Genetics and Care of the School Age Child

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Objectives

Describe the most common genetic conditions impacting the school age child

Recognize signs & symptoms of 2 common genetic conditions

Identify 3 national genetic resources

Do you know someone with a genetic disorder?

3% of all babies are born with a birth defect

1 in 9 children admitted to the hospital has a genetic disorder

1 in 12 adults admitted to the hospital has a genetic disorder

1 in 22 people in the US has a genetic disorder

50% of people with intellectual disability have a genetic basis

Common genetic conditions impacting the school aged child

- Chromosome abnormalities
  - Down Syndrome 1:700 children
  - Velo Cardio Facial Syndrome ~1: 2000 - 4000
- Sickle Cell Anemia: 1 in 625
- Cystic Fibrosis 1:3300
- Neurofibromatosis 1: 3500
- Duchenne Muscular Dystrophy 1:3500
- Marfan Syndrome affects 1: 5000 people

Why have a Genetic Evaluation?

1. Establish diagnosis
2. Anticipatory guidance
3. Medical management
4. Developmental intervention
5. Behavioral intervention
6. Psychological management
7. Recurrence risk

Velo Cardio Facial Syndrome (VCFS) or 22q11.2 deletion syndrome

VeloCardioFacial Syndrome (VCFS) is also called DiGeorge Syndrome or 22q11.2 deletion syndrome

Velo Cardio Facial Syndrome Characteristics

Affects Males and Females

Over 180 physical & developmental characteristics reported!

-Heart defects: (80%) (Ventricular Septal Defect, Aortic arch abnormalities , Tetralogy of Fallot, Truncus Arteriosis)

-Cleft palate (75%)

-Renal abnormalities (>30%)

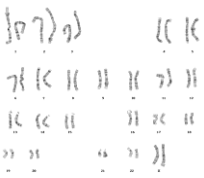
Velo Cardio Facial Syndrome or Deletion 22q11.2

DiGeorge Syndrome	Velocardiofacial Syndrome
Presenting in infancy	Childhood/adulthood
Severe heart defects ◦ Often lethal	Heart defects ◦ Usually mild
Severe infections ◦ Immunodeficiency	Weak palate; cleft palate ◦ Hypernasal voice
Seizures ◦ Low calcium levels	Long face and fingers

Velocardiofacial syndrome (VCFS)

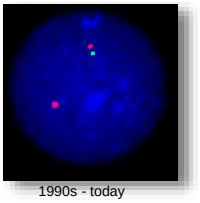
- Most common microdeletion syndrome in humans
- Too small to be seen with routine chromosome studies, new chromosome microarray test used
- 20-30 genes in a row are deleted

Traditional cytogenetic tests



1960s - today

Chromosome analysis
(under the light microscope)

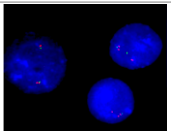


1990s - today

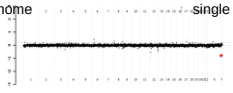
FISH analysis
(Fluorescent in situ hybridization)

Newer testing

Karyotype [4-5 Mb, whole genome

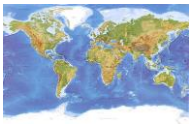


FISH [40 to 250 kb per probe, single site]



Chromosome MicroArray (2004-today)
[average resolution ~5 kb, whole genome]

How testing has improved!



Chromosome analysis

World Map
17,500 miles

Chromosome microarray analysis

Map of Houston
17.5 miles

Microdeletions 22q11.2: Velocardiofacial Syndrome

-Learning disabilities in 70-90%
(math concept, reading comprehension)

- Psychiatric illness in >40%- anxiety, ADHD, autism spectrum, schizophrenia

Psychiatric illnesses

Schizophrenia, bipolar disorder and anxiety disorder may start in teen years

Manifestations may range from mild to severe

Respond to treatment

Characteristics

- No feature occurs in all children
- No child has all of these features.
- The medical, developmental & psychological features are very different from person to person.
- Range from mild to severe

Diagnosis of Deletion 22q11.2
1 in 2,000 - 4,000

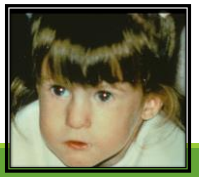
Birth	30%
By age 5 years	70%
By age 18 years	95%

Some people with deletion 22q11.2 are *never* diagnosed!

