.

We can allow that some of the adverse outcomes discussed below—principally anxiety and guilt—might at times be of small enough magnitude so as not to engender much moral concern, while also recognizing that anxiety and guilt can be significant to persons experiencing them. Furthermore, stigmatization and changes to a person's identity seem significant whenever they occur. Hence there is good reason to consider the possibility that risk communication can result in harm, as well as what risk communicators ought to do about this possibility.

At times, risk communicators have deliberately employed tactics that can be considered harmful, because they believe that such tactics are the most effective way to advance a public health goal. For example, risk communicators have purposefully used stigmatizing tactics in antismoking campaigns, and it has been argued that the social stigmatization of smoking was instrumental in reducing the prevalence of smoking behavior (Bayer and Stuber 2006). Similarly, anxiety- or fear-inducing messages might be deliberately used so as to spur an audience to action. In many cases, however, harms arising from risk communication are inadvertent—that is, unintended. To date, most ASD risk communication has been aimed primarily at providing people with information, rather than implicitly or explicitly persuading them to do something. Thus any harms arising from ASD risk communication would, at the present time, be more likely to be inadvertent than deliberately caused.

Furthermore, the ethical defensibility of deliberate harm-inducing tactics in risk communication is a point of controversy. Fear, anxiety, or guilt may in some cases effectively spur an audience to act in ways desired by risk communicators. It is also morally appropriate for people to experience guilty feelings at times (e.g., after acting in a way that is genuinely irresponsible). Nonetheless, even where persuasive risk communication is concerned, it is possible to persuade an audience to behave in a certain way without playing upon anxiety or fear, or invoking notions of culpability. Given the controversy surrounding the deliberate use of such tactics, we believe it most defensible for risk communicators to default to a position where they are not used without an exceptional justification. Therefore we focus on inadvertent harm in the present analysis, leaving the ethical analysis of deliberate harm-inducing tactics for another occasion.

One further note of qualification is in order. Ethical arguments often presuppose or depend upon certain empirical claims regarding the effects

of particular actions, practices, or policies. This is certainly true as concerns ASD risk communication, since the possibility of inadvertent harm may influence whether, how and/or when risk information is communicated. We recognize the importance of directly substantiating empirical claims whenever possible; however, this is not always possible, and as regards ASD risk communication specifically there are few direct empirical data regarding the occurrence of harmful outcomes. Despite this limitation, we believe that a plausible argument can be made that ASD risk communication is likely to result in inadvertent harm, since both conceptual considerations and empirical data from related domains support this conclusion.

Furthermore, while direct assessment of these possible outcomes is warranted in future empirical research, it seems ill advised to postpone the consideration of appropriate policy in ASD risk communication until “all the data are in.” Aside from the facts that science is never finished and that data are not self-interpreting (both of which undermine the notion that clear verification or falsification of empirical claims will always be possible), many if not most of the adverse outcomes discussed below are not trivial in nature. Given that risk communicators need to adopt *some* stance with respect to the large amount of ASD risk information currently available and forthcoming, it seems best to pay close attention to the possible adverse effects and ethical dimensions of such information, even if at times this attention is limited to informed speculation.

We also acknowledge that both the likelihood of inadvertent harm and risk communicators’ responsibilities with respect to preventing or mitigating inadvertent harm *may* differ between different risk communication contexts (at this point in our analysis we cannot confidently state a conclusion one way or the other). However, we believe that the types of inadvertent harm discussed below are sufficiently plausible across a variety of risk communication contexts so as to enable discussion at a general level; therefore our comments are intended to apply to all ASD risk communication contexts except where otherwise specified.

#### 4.1. *Anxiety and Worry*

Perhaps the most intuitively plausible adverse outcome of ASD risk communication is that it will result in worry or anxiety about the future occurrence of an ASD. The concept of risk signifies a possible harm or adverse outcome, and in light of risk’s association with harm and its inherent uncertainty, it makes sense that people may feel anxious when learning about their exposure to a particular risk. For example, parents of infants or

young children who have genetic testing performed before a firm clinical diagnosis of ASD may be anxious about whether their child will develop an ASD, as might prospective parents or pregnant women who have such testing performed.<sup>6</sup> Similarly, a pregnant woman taking SSRI antidepressants might experience anxiety because of the ASD risk associated with these medications (see Croen et al. 2011). While many investigations of anxiety *in* individuals with ASDs can be found in the academic literature, there appear to be few data available regarding ASD-related anxiety or worry in prospective parents, pregnant women, or parents of very young children not yet of sufficient age for diagnosis. However, examination of popular news media, blogs and other Internet resources suggests that ASDs are a significant source of concern for many persons. Empirical data from other domains, such as breast cancer, also show that risk communication can cause anxiety, sometimes significantly so (see, e.g., Tzeng 2010; Crotser and Dickerson 2010), though it should be emphasized that these data can be contradictory and difficult to interpret at times.<sup>7</sup>

