

The Role of Optometry in Early Identification of Autism Spectrum Disorders

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ABSTRACT

Autism spectrum disorder (ASD) is a diverse, behaviorally identifiable neurodevelopmental disorder that occurs in 1 in 150 children. The absence of eye contact, unresponsiveness to facial gestures, and/or difficulty in sharing joint visual attention are signs of abnormal or atypical visual development. Optometric utilization of various targets used for diagnostic testing in infants and toddlers may be among the earliest probes of preferred visual looking patterns conducted with this population. This potentially places Optometry in the vanguard of identifying infants and young children at risk for developing ASD characteristics.

Keywords: Autism spectrum disorders, early identification, eye contact, gaze avoidance neurodevelopmental disorder, preferential looking.

Introduction

Autism spectrum disorder is defined as a heterogeneous, behaviorally identifiable neurodevelopmental disorder that occurs in 1 in 150 children.¹ As a constellation of neurobehavioral disorders, ASD

is characterized by the absence of what is referred to as neurotypical development.² Neurotypical behavior at an early age most often centers on issues in nonverbal communication related to socialization. A classic example is the bonding of infants with caretakers that typically occurs through eye contact, often while the infant is feeding (Figure 1a). Normally developing infants have eye contact at 8 weeks of age, and at the age of twelve weeks the social smile and active interaction are present. The absence of eye contact, unresponsiveness to facial gestures, or difficulty in sharing joint visual attention are potential signs of abnormal or atypical visual development³ (Figure 1b).

Beyond socialization issues, newer models of autism acknowledge the role of motor, sensory, postural control and oculomotor systems.⁴ Signs of atypical neurobehavioral development may therefore be rooted in oculo-visual, cognitive, and language factors, or their interrelationship; and are increasingly acknowledged to require early identification and a multidisciplinary treatment approach.^{5,6} In advancing the case for the role of Optometry in early identification of ASD,⁷ we begin our discussion with concept of preferential looking.

Preferred Looking Patterns

The experimental psychologist, Robert Fantz, introduced preferential looking as a paradigm in 1958, initially publishing his results in 1962 in the *Journal of Comparative and Physiological Psychology*.⁸ It was not until 12 years later that Davida Teller and her colleagues published the first article on forced-choice preferential looking (FPL) and gave it clinical application to the measurement of visual acuity through the use of grating patterns.⁹ The impetus for the development of these procedures was to introduce a means of nonverbal response that would indicate an infant's preference in responding to a stimulus

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Press LJ, Richman J. The role of optometry in early identification of autism spectrum disorders. *Optom Vis Dev* 2009;40(3):141-149.



Figure 1a: Early interaction of a normally sighted infant versus an infant with visual impairment. A. At the age of three months, socio-visual communication of a normally sighted infant is an effective bonding function; the infant and the adult understand each other right from the start.



Figure 1b: A visually impaired infant may not have normal eye contact and may seem to look at the hair of the adult because of the more obvious contrast with the background, as compared to facial features. Pictures and text adapted from <http://www.lea-test.fi/en/assessme/vision.html>

that could be resolved, as opposed to a paired, blank pattern. FPL techniques have subsequently been adapted to other clinical measures, such as contrast sensitivity and stereopsis. It has also been used to probe visual function in populations with various forms of communication impairment.¹⁰ While visual acuity and stereopsis remain important clinical measures in applying the preferential looking concept to amblyopia and strabismus, they have overshadowed the significance of Fantz's original contributions about preferred looking toward faces.

Fantz's original observations included the ability of infants to discriminate between various stimulus pairs of target patterns. The stimulus pairs ranged from geometric patterns to face patterns. The paired face patterns included one pair with a schematic smile face opposite one with disordered features, and another pair with a schematic face versus a face photograph. Fantz recognized that the visual-preference method offered a simple and direct approach to preferential looking based on the duration of the infant's attention, though he was cognizant of the intensity of the looking response as well, and the need to factor novelty and habituation into the equation.¹¹

In assessing visual selectivity, Fantz felt that it was important to know the basis of selection by the infant and how it changes with age. Smiling face targets have been utilized to apply Fantz's principles in probing the infant's ocular motor competency for fixation, pursuits and convergence. As an example, in developing the Heidi fixation face paddle to which normally developing infants respond at three months of age (Figure 2), Lea Hyvarinen credits the face design

to Fantz's original studies. A similar face target, Patti Pics (Figure 3), was developed in 2003 by Precision Vision for fixation and acuity testing.

