

GI Issues in Dup15q Syndrome

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Gastrointestinal Basics

Three basic tasks:

1. Digestion
2. Absorption
3. Motility

DIGESTION

Taking what in what is needed and then breaking it down into the basic nutrients:

Fat

Protein

Carbohydrate

Miscellaneous others: vitamins, minerals, fiber

No studies have shown any basic difficulty with digestion of any of the basic nutrients in autistic disorders

Problems with Intake

Appetite driven by three factors in humans:

Glucose

Volume primarily stomach

Mental

Compiled data into the brain where processed to create appetite/hunger

Intake Issues

Multiple studies have documented intake issues in children with Dup 15q

No glucose issues

Volume issues? (More later)

Mental issues: Functionally defined feeding problems

Food Refusal: Don't get enough calories

Food Selectivity by Type: Eats only narrow range of nutritionally inappropriate foods

Food Selectivity by Texture: Don't eat developmentally appropriate food textures

Food Refusal

Problem with the central processing and integration

All evaluations are normal for any biologic issue

"Programming" says that intake/weight is fine

Interventions: medication, supplements, feeding tube

Metabolism

Either catabolic (↓) or anabolic (↑)

Children programmed for growth, adults can be static

Need to be anabolic to be healthy

GI Correlates of childhood feeding problems

Gastro-esophageal reflux was the most prevalent condition found among all children in the sample and was the factor most often associated with food refusal

Children with gastro-esophageal reflux had more severe feeding problems and higher prevalence of food refusal and dysphagia

Food Selectivity by type

Sensory issues effect preferences

Color, taste, etc.

Some very limited and skewed diets

Plenty of calories but nutritionally imbalanced

Food Selectivity by texture

Sensory as well

Prefer smooth to course

Lower to higher

Sometimes by type: crunchy versus chewy

ABSORPTION

No studies have demonstrated any difficulties with any transporters in the GI tract

No apparent absorption issues

Old concerns about inflammation and malabsorption

Type of IBD

Celiac disease

Casein and gluten sensitivity

MOTILITY

What goes in must eventually come out

Peristalsis is another description

GI tract has several muscle layers

Purpose:

Organized contractions to move nutrients along

Also to mix with digestive fluids

Housekeeping function

Motility Regulation

Intrinsic: Enteric nervous system, baseline rate of motility

Foods themselves: Carbohydrates fastest, proteins in the middle and fats slowest (alter diet alter motility)

Hormonal: Thyroid best example
Central nervous system
Regulate and modify the basal rate
Potential to slow down or speed up

SPECIFIC ISSUES

Constipation

Reported in 60% of children with 15q duplication (retrospective chart review)

Slow motility leads to constipation (colon's job to absorb water)

Selective dietary intake can lead to constipation

So does stool retention—potential for sensory issues and with-holding

Constipation Therapies

Alter diet?

Tend use stool softeners

Limited use of laxatives

Diarrhea

May be diet related

High juice intake

May be motility

Usually do diagnostic studies to ensure no other cause

Diarrhea Therapy

Alter diet?

Medication

Gastroesophageal Reflux (GER)

Reported in 56.7% of children with 15q duplication

Diagnosis cannot be made in infants on the basis of symptoms or cluster of symptoms

Etiologies

Primary versus secondary reflux

Primary

Lower esophageal sphincter (LES) relaxation

Impaired esophageal clearance

“Normal reflux”

Everybody has some

Increased with certain foods

Increased with overeating

Secondary reflux

Increased intra-abdominal pressure

Delayed gastric emptying

Mechanical impediments

Symptoms appear the same, treatments are different

Neurologic Influences

Previous data on “normal children”

Rates higher in children with neurologic difficulties

GER in 75% of neurologically impaired children

High rates of delayed gastric emptying with frequent vomiting

These are also the children with high rates of dysphagia

Primary or secondary?

Or both?!

Reflux Therapy

First line is acid suppression

What about surgical approach? (fundoplication)

Delayed Gastric Emptying

Vomiting/secondary reflux

Decreased appetite

Delayed Gastric Emptying Therapy:

Alter diet?

Medical therapies

If fundoplication get gas bloat or dumping

Intake Issues

No real appetite stimulant available (or being studied).

If malnourished may require feeding help

Behavioral interventions

Variety and insistence early on

Studies say even normal children require an average of 8 exposures to accept a food

Slow gradual introduction of new foods

Questions?