It stands to reason that risk anxiety will depend on the nature of the harmful outcome itself, in addition to the probability that the outcome will occur. Some outcomes are feared or disvalued more than others, and indeed risk perception research indicates that most persons are more concerned about risks when the risk relates to a “dreaded” outcome (Finkel 2009). As concerns ASD risk communication, the significant negative impact that ASDs can have on affected persons and their families may make ASDs a particularly concerning topic for many persons. The pediatric onset of ASDs might exacerbate this concern, since children’s vulnerable nature often evokes an especially protective response and increased anxiety about risk (Scott et al. 1998), and since childhood-onset disorders can disrupt both parent–child relationships and the trajectory of normal childhood in ways that deserve special note. Empirical studies show that parents of children, adolescents, and adults with ASD often experience significant stress, anxiety, and depression (Lee et al. 2008; Webster et al. 2008; Rao and Bidel 2009; Estes et al. 2009; Hines et al. 2012). To the extent that the experiences of families affected by ASDs become more broadly known amongst the general public, they may contribute to anxiety amongst persons who receive information regarding the risk of future ASD development.

A few additional considerations support concern about ASD risk communication causing anxiety. First, there is some evidence that risk anxiety may be worse for persons with a low tolerance for uncertainty (see, e.g., Crotser and Boehmke 2009). Many of the ASD risk factors identified thus

far are of small to moderate effect size, and uncertainty seems to be more pronounced regarding such risks. If the probability of an event is 90% or 0.1%, then a person can understand that the event is either very likely or very unlikely to happen to them. On the other hand, it seems comparatively more difficult to translate a risk of 5% or 10% into individually meaningful terms.

Second, as already discussed many ASD risk factors are not specific for a particular phenotype: they may be associated with mild or severe impairments in core ASD domains, as well as other conditions such as schizophrenia or attention-deficit and hyperactivity disorder (ADHD). This lack of phenotypic specificity means that not only will persons who receive this risk information not know whether their child will develop an ASD, they also will not know whether an ASD that might develop would be mild or severe. This lack of specificity may potentiate risk anxiety: while highly functioning persons may view their ASD as conferring benefits, for severely affected persons the prudential value of an ASD is more questionable, and when risk information relates to an uncertain phenotype parents may fear the worst.

A third consideration is more philosophical, and relates to how we understand the concept of “risk” itself. Numerous commentators in the philosophy and sociology of risk have argued that scientists and nonscientists tend to define risk differently. In the sciences, an “event-predicting” conception of risk dominates: risk is any probability of harm. However, most persons (cultural groups, social institutions) do not conceptualize risk in this way. Instead, they conceptualize risk in a “problem-classifying” way. On the problem-classifying view, risk is not *any* probability of harm. Rather, the concept conveys some situation that deserves special attention in light of additional moral considerations. To wit, Paul Thompson has argued that “the concept of risk functions as a category for organizing and prioritizing the expenditure of deliberative resources. Those things that are classified as risks are possibilities that demand attention and judgment, while those that are not classified as risks are left on automatic pilot, so to speak. . .” (Thompson 1999, p. 498). Furthermore, in ordinary language, “risk” is morally salient in two dimensions: it conveys a possibility of harm or loss, but it also implies moral responsibility, whereas the event-predicting sense of risk does not. The disparity between the event-predicting and problem-classifying sense of risk may have numerous upshots for risk communication. As concerns anxiety, it implies that risk communication may have the effect of moving a particular probability

of harm from the nondeliberative realm to the deliberative realm, as opposed to simply adding another probability of harm to a decision-making calculus that already focuses attention on the situation in question. By creating risk awareness where none existed before, risk communication may create anxiety and worry in and of itself, independently of the “magnitude” of a risk, and indeed preliminary empirical data seem to support this supposition (Lister et al. 2002).

#### 4.2. *Guilt, Blame, and Stigma*

A second category of adverse outcomes includes guilt, blame, and stigma. These outcomes are related in the sense that all concern judgments of moral culpability. Many of the conceptual considerations discussed above also explain why risk communication could result in guilt, blame, and/or stigma. One of the most fundamental moral principles is *nonmaleficence*—that we should not harm others—and this principle might be extended to risk-imposing actions by derivation of risk’s connection to harm. Risk-imposing actions may therefore be viewed by self or others as morally indefensible or irresponsible. For example, just as a woman taking SSRIs during pregnancy may feel anxious about the possibility that her child will develop an ASD, she may also feel guilty because she is “imposing” risk on her child—even more so if her child subsequently develops an ASD. *Mutatis mutandis* for other actions (e.g., reproductive decisions) that are associated with an increased risk of ASD.

When risk or health messages explicitly or implicitly persuade an audience to take a certain action or adopt a particular belief, risk communication becomes moralized in an additional dimension. Such persuasion creates an explicit or implicit “ought,” and when a moral obligation is created (or reinforced) in the minds of the audience, guilt, blame, and stigma seem more likely when persons do not or cannot comply with the message. For example, Guttman and Salmon (2004) describe how pro-breastfeeding messages have been shown to induce feelings of guilt and shame amongst low-income mothers whose life circumstances made breastfeeding difficult. At present, most ASD risk communication is nonpersuasive, but this may change. For instance, it is easy to imagine a public health campaign that identifies a number of environmental risk factors for ASD and tells people what they can (or should) do to reduce their or their child’s risk. To the extent that most persons conceptualize risk in a “problem-classifying” way, even nonpersuasive risk communication may implicitly create a sense of moral responsibility that could result in guilt.

