Course Description
• This course will provide an overview of common systemic diseases present in the pediatric population and review the ocular findings that may be present.
• Genetic and neurodevelopmental syndromes and the ocular findings that accompany these syndromes will be discussed.

Course Objectives
• Discuss genetic and neurodevelopmental syndromes and review ocular findings that are associated with these conditions.
• Become familiar with managing ocular manifestations of systemic diseases in the pediatric population.
• Review prevalent systemic diseases among children and managing these conditions for best results.

Do these patients have Special Needs?
• Will managing these patients in your practice invite more patients to be examined that have "Special Needs?"
• What defines this phrase, and how can you be prepared to deliver the best possible outcome for these patients?

How prevalent are special needs?

Patients will present with different needs and abilities. Patients will have different diagnoses and chief complaints. Managing the visual requirements of special needs patients in an accurate and efficient manner is a goal of this course.

Autism: 1 in 68

87% of families of children with special needs have income below the poverty level.

VshfHoQhhgy
Overview

Centers for Disease Control website says:

Prevalence
About 1 in 68 children has been identified with autism spectrum disorder (ASD) according to estimates from CDC's Autism and Developmental Disabilities Monitoring (ADDM) Network. [Read summary](http://www.cdc.gov/features/new‐autism‐data/index.html) [Read article]

ASD is reported to occur in all racial, ethnic, and socioeconomic groups. [Read summary] [Read article]

ASD is about 4.5 times more common among boys (1 in 42) than among girls (1 in 189). [Read article]

Studies in Asia, Europe, and North America have identified individuals with ASD with an average prevalence of between 1% and 2%. [Data table](http://www.cdc.gov/ncbddd/autism/documents/asdprevalencedatatable2016.pdf)

About 1 in 6 children in the United States had a developmental disability in 2006‐2008, ranging from mild disabilities such as speech and language impairments to serious developmental disabilities, such as intellectual disabilities, cerebral palsy, and autism. [Read summary](http://www.cdc.gov/ncbddd/developmentaldisabilities/features/birthdefects‐dd‐keyfindings.html)

Who has special needs?

Autism Spectrum Disorders

- Maybe verbal or non verbal
- May demonstrate stimming like flapping hands or repetitive behaviors
- May squeal or hum
- May get under chair or try to get in cabinet
- May touch walls
- May avoid eye contact
- May not respect personal space

Visual Conditions in Autism

- Myopia, Hyperopia, Astigmatism
- Strabismus
- Visual perceptual disorders.
- Vision information processing deficits.
- Side effects from medications: Dry eye, accommodative problems.

Autism Spectrum Office Behavior

- May be verbal or non verbal
- May demonstrate stimulating like flapping hands or repetitive behaviors
- May squeal or hum
- May get under chair or try to get in cabinet
- May touch walls
- May avoid eye contact
- May not respect personal space

Sensory challenges

- Eye exam can be frightening.
- Preparing the child for what to expect can set the stage for a more successful encounter
- As a doctor you may need to sit on the floor, or let the parent hold the child in another chair instead of the exam chair
- Choose your tests wisely, if the patient becomes agitated more than one visit may be needed to get the information needed to evaluate ocular health and refractive conditions.
- Children on the spectrum may be more sensitive to light, sounds, smells and distracted by too many people in the room.
Common Autism Spectrum disorders
Conditions that may have ocular involvement that present here every day.

- Attention Deficit Disorder
- Attention Deficit Hyperactivity Disorder
- Oppositional Defiant Disorder
- Autism Spectrum Disorders

What causes ADD or ADHD?
- Poor parenting?
- Poor prenatal care?
- Prenatal drug and/or alcohol abuse?
- Poor education?
- Genetics?
- Chemical imbalances?
- All of the above?
- None of the Above? IT DEPENDS?

What to watch for?
Expected attention span for focused task should be about one minute for each year of age. A 3 year old should have a 3 minute attention span. A ten year old should be able to pay attention for ten minutes.

Attention problems may present as active or passive or a combination of both.

Active presentation is that child that must touch everything. This child may wiggle during the exam, have trouble keeping their entire body still, may tap feet, tap fingers and talk constantly. This behavior is less tolerated and demands attention from teacher, parents, doctors and therapists.

