put together by Alex Yartsev: Sorry if i used your images or data and forgot to reference you. Tell me who you are. aleksei.igorevich@gmail.com

Prader-Willi Syndrome: 1 in 25,000

Clinical Manifestations:

Most obvious in infancy:

- hypotonic flaccid child, poor feeding which usually needs some sort of weird feeding tecdhniques
- gradually resolving flaccidity, feeding problems slowly improving after 12 months of age and becoming replaced by **UNCONTROLLABLE WEIGHT GAIN** before 6 years of age
- This is in part due to **UNCONTROLLABLE FEEDING BEHAVIOUR.** The child will forage for food around the house, obsessed with eating. Foods are preferentially carbohydrates.

Later:

- characteristic facial features develop
- secondary sexual characteristics fail to develop or are incomplete; eg. amenorrhoea, decreased facial and body hair, hypoplastic genitals
- Global developmental delay in the under-6s, and IQ of 50 to 70 in older children

Major diagnostic criteria are the above features, confirmed by a positive genetic test:

- **some sort of abnormality in chromosome 15q11-13:** Normally, you have a maternal and a paternal copy of 15(q11-13). The paternal copy is active while the maternal copy is methylated and inactive. The disorder results from either the absence of a paternal part (a deletion), mutation of the imprinting centre which is responsible for the methylation, or a maternal uniparental disomy where you end up with two identical maternal copies of the gene. Either way, the paternal copy is missing or useless.

Minor Features:

- osteoporosis
- Thick saliva
- Dental caries
- Sleep apnea
- Short stature
- Skin picking (resulting in sinister-looking little areas of missing skin)
- Small hands and feet
- Characteristic obsessive-compulsive behavior, punctuated with violent outbursts.
- Occasional stealing and lying

The reasons why these people die:

- Complications of obesity: They will present with sleep apnea, cor pulmonale, they get inchaemic heart disease earlier in life, they are young Type 2 diabetics.
- occasionally, because of **acute gastric dilatation and subsequent necrosis** (described in series of 6 pts by Whatron et al)
- **choking episodes** causing death in 7.9% of 152 patients (Stevenson et.al, 2007)
- **fractures** due to underlying osteoprosis

MANAGEMENT:

- surgeons may need to take care of the obstructive sleep apnoea and cryptorchidism.
- Replace calcium in the long term
- Manage the inevitable diabetes
- Monitor for scoliosis and cor pulmonale
- Behavioural issues may need to be managed with SSRIs. Interestingly, well-meaning attempts to replace pituitary hormones may aggravate the behavioural issues.
- Have a low threshold with PWS patients who present to emergency with sudden abdominal distension and pain.

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