The same sense of moral responsibility that may create guilty feelings may also result in blame or stigmatization of persons who are viewed as acting irresponsibly. For example, persons who expose their children to known environmental ASD risk factors, or who choose to reproduce knowing that they carry ASD-associated genetic variants, may be blamed or stigmatized by others who learn of these decisions and regard them as inappropriate. While such outcomes are concerning in their own right, they are particularly concerning in light of the fact that blame or stigmatization may occur in the absence of rational social discussion about *when* people are responsible for risk-bearing actions. Since almost everything a person does is associated with some probability of harm, it will not do to say that we are morally responsible for imposing any probability of harm—such impositions cannot be completely avoided. And since many ASD risk factors identified to date are of small effect size, it may well be the case that some actions associated with ASD risk are not actions that we should regard as morally culpable; this will depend on any number of factors, such as whether the risk can be avoided and at what cost.

In addition to these conceptual considerations, empirical and historical data also indicate that ASD risk communication may plausibly result in guilt, blame, and stigma. Many parents of persons with ASDs report feelings of guilt, either because they believe they have contributed causative genes, or because other persons have implicated the external environment (i.e., parenting-related decisions) as a contributory factor regarding their child's ASD (Mercer et al. 2006; Hines et al. 2012). While these findings have principally been documented in parents of a child or adult already diagnosed with an ASD—itsself one context for risk communication—there is every reason to think that similar feelings of guilt may attend to risk communication concerning the possible future development of an ASD. Guilty feelings have also been documented amongst family members who undergo genetic testing and test negative for disease-associated genes, since they may feel that they have unfairly “escaped” the burden of disease that their loved ones must live with (see, e.g., Crotser and Dickerson 2010).

Blame and stigmatization loom large in the history of ASDs. Despite a lack of empirical evidence to support the claim, in the 1950s and 1960s the leading view of ASD causation was the “refrigerator hypothesis,” which held that cold and detached parenting, and not innate biological factors, caused a child's ASD (Baker 2008). Hence parents, and particularly mothers, were blamed for causing autism in their child. This view still holds sway in certain countries, for example France, and anecdotal

accounts from parents in the United States indicate that some physicians in the United States may still express this view. While views about the causation of ASDs are increasingly shifted towards genetic causes (Mercer et al. 2006), to the extent that the “refrigerator hypothesis” has left a legacy it may contribute to stigmatization in the context of environmental risk. In addition, Farrugia (2009) has documented that parents of children with ASDs are often stigmatized on account of their child’s behavior in social situations. This is different from stigmatizing persons for risk-related behaviors, but it shows how social judgments about deviancy and perhaps parenting choices may relate to ASDs. It is a short and plausible step from this kind of stigma, to stigma consequent to judgments that certain risk-imposing behaviors constitute irresponsible parenting.

Genetic determinism may also contribute to guilt, blame, and stigma. In its broadest sense, “genetic determinism can be loosely defined as the view that genes (genotypes) cause traits (phenotypes),” but since this broad definition is almost trivially true, a better taxonomy would distinguish between strong, moderate, and weak genetic determinism. *Strong* genetic determinism holds that “gene G almost always leads to the development of trait T. (G increases the probability of T and the probability of T, given G, is 95% or greater).” *Moderate* genetic determinism holds that “more often than not G leads to the development of T. (G increases the probability of T and the probability of T, given G, is greater than 50%).” *Weak* genetic determinism holds that “G sometimes leads to the development of T. (G increases the probability of T, but the probability of T is still less than 50%)” (Resnick and Vorhaus 2006).

There are a few candidate genes that may be strongly determinative for ASDs, though even here one must be careful since common environmental exposures or epigenetic factors may also come into play. However, in many cases ASD-implicated genes are best described as weakly determinative, and the relevant concern is that they will be viewed as strongly determinative. If this is the case, then risk message recipients will be more likely to experience guilt, or be blamed and stigmatized, for “causing” their child’s ASD. There is some empirical evidence that adding genetic information to risk messages can have a deterministic effect, even when overall risk is conditioned by both genetic and environmental factors, though the general extent of this deterministic effect in genetic risk messages is still unclear (Smerecnik et al. 2009; Smerecnik 2010). As concerns ASDs specifically, a recent survey of parents’ views about causes indicated that 25% of participants believed that ASDs were caused by a single gene (Mercer et

al. 2006). Genetic determinism has also been raised as an issue in other specific risk communication contexts, including breast cancer and type-II diabetes (Haga 2009; Crotser and Boehmke 2009).

#### 4.3. *Alterations of Identity*

A third kind of adverse outcome that may occur consequent to ASD risk communication is that genetic risk information may alter a person's self-identity, or their identity in the eyes of others, in ways that are ultimately detrimental to them. Where genetic risk factors confer only partial liability, such risk factors cannot serve as a valid substitution for a clinically diagnosed ASD. However, the use of genetic screening technologies such as chromosomal microarray in the prenatal, neonatal, or very early life setting would mean that genetic risk information would be available before a firm clinical diagnosis of ASD could be made, the result being that children might be labeled as "having an ASD" based on predispositional genetic information alone. Since not all children who bear such genetic markers will go on to develop an ASD, some children might be inappropriately labeled, especially in light of tendencies toward genetic determinism. This could negatively impact children in a few ways.