Inactive presentation is the child that looks out the window daydreaming and seems to be "in their own world." You may ask them a question and they will give you a blank look. The child may also have some restless body support. Because this behavior doesn't disturb others these children often fall behind in their school work without being noticed.

Active presentation
- Fidgets with hand or clothing
- May be on a cell phone engaged in a video game and block out all other input visually or otherwise
- Not aware of "personal space" may get in your face or react aggressively if you get in theirs
- Constant talking
- May cooperate better if a slow direct approach is taken with explaining what is about to happen at each step. "I want you to follow this toy, I am going to come close to you with this bright light."

Inactive presentation
- May be very shy or frightened about eye exam
- May respond with crying
- May not make eye contact or cooperate with verbal instructions
- May not have verbal communication to answer questions. When taking visual acuity they may not be able to read pictures: Heart, Bird, Cake, but if asked to point to the bird they may be able to successfully respond. A matching card with shapes can be used as well they may be able to match even if the language development is absent.

Case History General
- Ask developmental questions. Was the child born prematurely at low birth weight? Were there complications with the pregnancy? Any history of drug or alcohol use during pregnancy?
- Motor questions. When did the child crawl? When did the child walk?
- Does the child take medication?
- Does the child have other issues that require speech, occupational or physical therapy?
- Is the child a picky eater?
- Does the child sleep?
Behavior Medications

Common to encounter medications used to help children stay calm, focus their attention and to rest at night.

CLASS
Anti-psychotics or Neuroleptics

DRUGS: Haldol, Mellaril, Stelazine, Thorazine

BENEFIT: Reduce agitation, anxiety, aggression, hyperactivity, stereotypic and self-stimulatory behaviors, and temper outbursts

SIDE EFFECTS
Addiction
Blurred vision
Dyskinesia
Psychosis
Sedation
Tremors

Typical Neuroleptics

CLASS: Typical Neuroleptics

DRUGS: Abilify, Clozaril, Geodon, Risperdal Seroquel, Zyprexa

BENEFIT: Reduce aggression, agitation, self injurious Behavior

SIDE EFFECTS
Agitation, Increased appetite
Lowers white blood cell count
Tardive dyskinesia

Anti Depressants

DRUGS: Anti-depressants Anafranil, Celexa Elavil, Lexapro, Luvox, Paxil, Prozac, Tofranil, Wellbutrin Zoloft

BENEFIT: Raise serotonin levels, Reduce anxiety
Reduce obsessive-compulsive & ritualistic behaviors

SIDE EFFECTS
Arhythmias
Blurred vision
Constipation, Dry mouth
Dizziness
Hyperactivity & Impulsivity
Lowered threshold for seizures
Sleep disturbances

Stimulants

DRUG: Adderall, Cylert / Daytrona patch Dextedrine, Focalin, Ritalin

BENEFIT: Affect dopamine
Improve focus and regulation
Monitor arousal system
Decrease impulsivity

SIDE EFFECTS
Depression
Increase in perseveration & repetitive behaviors
Irritability, Palpitations
Sleep disturbance
Stimulants

<table>
<thead>
<tr>
<th>Trade Name</th>
<th>Generic Name</th>
<th>Approved Age</th>
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<tbody>
<tr>
<td>Adderall (XR)</td>
<td>amphetamine</td>
<td>3 and older</td>
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<tr>
<td>Concerta</td>
<td>methylphenidate (long acting)</td>
<td>6 and older</td>
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<tr>
<td>Dexmethylphenidate</td>
<td>methylphenidate hydrochloride</td>
<td>6 and older</td>
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<tr>
<td>Dexedrine</td>
<td>desmethylamphetamine</td>
<td>3 and older</td>
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<tr>
<td>Dextrostat</td>
<td>desmethylamphetamine 3 and older</td>
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<tr>
<td>Focalin (XR)</td>
<td>methylphenidate (long acting)</td>
<td>6 and older</td>
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<tr>
<td>Metadate ER</td>
<td>methylphenidate (extended release)</td>
<td>6 and older</td>
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<tr>
<td>Metadate CD</td>
<td>methylphenidate (oral solution and chewable tablets)</td>
<td>6 and older</td>
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<tr>
<td>Methylin</td>
<td>methylphenidate (oral solution and chewable tablets)</td>
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<td>Ritalin</td>
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<td>Strattera</td>
<td>atomoxetine</td>
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<td>Vyvanse</td>
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Anti-hypertensives