The Richman Face Dot Test

A related example of the application of Fantz's original preferred looking patterns is the Richman Face Dot Test (RFDT). The RFDT is a composite test, utilizing a smile face target in a forced choice paradigm. This approach has been developed for measurement of the visual acuity of children. These tests are also known as vanishing optotypes and employ the use of high pass spatial frequency figures. The test charts are face figures created by alternating black and white dots on a white background. Any cross-section of the dots has a Fourier transform with a zero frequency component equal to the luminance of the background. When the dots are out of focus, they fade rapidly into the background causing them to become invisible, hence the concept of "vanishing optotypes." Rather than merely blurred as in a typical Snellen chart, the face pattern disappears as a function of the visual acuity level. The examiner first confirms the patient's preference for looking toward faces by holding a bold dot pattern smile face paddle in one hand, and a blank pattern in the opposite hand. The examiner then holds a blank paddle in one hand and a contrast dot smile face pattern in the opposite hand (Figure 4). The results are converted into an approximate visual acuity based on the testing distance at which the patient no longer exhibits a preference for looking toward the face pattern as opposed to the blank paddle.



Figure 2: Heidi Face Paddle. From: <http://www.lea-test.fi/en/vistests/instruct/2530/index.html>



Figure 3: Patti Cake™ Large Fixation Paddle. From: http://precision-vision.com/index.cfm?fuseaction=feature.display&feature_id=6

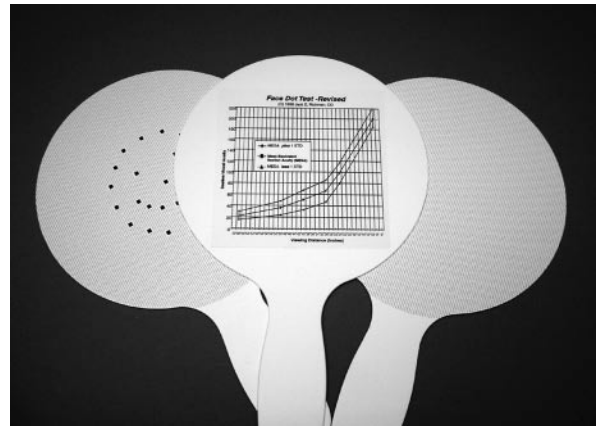


Figure 4: Richman Face Dot Paddles

The concepts utilized in Preferential Looking testing to discriminate between various stimulus pairs; especially faces, are of principal importance when trying to engage patients with ASD. Aversion to face targets in general and eyes in particular, may account for poor performance. Evidence suggests that the face processing problem is not an isolated issue in social engagement, but a fundamental alteration of visual processing in autism.¹² Optometric utilization of face targets used routinely for clinical testing of ocular fixation, pursuits, saccades, and stereopsis in infants and toddlers may be among the earliest assessments of preferred visual looking patterns conducted with this population. This potentially places Optometry on the front lines of identifying infants and young children at risk for developing or expressing ASD characteristics.

Eye Contact and Autism Spectrum Disorders

One of the hallmark features of autism is gaze avoidance. Inattention to faces, particularly the eye region, is one of the earliest and most consistent signs of ASD.¹³ Klin and his colleagues used infrared eye tracking technology to measure scan paths of individuals being screened for autistic spectrum disorder as they viewed movie clips involving social interactions.¹⁴ Individuals with ASD focused much more intently and more frequently on the speaker's mouth, and rarely on the eyes. How early is this pattern of gaze avoidance in place? Klin and colleagues narrowed their eye tracking investigations to fifteen 2-year-old children with autism compared with 36 typically developing children and with 15 developmentally delayed but nonautistic children.¹⁵

Preferential attention was measured as percentage of visual fixation time to 4 regions of interest: eyes, mouth, body, and object. Level of social disability was assessed by the Autism Diagnostic Observation Schedule. They determined that looking at the eyes of others was significantly decreased in children with autism, while looking at mouths was increased in comparison with both control groups. The two control groups were not distinguishable on the basis of fixation patterns. In addition, fixation time on eyes by the children with autism correlated with their level of social disability with less fixation time on the eyes predicting a greater social disability. Their results indicate that fixation patterns involving eye contact serve as a potential biomarker for quantifying autism spectrum disorders at two years of age. The possibility exists that this biomarker is reliable at younger ages, though definitive studies have not as yet been published.