First, the provisional ascription of an "ASD" identity to young children prior to a firm clinical diagnosis might cause these children to be "placed under a microscope" in terms of scrutiny for ASD-related behaviors; this level of scrutiny may not be good for children in terms of their development (e.g., by problematizing potentially normal behaviors). Second, identity alterations *per se* may be viewed as ethically problematic insofar as they restrict the options available to individuals in terms of their lived identities. Self-determination is a core moral good, and "labeling" that restricts the identities available to a person can be viewed as a moral bad and a violation of a child's "right to an open future," a notion philosophers have invoked to capture the idea that the future autonomy of children is held in trust by the adults who care for them, and that it is wrong for parents (guardians, etc.) to act in ways that detract from a child's ability to make future autonomous choices (Barnbaum 2008).

Defenders of prenatal or early life genetic screening for ASDs might point out that it is just that—screening—and that no child will be diagnosed solely on the basis of predispositional genetic information. However, it is plausible to think that for some "borderline" cases, genetic information may make a difference to whether a child receives a diagnosis. Excessive scrutiny of children because of their genetic profile may also result in dif-

ferent parental reports of a child's behaviors in the home environment. Finally, even if a child who carries certain genetic risk factors were not to eventually receive a diagnosis from a physician, their families might nonetheless persist in ascribing an ASD identity to them. Disease identities are not simply imposed by the medical establishment; they are also actively constructed by individuals, families, and cultural groups (Bagatell 2007). It may be naïve to think that the widespread use of genetic screening technologies will not result in any changes to the way we view ourselves simply because in the ideal world, such screening tools would not be considered meaningful unless confirmed by a clinical diagnosis that would have eventually been made anyway.

Finally, ASD risk communication could result in our collective identity being altered in undesirable and ultimately indefensible ways. A principal concern expressed by self-advocates and members of the neurodiversity community is that it is inappropriate to broadly pathologize ASDs; that not all aspects of the autism spectrum should be described as "disorders." Should genetic risk communication to prospective or expecting parents commonly result in decisions not to reproduce or to terminate a pregnancy, it could have the effect of removing or significantly decreasing certain personality types in the population. It is too early to know how likely this is to occur. Though the only guidelines for interpretation of chromosomal microarray screening for autism exist for postnatal applications (Miller, David, et al. 2010), there is still ongoing debate about the clinical utility of these tests in this setting (Kaminsky et al. 2011), and, despite that, these tests are being increasingly used in the prenatal setting (Singer 2012). It is known that as concerns other prenatally discovered neurodevelopmental disorders such as Down syndrome, positive test results often lead to pregnancy termination (Skotco 2009). The present inability to associate genetic ASD risk factors with specific phenotypes *may* increase the possibility of selective abortion after receiving ASD risk information prenatally, since prospective parents will not know how severely their future child might be affected. The disposition of this issue obviously depends on the morality of both selective abortion and abortion more broadly, but it also depends on arguments about the prudential value of a life with an ASD. We cannot enter these debates here, but do emphasize that even if abortion is deemed morally permissible in some circumstances, broad reproductive changes consequent to ASD risk communication could result in inadvertent harm, because pregnancies might be terminated on account of an inappropriately pathologized condition; because the future child would not in fact have

developed an ASD; or because selectively terminating pregnancies on account of ASD risk information would decrease the diversity of personality types in the world in a detrimental way. This further underscores the need for additional philosophical work concerning neurodiversity.

#### *4.4. Taking Actions Contrary to One's Interest*

Finally, for a number of reasons risk communication may result in individuals taking actions not in their interest. A large amount of empirical research in risk perception indicates that many persons (sometimes including scientists) have difficulty understanding numerical and probabilistic information, and/or may display certain cognitive biases when it comes to assessing probabilities (Rector 2008; Finkel 2009). If people misunderstand risk information they may make different decisions than if they understood it properly. For example, if people misunderstand the nature of a particular genetic risk, then they may make reproductive decisions that are ultimately contrary to their values or preferences. Overestimation of general recurrence risks, which has been documented as regards ASDs (Mercer et al. 2006), may cause a couple to forego reproduction when they otherwise would choose to reproduce. A misunderstanding of environmental risk may result in avoidance behaviors not proportionate to the risk. Overestimating the significance of a particular genetic risk may prompt people to chase unnecessary diagnostic or therapeutic interventions.

While risk comprehension can be an issue in any context, genetic determinism and the inherent difficulty in communicating complex risk may make comprehension a particular concern for ASDs. One issue is that much ASD risk information is of small effect size, and comprehending small probabilities may be particularly difficult for many persons (Keller et al. 2006). Therefore, as compared to genetic markers that confer a very high probability of disease, genetic markers of small to moderate effect size may be inherently more difficult to understand—a problem that has also been raised with respect to other complex diseases, such as type-II diabetes mellitus (Haga 2009). In addition, a version of the availability heuristic might contribute to the exaggeration of small probability risks. The availability heuristic describes people's tendency to overestimate the probability of risks that come easily to mind (Keller et al. 2006). The "availability" of ASDs may be influenced by such things as media coverage. In addition, to the extent that risk functions in a "problem-classifying" way for most people, the very act of communicating ASD risk information may make it

more salient and hence predispose people to overestimate ASD risk when they were previously unaware of specific risks.