**DRUGS:** Clonidine, Tenex
**BENEFITS:** Calm and improve sleep
Decrease hyperactivity & impulsivity
**SIDE EFFECTS:** Irritability, Lower blood pressure, Sedation

Dopaergic Re-uptake inhibitors

**DRUG:** Depakote, Dilantin, Keppra, Phenobarbitol, Tegretol, Trileptal
**BENEFITS:** Calm behavior, Lessen mood swings, outbursts
**SIDE EFFECTS:** Affects kidney function
Vision impairments such as rapid eye movements

Visual Acuity Matching

- Product available at Goodlite.com
- Can hand the child the cards with picture and have them place them on the card at near distance point to the picture and have them hold up the correct card or point to the matching picture on the near card as the distance picture is held up.

Case History Ocular

- Does the child struggle to see near or far?
- Does an eye turn in or out?
- Is the eye turn constant or intermittent?
- Does the eye turn alternate of stay with the same eye?
- Any trauma?
- Any surgery?
- Any styes or eye rubbing?
Anterior Segment Evaluation

- Slit lamp, Burton Lamp or
- 20 diopter lens and transilluminator, penlight.

Posterior Segment Evaluation

- Direct ophthalmoscopy
- Direct ophthalmoscopy with 20 D if pupils are large 6-7mm
- Slit lamp with 90 Diopter lens
- Fundus photography. Not always possible to complete this due to ability to fixate. Reported success with children as young as age 2.

Posterior segment exam

Fundus Photo

Estimation of Refractive Status

- Retinoscopy, Distance and Near
- Autorefraction
- Welch Allen spot screener, user friendly and a good estimation.
- Subjective

Retinoscopy
Visual conditions

- Myopia, Hyperopia, Astigmatism: Objective tests may prove more reliable than subjective tests. A proper visual correction may be key to improving outcome. Patient in school for Autism no longer needed special services after being prescribed -5.00. His sensory delays were overcome after his visual correction was applied.
- Amblyopia: Impairs acuity, depth perception and contrast sensitivity. May present as refractive, strabismic or a combination of both.
- Strabismus: Esotropia, Exotropia, Hypertropia, Hypotropia
- Visual perceptual disorders.
- Vision information processing deficits.

Exotropia, Infantile

Exotropia, with telecanthus

Measuring intrapalpebral distance

Eyes should show symmetry

Measure from the outer canthus to the inner canthus on each eye to get the size of the palpebral fissure. Measure inner canthus to inner canthus to get the interpalpebral distance.

Binocular Dysfunctions

- Convergence insufficiency
- Convergence excess
- Divergence insufficiency
- Divergence excess
- Accommodative insufficiency
- Accommodative Spasm
Ocular Motor Dysfunctions

- Ocular motor dysfunction of Saccadic Movements
- Ocular motor dysfunction of Pursuit Movements

Special Needs Defined

- Non verbal: infants, children with speech delays, stroke patients that mastered language at some point and then suffered a loss.
- May not be cooperative. Objective testing must be performed quickly with accuracy.

Autism Spectrum Disorders

- Asperger Syndrome
- Attention Deficit Disorder
- Attention
- Deficit Hyperactivity Disorder
- Autism
- Rett’s Syndrome
- Pervasive Developmental Disorder

Autism Diagnosis

https://www.autismspeaks.org/what-autism/diagnosis/dsm-5-diagnostic-criteria

- Autism speaks website link
- Some children may be already diagnosed with Autism when you meet them
- Other children may give you signs and symptoms that merit referral for diagnosis

Indications for Autism Referral

- Lack of eye contact
- Interacting with humans like they are objects or tools
- Restrictive repetitive patterns for example lining up toys, flipping objects, echolalia
- Inflexibility in routines, rituals, examples: having a meltdown if the wrong spoon is used, only being able to drink out of one cup, unable to go to bed without completing all the bedtime steps.
- Lack of speech language development