There are many theories as to why young children who are on the spectrum of autistic disorders have difficulty attaining or maintaining eye contact. There may be sensory overload involving vision, with ASD individuals finding it more soothing or calming to view peripherally, or look beyond objects of regard. At a very basic level, the term "autism" implies being within oneself, and gaze aversion helps limit interaction with other than self. The earliest signs of autism most parents noted were that their children seemed to "look right through them," or even appear to avoid looking at them. However, current research perhaps indicates another view of this behavior. The most influential school of thought in this regard stems from the work of Simon Baron-Cohen et al.

in the United Kingdom who conceived a Theory of Mind that has most recently surfaced in the context of mirror neurons in the brain.¹⁶ Baron-Cohen and his colleagues show, that ASD children don't actually avoid making eye contact, but rather, they fail to make eye contact at appropriate times. The researchers speculate that part of the explanation for the gaze abnormalities in autism may be a failure to comprehend that the eyes convey information about a person's mental states and emotions. They believe their findings indicate that children with autism have difficulty with or lack understanding the concept or Theory of Mind of other people having thoughts and feelings of desire, goal, intention to refer and think, resulting in them not mapping these onto behavior such as eye-direction. In short, they say, ASD children may be blind to the mental information that other people convey with their gaze.

A key factor in Baron-Cohen's model is the presence of an eye-direction detector (EDD). They found that infants are very sensitive to eye-direction, particularly for distinguishing between eyes that are looking at them and eyes that are looking away. This appears to be the simplest stage of the mechanism that decides a basic question, "am I being observed?" Essentially, this is the eye-contact detection mechanism. One cue that can be utilized to infer what another is likely to do, i.e., intending thought or action, is eye-direction detection (EDD). It allows infants to develop knowledge of where an adult is looking, and engage in social interaction through shared emotion. The brain regions involved in gaze processing in ASD related to EDD appears to be localized neuroanatomically in the superior temporal sulcus and amygdala, and synchronizes its actions with the orbitofrontal cortex. This region of the superior temporal sulcus (STS), based upon event-related functional MRI (fMRI) studies was not sensitive to intentions conveyed by observed gaze shifts. There is a clear difference in the response of brain regions underlying eye gaze processing in autism.¹⁷

It is clear that infants developing normally rely on information from eye contact to bootstrap or augment and turn on other developing systems, and that children with autism typically remain in the stages exhibited by young infants.¹⁸ It has been suggested that oculomotor abnormalities may play a role as a sensorimotor defect at the root of impairments in later developing functional systems, ultimately resulting in socio-communicative deficits.¹⁹ Collectively, based

on this recent work, it appears that at numerous levels, vision can be seen to play a critical role in the development of theory of mind.²⁰

Seeing The World Differently: Local vs. Global Processing

Uta Frith is a cognitive psychologist who collaborated with Simon Baron-Cohen in proposing the Theory of Mind that helps to account for the looking patterns of ASD children.²¹ As a framework for their perspective, bear in mind that in neuroscience, there are distinct strategies of information processing. One of the more established concepts is the distinction between top-down and bottom-up strategies, considered to be strategies of information processing and knowledge ordering. A bottom-up approach is the piecing together of a scheme to expand into a larger organization method. In a bottom-up approach the person's initial parts of the system are specific and in great detail. These parts are then linked together to form larger subsystems, which then in turn are linked, sometimes in many levels, until a complete top-level system is formed. The study of visual attention provides an example of this bottom-up approach. If your attention is drawn to a bird in a tree, it may be simply that the bird is more visually prominent than the surrounding tree. The information that caused you to attend to the bird came to you in a bottom-up fashion — your attention was not dependent upon familiarity of the bird.