Second, we should also consider the possibility that beliefs about ASD risk may depend on noncognitive factors (as opposed to cognitive understanding of numerical information). In general, “affect” or emotion has been shown to influence people’s estimation of probabilities (Klein and Stefanek 2007). Risk communication may figure prominently in providing a sense of explanation to families affected by ASDs. To date, most cases of ASDs (85–95%) are considered idiopathic, and studies of ASDs have highlighted many ASD-affected families’ desire to find an explanation for why they or their child has been affected by an ASD. For example, a recent examination of parents’ views regarding the return of ASD-related research results indicated that most parents considered a result most relevant when it provided explanation for why their child developed an ASD (Miller, Fiona, et al. 2010). Considered in itself, the ability of risk communication to provide explanation is a good thing, but ethical concerns arise in relation to what counts as a legitimate explanation, as well as how families’ desire for explanation might affect their interpretation of risk information. The peace of mind and certainty provided by explanation may plausibly make ASD-affected families more likely to view a weakly determinative risk factor as strongly determinative, and therefore may make inadvertent harms associated with genetic determinism more likely.

A third reason why risk communication may prompt persons to act contrary to their interests is that the risk information itself might be too poorly supported by evidence to rely upon. In such circumstances, someone may base their decisions(s) upon risk information that turns out to be false. In light of difficulties concerning the adequate powering and replication of genetic studies, this issue has already received some attention as concerns ASDs and the return of research results (Miller, Fiona, et al. 2010). As discussed in the next section, the appropriate way to address this issue is not straightforward since judgments about “sufficient” or “appropriate” evidence levels are evaluative.

Finally, risk communication may lead individuals to take actions contrary to their interests because risk messages themselves are poorly designed. For example, Tabor and Cho (2007) describe how many genetic studies in the primary literature use conflicting standards for making causal claims, or claim that associations are causal in nature when the evidence does not support this. To the extent that genetic counseling or other forms of risk communication are based upon claims made in the primary litera-

ture, misleading claims made there may have downstream effects. Even when causal claims made in the primary scientific literature are defensible, they may be distorted by press offices or the media (Yudell et al. 2012).

##### 5. POSSIBLE RESPONSES TO INADVERTENT HARM

The possibility that ASD risk communication may cause inadvertent harm raises questions about what risk communicators can and should do to mitigate or otherwise address such harms. One obvious and important step is to gather more information about the possible adverse effects of ASD risk information. While the adverse outcomes discussed here are plausible, they are based on conceptual arguments and limited (and often indirect) empirical data. Direct empirical data regarding how individuals respond to ASD risk are needed. In addition, research methodologies may in some circumstances need to be further developed, since some of the relevant empirical questions (e.g., concerning worry and anxiety) may not be ideally addressed using current analytical tools. Specifically, it is important in future empirical work to address the following issues: (1) Does ASD risk information make people anxious or worried; does anxiety or worry vary with the nature or magnitude of the ASD risk; and do different research methodologies yield different answers about affective responses to ASD risk? (2) If affective responses vary with research methodology, do we have good reason to privilege one methodology over another when trying to ascertain what people “really” feel? (3) Does the process of ASD risk communication, either in persuasive or nonpersuasive form, make people feel guilty? Do guilty feelings result in different actions in response to risk? (4) To what extent is broad public health communication about ASD risk liable to implicate notions of responsible parenting and responsible risk avoidance into social discourse about autism? (5) What kinds of risk information offer “reasonable” or “genuine” explanation in the view of ASD-affected families? (6) How, if at all, are people likely to act in response to risk information of small effect size in terms of risk avoiding or risk-reducing behavior?

It may take some time to accrue the relevant empirical data, and research on ASD risk continues apace; some present thought to risk communication policy is therefore warranted. Assuming the plausibility of the harms discussed here, the next question is what risk communicators ought to do about them. While we cannot provide a comprehensive normative analysis here, we do identify several key issues as a way of setting a normative research agenda.

One relevant normative question concerns the extent to which risk communicators are morally responsible for inadvertent harms that occur. It might be argued that when risk communicators neither directly cause such harms nor intend for them to occur—conditions that may apply to many of the harms discussed here—they are not morally responsible for their occurrence, or are at least less responsible than they would have been had the harms been intended (see, e.g., Davis 1993). (This argument is often discussed under the heading of the “intention/foresight” distinction.) Alternatively, it might be argued that since risk communication can causally mediate some harms, risk communicators are responsible for all reasonably foreseeable harm, regardless of their intentions or whether their actions were causally sufficient to bring about the harm. Furthermore, the notion of “degrees of moral responsibility” requires some interpretation—this notion may be intelligible as concerns blameworthiness, but not permissibility. These questions about intention, foresight, and degrees of responsibility may bear upon the extent to which risk communicators are obligated to expend resources to prevent or minimize inadvertent harm.