Stimming

- Stimming
  - Common to flap hands or fingers, rock back and forth, seeking motor stimulation and feedback.
Pediatric Ocular Manifestations of Systemic Disease and Neurodevelopmental Syndromes

Course Description
Children may present with medical conditions and disorders with known ocular manifestations. A review of common medical, genetic, metabolic and traumatic issues which may present in a clinical setting. Chronic conditions such as diabetes and high blood pressure are observed more and more in the pediatric population. Genetic disorders such as Marfan's Syndrome, Fragile X Syndrome, Turner Syndrome, Edwards Syndrome, Down Syndrome, Klinefelter Syndrome are prevalent in the special needs population and present with special visual challenges. Neurodevelopmental disorders such as Aicardi Syndrome, Angleman Syndrome, Bechet's Disease, Cerebral Palsy, Fetal Alcohol Syndrome, Prader-Willi Syndrome, Rett Syndrome and Stickler Syndrome will be reviewed for diagnosis and management for optimal management and positive visual outcomes.

Chronic Metabolic Conditions
Diabetes and High Blood Pressure are increasing in prevalence as poor eating and fitness habits in all ages of people lead down the road to OBESITY

Diabetes
- Diabetes is prevalent and diagnosed at an early age.
- Retinopathy is more likely to appear if sugar levels are poorly controlled and diabetes has been present more than ten years.

Prevalence of Diabetes
- About 208,000 people younger than twenty years have diabetes (type one or type two)
- This represents 0.25% or 1 in 400 of all people in this age group
- 18,346 youth are newly diagnosed with type one diabetes annually
- 5,089 youth are newly diagnosed with type two diabetes annually

Diabetic Retinopathy
- Diabetic retinopathy can be classified as non proliferative diabetic retinopathy (NPDR)mild, moderate, severe or proliferative.

![Diabetic Retinopathy Images]
Mild Diabetic Retinopathy
- Small vessels are beginning to leak
- Small dot blot hemorrhages will be evident
- Crossing defects present
- Tortuosity and vessel attenuation becomes more visible
- Only one dot blot hemorrhage needs to be observed to meet this criterion

Moderate NPDR
- More than microneurysms but less than severe presentation
- Could be some exudates, cotton wool, larger hemes, any combination any location

Severe NPDR
- "Blood and Thunder" fundus appearance
- Hemorrhages of all kinds, dot blot, flame, boat
- Exudates
- Cotton Wool
- Venous Beading
- A little bit of everything EXCEPT Neovascularization
Proliferative diabetic retinopathy (PDR) is the more advanced form of the disease. At this stage, circulation problems deprive the retina of oxygen. As a result, new, fragile blood vessels can begin to grow in the retina and into the vitreous, the gel-like fluid that fills the back of the eye.

Diabetic Retinopathy - American Optometric Association
aoa.org/patients-and-public/eye-and-vision-problems/glossary-of-eye-and-vision-conditions...

Genetic Disorders

Marfan’s Syndrome, Fragile X Syndrome, Turner Syndrome, Edwards Syndrome, Down Syndrome, Klinefelter Syndrome

Marfan Syndrome

Marfan’s Syndrome

Marfan Syndrome
Marfan Syndrome

- **Eyes**: nearsighted (myopic), eye (or cataract) lens dislocation, retinal detachment
- **Lungs**: spontaneous lung collapse (pneumothorax)
- **Cardiovascular System**: aortic widening or dilatation, aortic aneurysms, mitral valve prolapse / leakage
- **Skeleton**: curvature of the spine (coliosis), pseudo or funnel chest (pectus deformity), tall stature (tall stature)

[Image: https://www.pinterest.com/pin/115756652895801791/]

Fragile X Syndrome

- Proper test: hpgfxxghubh dfrvhir@phahwodak gldekle
- Proper test: prwirtp p qphbqz qth ljgwhfw @f dhrhup
  5K | Vfdshwhdhvpfogeh = surp hghp#
  hdru/p dfrwshkw /Adhrq | @hfrh /p @hfrh#
  K | srsevli/dlrkjkshfkhqir@shui/kr shui/krqlehk#
  mhrh/shufhrkhu/#
  http://shufhrkhu/#
  mhrh/shufhrkhu/#