In a top-down approach a general idea of the search plan is first put together, specifying but not detailing any specific subsystem to use. Each subsystem strategy for searching is then refined in greater detail, sometimes in many additional subsystem levels, until the entire plan is compacted to satisfy the goal at the outset. Consider the top-down approach or strategy in our situation of looking for a bird. You have a representation in your mind of what you are looking for, i.e. the bird. When you see the object you are looking for, it becomes important and further prompts your visual attention. This is an example of the use of top-down information.

In cognitive terms, the two thinking approaches are distinctly different from each other. A "top down" (or big chunk) approach is used by the individual who sees the larger picture and overview, and works downward from there. Such people focus on the big picture, deriving the details to support that picture. In contrast, a "bottom up" (or small chunk) thinking

strategy is utilized to focus primarily on the details rather than the entirety of the background. This has led to the expression that, “The person does not see the forest for the trees”.

How does this apply to ASD? Elements or details that might be hard to extract, and therefore remain hidden from the visual experience of normal observers, might be perceived very easily by observers with ASD. Conversely, individuals with ASD may not grasp broader concepts because they are focused on details. Weak central coherence is the term that Frith and Baron-Cohen coined to capture the individual’s preference for local details over global processing. The weak central coherence theory (WCC), also called the central coherence theory (CC), suggests that a specific perceptual-cognitive style, loosely described is a limited and restricted ability to understand context or to see the big picture, and underlies the fundamental disturbance in autism and related autism spectrum disorders (ASD).²² Another way of expressing this is that many individuals with ASD have a preference for the parts of objects rather than the whole. One might also describe this as a bottom-up or small chunk thinking strategy.

While we must be cautious in making sweeping overgeneralizations, it is fair to say that a key point in many of the theories about ASD behavior are rooted in a mismatch between top-down and bottom-up processes. Another point to be factored into theories of ASD is the influence of executive control. This is a theorized cognitive system that controls and manages other cognitive processes. It is also referred to as the executive function, supervisory attentional system, or cognitive control. The concept is used to describe a loosely defined collection of brain processes which are responsible for planning, cognitive flexibility, abstract thinking, and rule acquisition, as well as initiating appropriate actions and inhibiting inappropriate actions, and selecting relevant sensory information. These top-down and bottom-up processes are modulated and adjusted through executive control.²³

Dakin and Frith,²⁴ in their article regarding atypical visual perception of children with autism note that those who have ASD often exhibit above normal abilities in various psychological tests, e.g., the Block Design²⁵ and the Ebbinghaus illusion²⁶ (Figure 5A and 5B). An example of a clinical test frequently used by optometrists that might tease out visual processing styles in ASD is the Test of Visual Perceptual Skills (TVPS), with normative data available as early as 4



Figure 5a: Tasks used to probe perceptual processing that elicit supranormal performance in observers with ASD: Block Design subtest of the WISC

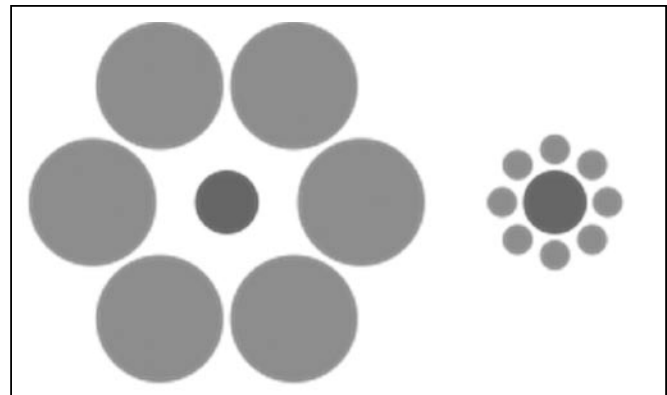


Figure 5b: Tasks used to probe perceptual processing that elicit supranormal performance in observers with ASD: Ebbinghaus illusion (surrounding targets change apparent size of central target)

years of age. Supranormal performance would be expected on the figure-ground subtest, which requires relatively local processing, and subnormal performance would be expected on the visual closure subtest, which requires global processing. Another example would be superior ability on tests of local stereopsis such as Wirt Circles and weak cognitive responses to random dot stereograms (which are global in nature and require central coherence which are deficient in ASD). Patients with ASD are most likely functioning in these stereotests with a bottom-up (or small chunk) thinking strategy.