A second issue relating to moral responsibility concerns the degree of risk communicators’ positive obligations of assistance. It might be argued that some specifications of nonmaleficence concerning the prevention, removal, or mitigation of harm are better viewed as specifications of beneficence. This distinction is more than semantic, as moral philosophers often seem to agree more about our negative obligations of noninterference than they do about our positive obligations of assistance. Physicians sometimes act as risk communicators, and where this is the case we might appeal to their fiduciary duty to explain why they have obligations to prevent, remove, or mitigate harm (explaining the source and authority of this fiduciary obligation is, of course, another matter). But when risk communication occurs in the public health context the matter is less clear. It might be argued that public health communicators have positive obligations of assistance to their audience, either because they bear some sort of quasi-fiduciary duty analogous to a physician’s fiduciary duty, or because such specific obligations can be derived from general moral principles and circumstance-specific factual considerations (e.g., risk communicators’ power over an audience in virtue of their ability to shape their audience’s perception). Though we will not defend such arguments here, we do believe that risk communicators have obligations to try and minimize inadvertent harm. However, we recognize the need for a more developed argument taking into account the considerations raised here.

When trying to minimize inadvertent harm, perhaps the most fundamental question is when sufficient evidence exists to recognize a risk as “real.” In light of difficulties concerning adequate powering, replication, and inconsistent experimental methodologies, numerous commentators have raised this issue as regards the return of results in genetic research, and specifically autism (see, e.g., Miller, Fiona, et al. 2010). However, to date no specific and/or adequate proposals have been advanced to deal with determinations of “sufficient” evidence. Shalowitz and Miller (2005) recognize the importance of information quality, but argue that research participants’ requests for information should be honored even when that information is highly uncertain. The basis for their argument is that a principle of “respect for persons” requires such disclosure, but this is more asserted than argued for. Furthermore, honoring all requests for disclosure will likely result in inadvertent harm in situations where risk communication does not provide any substantive benefit to information recipients; we therefore have significant reservations about this approach. Other commentaries (Ravitsky and Wilfond 2007; Miller, Fiona, et al. 2010) suggest that risk information should be disclosed to research participants only when that information is scientifically valid, but leave open the question of how, exactly, we should make this determination.

It is important to emphasize that determinations of scientific validity are value-laden; there is no value-free, “objective” algorithm for making them. “Normal” science operates with a set of conventions for statistical inference designed to minimize false-positive (type-I) error, but as concerns environmental risk numerous philosophers have argued that a greater emphasis on minimizing false negative (type-II) error is warranted. This is because ‘risk’ as a concept is action-guiding, and serves to avert possible harm; risk is not an inert scientific concept intended only to describe the world (Shrader-Frechette 1991; Hansson 2007).<sup>8</sup> As such, the set of values driving risk assessment, management, and communication is different than the set of values governing normal science and its accumulation of knowledge. This same argument can be extended to ASD risk communication, whether genetic or environmental. There is an inherent tension between minimizing harm and maximizing benefit when it comes to setting evidential thresholds: a high threshold of evidence minimizes any inadvertent harm that may arise from acting upon faulty risk information, but denies persons the benefit of risk information in some situations where a lower threshold of evidence may be appropriate. A low threshold of evidence maximizes the chance that persons will receive potentially useful informa-

tion, but also carries with it a higher chance of inadvertent harm. While general considerations about the relative importance of type-I and type-II errors might guide determinations about sufficient evidence, it may be the case that such determinations can be made only in a procedural and not a substantive fashion (Shrader-Frechette 1991). However, much additional work is needed to practically apply the general philosophical arguments cited above to specific contexts such as ASDs.

Assuming that the risk information in question is scientifically valid (however this is determined), the ethically ideal approach would be one that looks to minimize harm while still honoring message recipients' preferences regarding the type and amount of risk information that they receive. Persons who do not wish to receive some kinds of risk information may "opt out" of it, but persons who desire all scientifically valid risk information (e.g., any positive association between a risk factor and ASDs, regardless of effect size or ability to infer causality) would have the option of receiving it. A number of tools may help risk communicators to reduce the likelihood of inadvertent harm arising from lack of comprehension or emotional distress, while still honoring message recipients' information preferences. Graphical representations of risk such as risk ladders have been shown to increase comprehension of risk (Keller et al. 2009; Tait et al. 2010). Genetic counseling can both increase understanding of risk and also help to alleviate risk-related anxiety (see, e.g., Pieterse et al. 2005; Senay and Kapingsht 2009; Kladny et al. 2011).

However, these approaches to minimizing harm are not 100% effective. For example, risk ladders may be effective only amongst "high-numeracy" populations (Keller et al. 2009).<sup>9</sup> The efficacy of genetic counseling in correcting for misunderstanding of risk or in lowering anxiety is similarly variable (Pieterse et al. 2005, 2007; Senay and Kapinghst 2009). Some of the harms discussed here, such as stigmatization, are socially mediated, and providing genetic counseling to risk information recipients or changing message formats therefore seems unlikely to do much to address these kinds of harms. It might also be questioned whether genetic counseling will be widely available as ASD risk communication is extended from the contexts of clinical care and return of research results to the wider public health context. For example, if chromosomal microarray (CMA) testing becomes routine practice in the prenatal or neonatal settings, will genetic counseling be routinely available for individuals whose results indicate ASD-associated variants? Certainly genetic counseling cannot be provided in the context of environmental risk communication undertaken by public

health agencies, so whatever benefits it provides would be moot here. Finally, as concerns risk communication in the broader public health setting, honoring individuals' preferences for information will likely prove difficult. Persons who do not wish to receive information about environmental risk may still be presented with that information regardless.