Evaluation and Diagnosis

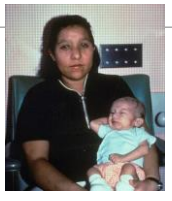
Physical exam and presence of signs and symptoms of VCFS

Blood test: Chromosome microarray testing



Inheritance

Autosomal Dominant



~ 95% are new deletions

~ 5% are inherited from a parent

- Because of variability, parent may not have been diagnosed

Genetic Counseling

If inherited, each sibling has a 50% risk to be affected

If new deletion, siblings have low risk

Any child of the affected person will have a 50% chance to be affected

www.positiveexposure.org

www.vcfstexas.com

Treatment

Depends on symptoms:

- Surgery to correct cleft palate and/or heart defect
- Speech therapy
- Psychological counseling, psychiatric care
- Medication

Knowing the diagnosis

Early intervention may impact the long term outcome

May help family and teacher know educational strengths and weaknesses

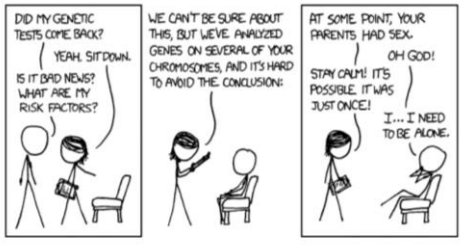
Help obtain needed therapies

Dispel misinformation

Screen for association complications or disabilities before symptoms seen

VCFS Resources

- International 22q Foundation: www.22q.org
- 22q and You: http://www.chop.edu/service/22q-and-you-center/home.html
- VCFS Texas: www.vcfstexas.com



Marfan Syndrome

Speaking out about Marfan Syndrome

Marfan Syndrome

Affects connective tissue of the body:

Eyes, heart, lungs, skin, skeletal system

Affects males and females

What is the Cause?

Mutation or change in fibrillin-1 (FBN1) gene on chromosome 15.

Skeletal abnormalities

Long narrow face with high arched palate

Disproportionately long fingers and limbs

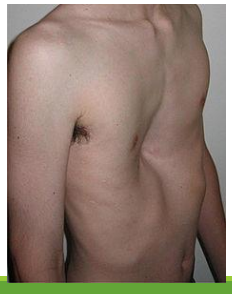
Chest abnormalities: pectus excavatum or pectus carinatum

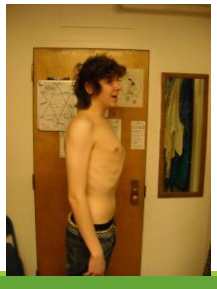
Scoliosis- seen in about 50%

Joint hypermobility

www.marfan.org

Pectus Excavatum Pectus carinatum





Cardiac Features

Aortic dilatation and aortic aneurysms

Predisposition for aortic tear and rupture

Mitral valve prolapse: 'billowing motion of mitral valve'

Aortic regurgitation: valve not fully closing and blood leaks back into heart

Eye Findings

Dislocated Lens

Severe Myopia

Detached Retina

www.marfan.org

Evaluation:

Physical exam

Family history

Echocardiogram

Ophthalmologic exam

Diagnosis of Marfan Syndrome

Diagnosis based on clinical criteria, not genetic testing

Fibrillin gene on Chromosome 15 causes Marfan syndrome. Over 300 mutations in this gene have been found.

Children also evaluated for a newly diagnosed disorder, Loeys-Dietz syndrome which includes features in common with Marfan Syndrome

Inheritance

Autosomal Dominant

75% have an affected parent

25% due to a *new mutation* or change

Genetic Counseling

If familial, siblings have a 50% risk

If new mutation, siblings have low risk

Any child of the affected person will have a 50% chance to be affected

Treatment includes regular visits to:

Cardiologist

Orthopedist

Ophthalmologist

Geneticist

May need appointments with orthodontist and pulmonologist

Treatment may include:

Antihypertensives: beta blockers: to lower BP and reduce force of heart beats

Surgery for scoliosis or chest deformities

Anticoagulants if have mechanical valve

Headache and/or pain management

Antidepressants

Physical Activity Guidelines

Recommend non-contact, non-strenuous, non-competitive activities

Encourage brisk walking, slow jogging, cycling on level ground, shooting baskets, slow paced tennis.