Fragile X


Turner Syndrome

Turner Syndrome

Edward's Syndrome

Trisomy 18, also called Edwards syndrome, is a chromosomal condition associated with abnormalities in many parts of the body. Individuals with trisomy 18 often have slow growth before birth (intrauterine growth retardation) and a low birth weight. Affected individuals may have heart defects and abnormalities of other organs that develop before birth. Other features of trisomy 18 include a small, abnormally shaped head; a small jaw and mouth; and clenched fists with overlapping fingers. Due to the presence of several life-threatening medical problems, many individuals with trisomy 18 die before birth or within their first month. Five to 10 percent of children with this condition live past their first year, and these children often have severe intellectual disability.


Edwards Syndrome

Down Syndrome


http://2.bp.blogspot.com/-oCmJ0NXkVt0/UGrsb5c8G_I/AAAAAAAAAQ0/aUHYQ9i0cwc/s1600/brushfield+spots001.jpg
Klinefelter Syndrome

Facts on Klinefelter Syndrome
Cause: Boys born with an extra X chromosome
Frequency: 1 in 1000 males.
Characteristics: Learning problems, tall stature, small testes, breast development, infertility, skeletal abnormalities.
Treatment: Psychosocial care, testosterone replacement therapy, infertility treatment.


Klinefelter Syndrome


Neurodevelopmental disorders

Aicardi Syndrome, Angleman Syndrome, Bechet’s Disease, Cerebral Palsy, Fetal Alcohol Syndrome, Prader-Willi Syndrome, Rett Syndrome and Stickler Syndrome

Aicardi Syndrome

Dysgenesis of the corpus callosum
Infantile spasms epileptic like seizures, occipital flattening, mental retardation, end stage kidney failure, hearing loss.
Optical Manifestations: Chorioretinal lacunae, optic nerve colobomas, optic nerve hypoplasia.

Aicardi Syndrome

http://dxline.info/img/new_ail/aicardi-syndrome.jpg

Optic Nerve Hypoplasia

Optic Nerve Hypoplasia

This is a tiny nerve. It will be a long way to the macula from here in disc diameters (5 disc diameters to the fovea). Macular area may appear much larger than the disc.

Normal Nerve

Angelman Syndrome

Bechet’s Disease

Angelman Syndrome

Bechet’s Disease I

Diagnostic Criteria

Recurrent oral ulcers: Aphthous ulcers noted by patient/physician, 4 episodes in 12 month period

Plant 1 of Inflammation:

Recurrent genital ulcers: Aphthous ulcers on anogenital skin noted by patient/physician

Eye lesions: Anemia/pigmentary reti- na changes noted by ophthalmologist

Skin lesions: Aphthous ulcers noted by patient/pain/discoloration noted by ophthalmologist

Pathergy test: Incision noted at 48-49 hours
Cerebral Palsy

Disorder of movement and posture secondary to damage to motor control connections

Hypotonicity, neurologic impairment

Strabismus, nystagmus, optic nerve pallor, cataracts, myopia

Fetal Alcohol Syndrome
Fetal Alcohol Syndrome

- Small head
- Low nasal bridge
- Epicanthal folds
- Small eye openings
- Flat midface
- Smooth philtrum
- Underdeveloped jaw

Telecanthus

- Narrow temple distance and nasal bridge
- Almond-shaped eyes
- Mild strabismus
- Thin upper lip
- Downturned mouth
- Overweight

Prader-Willi Syndrome

- Deletion of paternal genetic material on chromosome 15
- Obesity, hypotonia, short stature, learning disability
- Strabismus, almond shaped palpebral fissures, myopia

Rett Syndrome

- Mutation of binding protein (MECP2) that alters the development of gray matter
- Deficits in motor control and communication skills
- Observable hand wringing behavior
- Difficulty maintaining eye contact
Rett Syndrome

Stickler Syndrome

Stickler Syndrome

Conclusion

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