A third approach to mitigating inadvertent harm would be to control what risk information message recipients have access to, or when they have access to it. For example, alterations of identity are more of a concern were genetic testing to be undertaken before a firm clinical diagnosis of ASD could be made, and where the genetic risk factors revealed are not sufficient to cause an ASD. By restricting the use of genetic testing to the confirmation of clinical diagnoses or the workup of individuals strongly suspected of having an ASD, concerns about the alteration of identity might be significantly curtailed. Where risks arising from the environment or gene–environment interactions are concerned, risk communicators might try to target those individuals or populations who are most expected to encounter the environmental exposure, or to have the appropriate combination of environmental exposure and genetic profile. Targeting higher-risk populations in this way may help to reduce generalized risk anxiety (by limiting overall risk message exposure), or the possibility that people may act upon risk information in ways detrimental to their interest.

Risk communicators might also withhold scientifically valid risk information when that information does not appear to be useful or relevant. Exactly when risk information is useful or relevant has been subject to some debate. Some guidance documents (e.g., NBAC 1999) have defined “useful” in terms of *clinical* usefulness, as concerns either diagnosis or therapy. However, other commentators have rightfully pointed out that risk information may also be useful or relevant in other circumstances. For example, risk information can provide explanation and meaning, and even where it cannot be used clinically, risk information may allow individuals to plan certain life decisions differently, for example as concerns reproduction (Shalowitz and Miller 2005). In our view, the key issue here is whether a threshold exists for calling scientifically valid risk information relevant. For example, is an ASD risk factor associated with an incremental risk of a few percent relevant? Can it be said to legitimately provide explanation for why a child has developed an ASD? Will such information make a difference to the life-planning or risk avoidance decisions of a rational actor? Clearly these are philosophical determinations that require workable conceptions of both rationality and explanation.

Determinations about access to risk information also invoke moral concerns. Most obvious is the question of whether individuals have a right to all information about themselves, or whether respect for persons requires the disclosure of any requested information. Despite numerous attempts to use a principle of “respect for persons” as grounding for an obligation to return all individual results to research participants, it is not clear whether and why the principle supports this specific conclusion. If participation in a research study creates ownership over genetic information, then this needs to be argued, but to date no such argument appears to have been made. If a good case cannot be made that risk information is relevant to a rational actor, then it is hard to argue that withholding that information *substantively* diminishes a person’s autonomy. While thwarting a person’s *preference* for risk information might be viewed as violating autonomy in some sense, whatever obligations exist to honor such preferences may conflict with risk communicators’ obligations to prevent or minimize harm, and if the preference itself is irrational then such obligations *may* trump. On the other hand, arguments against risk disclosure that are based on the information-gathering purpose of research seem to miss the point (see, e.g., Clayton and Ross 2006). Unless disclosing risk information in some way thwarts important research goals, the fact that the primary goal of research is knowledge-building and not therapy seems irrelevant to the question of whether researchers should disclose potentially beneficial risk information that happens to be produced as a byproduct of this knowledge-building. Clearly more philosophical work remains to be done as concerns these difficult questions.

Finally, apart from the question of whether respect for persons requires the disclosure of all individual information, there are risk–benefit considerations to be dealt with in the policy setting. For example, limiting genetic screening to the confirmation of clinical ASD diagnoses or the workup of individuals strongly suspected of having an ASD would help to minimize certain inadvertent harms, but would entail the loss of any benefits of early detection. Hence there is a tradeoff between potential benefit and risk. In addition, risk communication in the policy setting needs also to consider economic costs as relates to the potential benefits of such screening. As is the case when evaluating research interventions, risk–benefit determinations as concerns screening tools are inherently evaluative and contestable.

## 6. CONCLUSION

Autism spectrum disorders (ASDs) are an issue of significant and growing public health importance. In recent years a large amount of information about genetic ASD risk factors has been published, with more information about genetic and environmental ASD risk expected to follow in the near future. The scientific validity of much of this risk information is uncertain, and even where ASD risks are well characterized their ultimate value or relevance is frequently questionable in light of small effect sizes and gene-environment interactions. While risk communication in any context raises ethical issues, the complexity and uncertainty frequently characteristic of ASD risk make this topic particularly difficult to navigate. In the present article we have discussed the possibility that ASD risk communication may lead to a number of inadvertent harms, including anxiety and worry; guilt, blame, and stigma; alterations of identity; and individuals acting contrary to their interests. Many of these harms are speculative, though conceptual arguments and empirical data from other risk communication domains support their plausibility. Nonetheless much additional empirical research is warranted.