Frith concludes that individuals with ASD either really do see the world differently, or attend to it in a radically different manner. Most recently, Frith’s research has cross-fertilized observations in autism and schizophrenia that may provide further behavioral insights. Both disorders involve abnormalities in social cognition, as well as executive function impairments. Indeed, they may be opposite sides of the same coin. Whereas individuals with ASD under-utilize Theory of Mind, those who are paranoid-schizophrenics see

meaning and communication in situations where it is not intended.²⁷

Frith's line of research is in understanding how individuals with ASD experience the world is also quite comparable to that of an optometrist, Melvin Kaplan. Kaplan has followed a similar line of investigation in behavioral optometry, making connections between focal vision (central processing) and ambient vision (global processing) in both schizophrenia and autism.²⁸ Padula has applied this dichotomy or duality to patients with acquired brain injury.²⁹ If one substitutes focal for local or a bottom-up strategy, and ambient for global or a top-down strategy, a striking parallel can be drawn between the observations by Frith and by Kaplan. Kaplan suggests combining standardized tests such as the Gesell Developmental Assessment (block play, incomplete man, circus puzzle, form puzzle and copy form) with his nonverbal battery to help differentiate the patient's style as focal or global.³⁰ As a complement to insights from traditional optometric measures and probes, an appreciation of the individual's visual information processing style aids in creating a management plan that initially builds on the patient's strengths while moving toward addressing weaknesses.

With specific regard to autism, Kaplan has elaborated the use of yoked prism lenses on helping to reorganize the balance between focal/local and ambient/global processing, an approach which others have applied successfully.³¹ He further notes that in most instances these changes with prisms are temporary, and that it takes months of guidance and training to transform temporary changes into permanent neural alteration.³² An example of this visual rehabilitation therapy might be the use of parquetry blocks to help the child transition from emphasis on parts, to recognition of a global pattern that emerges from a particular arrangement of the parts.

Lastly, considerations of local versus global processing do not occur in a vacuum, and must be factored into speed of processing and integrative connectivity.³³ For optometrists and therapists experienced in optometric vision therapy, these principles and techniques are recognizable. By extending one's repertoire of observations to the unique traits of the ASD spectrum, and through learned skill and practice, Optometry can help individuals with autism develop a more balanced visual approach to seeing.

A Unique Opportunity for Early Intervention

As optometrists become progressively more involved in the care of infants and young children, motivated by programs such as the AOA's InfantSEE®, we are presented with the opportunity to monitor early childhood/infant development from a unique perspective. Based on our preceding discussion, it should be apparent that many individuals with ASD have specific visual behaviors that are different from what the clinician will encounter with children with typical visual development including:

- Avoidance of eye contact
- Aversion of gaze and gaze following
- Insensitivity to joint visual attention with another individual
- Difficulty integrating peripheral/global vision with central/focal vision and
- Paradoxical fixation and perseveration on one particular object of interest

We can summarize these visual behaviors in ASD children by saying that they have inverted or reversed preferred looking patterns from what is normally expected. In other words, they exhibit visual behaviors opposite to what one would expect to observe when examining non-ASD or neurotypical infants and toddlers. Young children normally tend to look at the examiner's face rather than the target being presented. This makes collecting data more challenging, but is an aspect of the examination resulting in unintended versions of peek-a-boo that makes examining normal infants so enchanting. As an example, when the examiner is attempting near retinoscopy or using an OKN drum, normally developing young children will often try to look around the stimulus to find the examiner's face which has suddenly disappeared.