Keep heart rate < 100.

Avoid heavy backpacks, may want to have a 2nd set of text books at home or use rolling backpack.

Athletics

Avoid weight lifting, competitive sports

Guide children away from sports at a young age

Help PE teachers develop adaptive PE plan, do not test the child's physical limits

The School Nurse

Screening: vision, posture, Pre-Sports physicals

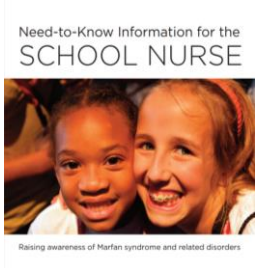
Referrals: convey need to parents, help with referral, follow-up

Manage medication, psychosocial care.

Encourage activities other than sports: computers, music drama, team managing

The Marfan Foundation

http://www.marfan.org/resources/patients/school



Have emergency plan

Marfan Syndrome is a manageable medical condition

Teens almost never have an aortic dissection

How to recognize emergencies

Aortic rupture or dissection: rare in school aged child. Usually painful, has been described as 'tearing pain boring through'. May have syncope or shortness of breath.

Pneumothorax: shortness of breath, chest pain worse with deep breath

Retinal detachment: flashing lights, spots in vision, sudden loss of vision

Roles and Responsibilities of All Nurses

ASSESS and IDENTIFY: patients who may benefit from a genetic evaluation

REFER: know how to make appropriate referrals, prepare family for consult

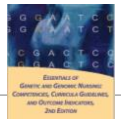
SUPPORT: providing initial and ongoing care and support

EDUCATE: prepare patients for specialized testing,
help families process the information learned

COORDINATE: testing, procedures, follow–up.

RESOURCES: Find trusted accurate resources and help families to learn of support groups and clinical trials

Core Competencies



Core Competencies for All Health Professionals 2007
www.nchpeg.org

Essentials of Genetic and Genomic Nursing: Competencies, Curricula Guidelines and Outcome Indicators, 2nd Ed, 2008

https://www.genome.gov/27527634#al-1

Where to find genetic services?

Families can be referred to genetic professionals by any health professional or self-referred depending on insurance.

American College of Medical Genetics www.acmg.net

Genetic Providers in Texas
http://www.dshs.state.tx.us/genetics/provider.shtm

Adult Genetics clinic: Baylor Clinic: 713-798-7820

Pediatric Genetics clinic: Texas Children's Hospital, 832-822-4293

University of Texas Medical Genetics: 713- 500-6727

Web resources

Genetics Education Materials for School Success: GEMMS- www.gemssforschools.org

Genetics Home Reference: http://ghr.nlm.nih.gov

National Organization for Rare Disorders: www.rarediseases.org

Unique : www.rarechromo.org

Web Resources

- Gene Tests www.genetests.org
- Genetics and Rare Diseases (GARD) Information Center: http://rarediseases.Info.nih.gov/GARD/
- ISONG.org- International Society of Nurses in Genetics
- National Coalition for Health Professional Education in Genetics (NCHPEG) www.nchpeg.org

My Family History Tool

https://familyhistory.hhs.gov/fhh-web/home.action

Summary

Genetic conditions impact a significant portion of the general population.

Children with 22q11.2 deletion syndrome and Marfan syndrome need specific management and care

Nurses have an important role in assessing, referring, coordinating, evaluating and educating individuals, families.

You are not expected to "know it all", but need to know how to access accurate information!

References

- American Academy of Pediatrics Committee on Genetics "Health supervision for children with Marfan Syndrome." *Pediatrics September 2007; 120:3 683-684; doi:10.1542/peds.2007-1643*
- Bassett, A.S., et al, Practical Guidelines for Managing Patients with 22q11.2 Deletion Syndrome *J Pediatr.* 2011 Aug;159(2):332-9.e1.
- Vorstman, Jacob A.S. et al. "A Cognitive Decline Precedes the Onset of Psychosis in Patients with the 22q11.2 Deletion Syndrome." *JAMA psychiatry*72.4 (2015): 377–385. *PMC*. Web. 4 Apr. 2016.