Similarly, additional normative research is warranted, since existing discussions of the ethics of genetic risk disclosure often operate with conceptually simplistic definitions of risk, rely upon concepts such as “relevance” or “utility” that require further clarification, or advance normative arguments around disclosure that are insufficiently motivated. While our primary aims in this paper have been to describe how ASD risk communication might lead to inadvertent harm and what possible responses risk communicators might take, we do provisionally suggest that the routine disclosure of all ASD risk information known to be scientifically valid, regardless of effect size or causal attribution, is ill advised. Given the high level of public scrutiny and concern about ASDs and the ways in which risk information might lead to harm, communicating information of little ultimate use to individuals seems to have distinct disadvantages and few advantages, apart from honoring some persons’ preferences regarding what information they would like to receive. Whether such preferences correlate to a *right* to this information, or an obligation on the part of risk communicators to disclose it, is a matter that remains to be adequately resolved.

## NOTES

1. There is ongoing debate about whether this prevalence increase reflects a rise in real risk, with changing diagnostic tendency an alternative explanation. However, some studies have attempted to take diagnostic tendency into account and have still concluded that some of the prevalence increase is genuine. For further discussion, see, e.g., Herbert (2010).
2. It should be noted that the notion of “better outcome” presumes some therapeutic goal, such as changing a child’s social interactions, that might be contested. Hence questions about early intervention and outcomes are intimately related to questions about neurodiversity.
3. For example, a review of the role of rare DNA copy-number variations (CNVs) in autism spectrum disorders estimated the population-attributable risk for *all* CNVs at 3% collectively (Pinto et al. 2010).
4. A recent review of studies examining the effect of common alleles in ASD risk reports odds ratios of 1.1–1.2 (Devlin et al. 2011). A recent examination of the functional impact of rare copy-number variations in ASDs found that ASD cases showed a statistically significant increase in the number of genes intersected by CNVs, but the effect size was relatively small at a 1.19-fold increase. The same study also examined whether there were significant differences between cases and controls as regards the number of ASD-implicated genes intersected by CNVs and again found differences between cases and controls; however, the effect size was again modest, with an odds ratio of 1.8 and a risk difference of about 2% between cases and controls (Pinto et al. 2010).
5. For example, a recent examination of the role of SSRIs in the development of ASDs reported a positive association, with an odds ratio of about 2 and a risk difference of about 3% between cases and controls (Croen et al. 2011).
6. Currently some groups recommend the use of chromosomal microarray testing as part of the diagnostic workup of individuals with ASDs, though it is acknowledged that the clinical significance of many genetic variants associated with ASDs is uncertain (Shen et al. 2010). However, it seems likely that CMA testing will be more widely used in the prenatal, perinatal, and early life settings, before a conventional diagnosis of ASD can be made based on clinical signs. Already CMA is being used in some quarters as part of prenatal genetic testing (Singer 2012). Some discussions of translating genetic research on ASDs into the clinical setting have also broached the possibility of using CMA as part of routine neonatal disease screening panels (see, e.g., Pellicano and Stears 2011). Finally, a number of instruments are being developed to aid in the early detection of ASDs, prior to the time when an ASD could be

- diagnosed conventionally. These early detection tools are promising, but carry with them the possibility of false positive and false negative results as compared to conventional clinical diagnosis (see, e.g., Zwaigenbaum 2010).
7. A number of reviews examining anxiety and distress consequent to genetic risk communication have concluded that for most persons, these outcomes are either unlikely to occur, and/or are low-level and transient (e.g., Broadstock et al. 2000; Crotser and Boehmke 2009). On the basis of this information, some ethical commentaries on the return of research results (e.g., Shalowitz and Miller 2005) have concluded that inadvertent harm is not a problem. Aside from the fact that such commentaries ignore harms not related to distress, there is reason to doubt that the empirical data cited above tell the whole story. Other studies (e.g., Tzeng 2010) have indicated that a significant percentage of persons (~25%) may experience distress in response to risk information, and in addition there is evidence suggesting that the very process of bringing risk information to individuals' attention increases their anxiety (Lister et al. 2002). Furthermore, many of the risk communication studies failing to document significant anxiety use quantitative measures such as the State-Trait Anxiety Inventory (STAI). Qualitative studies more strongly support the conclusion that risk communication may cause anxiety (Crotser and Dickerson 2010), and quantitative measures of anxiety have been criticized for potentially underestimating the extent of individuals' anxiety (Evers-Kiebooms 2001; Lister et al. 2002). Finally, anxiety and worry may be distinct constructs (Davey et al. 1992; Constans 2001), and what measures one may not measure another. For example, it has been documented that worry propensity may not track with anxiety, and that individuals who do not exhibit clinically significant increases in anxiety as measured by the STAI may still worry excessively (Constans 2001). All of this suggests not only that additional refinement of empirical methods of assessing worry and anxiety are needed, but also that the dismissal of anxiety and worry as significant outcomes of risk communication is premature—especially in light of the fact that anxiety and worry are such *intuitively plausible* outcomes where ASD risk communication is concerned.
  8. Furthermore, it is not clear that standard scientific conventions concerning, say, significance testing are sufficient to address the issue of replication between studies.
  9. More fundamentally, the structure of some risk perception studies is such that it is not entirely clear whether risk ladders actually increase cognitive understanding as opposed to simply changing a person's perception of risk level (Keller and Siegrist 2009).

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