Based on the authors' clinical experiences, then, inverted preferred looking patterns may be noted during the examination by:

- The infant's disinterest in the doctor's face or expressions
- Disinterest in visual stimuli featuring facial expression, but attention to faceless targets
- Paradoxical response to stimuli such as Richman Face Dot paddles (infant prefers to look toward paddle with the face less distinct or absent rather than toward the obvious smile face target)

The key point in observing inverted preferred looking patterns away from faces or face targets is

that that a child may be at risk for emergent ASD behaviors at an early age. Clinicians who are comfortable with examining infants will note that the normal social context of the examination, in which the examiner and infant share joint attention, is lacking. This affords a unique opportunity to assess whether an infant's visual behavior is typical, demonstrating the normal patterns of preferred looking toward face-like targets and eyes, or atypical and placing them at risk for developmental delay.

Why is documenting the looking pattern of infants toward faces an important optometric screening tool for ASD? First, it may well help identify whether an infant's visual behavior was normal prior to 12 months of age. Particularly when an optometric clinician is able to periodically follow a child through a developmental timeline, documentation is available regarding deviations in the child's looking behavior. Information from early assessments and subsequent examinations may ultimately help developmental pediatricians and pediatric neurologists decide if a child had been developing normally, but is now experiencing regression or disintegrative disorder.³⁴ This becomes a crucial construct if we accept that ASD is a syndrome of developmental disconnection between key cortical areas³⁵ a manifestation of which can be the failure to integrate ocular motor systems with broader cognitive functions.³⁶ From an optometric standpoint, visual interventions are available in early childhood that can help guide visual development and connect vision with multimodal higher-order association processes.^{36,37} Any interventions effective for ASD children should be implemented at as early an age as possible, and a variety of behavior checklists and observation schedules have been recommended to assist in early detection.³⁸

Caveats in Assessing Looking Behavior

Caveat #1: Before making any assumptions about the cause of atypical looking behavior, the optometrist must check carefully for uncompensated refractive abnormality by conducting retinoscopy and testing with appropriate lenses. In some instances high uncompensated ametropia may result in gaze aversion. An example is this 4 month old infant diagnosed as having infantile autism because she did not have normal eye contact with adults and seemed to avoid eye-contact. She did not



Figure 6: This 4 month old infant was diagnosed as having infantile autism because she did not have normal eye contact with adults and seemed to avoid eye-contact. She did not accommodate to usual fixation targets. Plus lenses were used in trial form and the infant immediately looked surprised and after a few seconds developed a normal social smile and good eye-contact with her mother. Adapted from: <http://www.lea-test.fi/en/assessmelvision.html>

accommodate to usual fixation targets. Plus lenses were used in trial form and the infant immediately looked surprised and after a few seconds developed a normal social smile and good eye-contact with her mother (Figure 6).

If lenses are indicated by retinoscopy, an assessment of looking behavior can then be conducted with the lenses in place. We suggest that looking behavior toward face targets be then investigated by picture faces such as the Patti Cake or Heidi Fixation Paddles. If the patient shows normal interest in the target, then proceed to assess visual acuity with the Face Dot Test or an equivalent measure. However, if the patient shows gaze aversion to the face fixation target, reassess fixation attention and gaze preference with life size pictures of actual faces at near, such as a photograph or magazine picture. This picture should have a viewing hole for the examiner to observe the infant's gaze pattern. At a more basic level, reassess how well the patient is able to make eye contact with the examiner. If continued and repeatable aversion to looking at faces, particularly the eye region is noted, consultation with a developmental pediatrician or early intervention specialist should be considered. (See flow chart that follows these steps, Figure 7).

Caveat #2: Another caveat to bear in mind is the overlap between cortical visual impairment (CVI) and ASD gaze patterns. As note by Pring,³⁹ since Fraiberg's classic descriptions,⁴⁰ researchers have increasingly noted the commonalities between children with autism and those with severe and profound visual impairment. Earlier we mentioned the work of Hyvarinen in developing clinical tests of looking behavior patterned after Fantz's original studies on faces. In developing the Heidi Expressions Test Game⁴¹ to improve the early evaluation of vision for communication, Hyvarinen stressed that some visually impaired children have deficits in visual recognition of facial expressions as part of their extensive loss of visual function (which she calls a pathway problem),

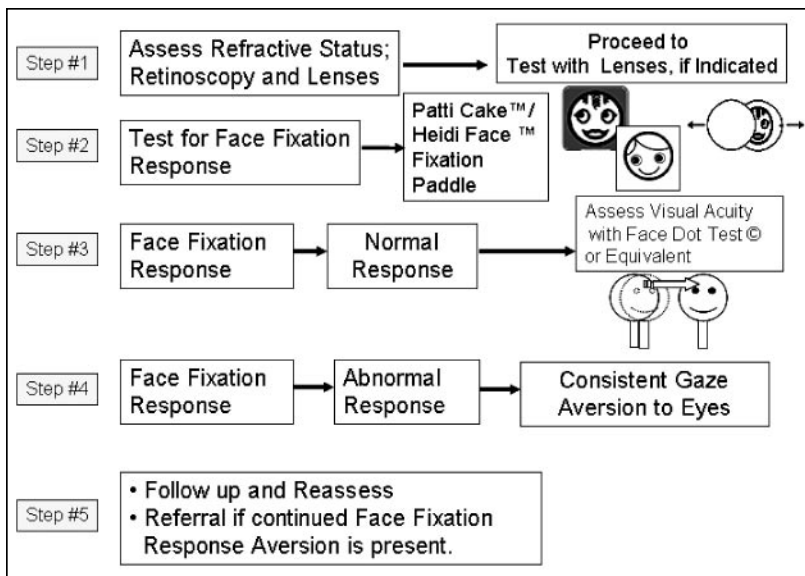


Figure 7: Flow chart for investigating gaze patterns



Figure 8: Heidi Expressions Test Game
<http://www.lea-test.fi/en/vistests/instruct/heidiexplheidi.html>

whereas others show near normal visual acuity and fields but do not respond to expressions (which she refers to as a cognitive visual problem).

The Heidi Expressions Test Game consists of a series of high contrast, black and white cards with various facial expressions that can be used in a matching game format when the child is not functionally blind (Figure 8). Hyvarinen notes that without early intervention, a child with gaze aversion to faces may experience social play so stressful and confusing that he or she withdraws and may be diagnosed as having autistic behaviors. In essence, overlap or in some instances, a misdiagnosis can occur in either direction. Children with CVI can have autistic behaviors, and children on the autistic spectrum may appear to have CVI specifically when averting gaze toward faces.

Conclusion

Most authorities now believe that subtle signs of ASD are present under 12 months of age,⁴² and eye tracking technology has been used experimentally to detect gaze patterns at progressively younger ages.⁴³ Research will continue to update clinical practice and contribute to clinically useful tools, including electrodiagnostic and brain imaging assessments. Yet one of the basic observations that will remain within the chair side province of the clinician is engaging the infant and noting that eye contact has begun to replace physical contact by 9 months of age. Retinoscopy, the use of lenses, and assessment of visual acuity with targets other than faces is essential in making a tentative differential diagnosis. If eye contact is lacking

Table 1: Early Socio-Visual Signs of Autism Spectrum Disorders

(Adapted from <https://aap.org/healthtopics/autism.cfm>)

- Doesn't keep eye contact or makes very little eye contact
- Doesn't respond to parent's smile or other facial expressions
- Doesn't look at objects or events parents are looking at or pointing to
- Doesn't point to objects or events to get parents to look at them
- Doesn't bring objects to show to parents just to share interest
- Often does not have appropriate facial expressions
- Does not perceive what others might be thinking or feeling by looking at their facial expressions

to fixation targets with facial features, but present to other stimuli, the parent should be quizzed about how indicative this is of behavior at home.

If the parent is unaware that the infant does not exhibit appropriate eye contact, the optometrist can re-schedule the child for a visit in one month to see if the gaze aversion is consistent. In the interim, the parent can maintain an informal inventory of visually-based social behaviors such as found in Table 1. If eye contact is improved, visual development should be monitored in three to six months and on a routine basis thereafter. However, if the infant still exhibits poor eye contact, and exhibits other possible signs and indicators of ASD, collaboration with appropriate professionals should be arranged. In doing so, the optometrist can make a valuable contribution by referring for appropriate early intervention.

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