

# Establishing the diagnosis

- Start with DNA methylation analysis - will diagnose >99%
- Also start with an oligo-SNP microarray which will pick up the deletion class and size, as well as microdeletions that include the IC & *SNORD116*, as well as 70% of uniparental disomy 15
- FISH will confirm a deletion if present, but will not reveal size of deletion & does not detect UPD or an IC deletion
- If DNA methylation positive, but the microarray or FISH is normal, then need to DNA polymorphism analysis
- See GeneReviews for details of PWS testing algorithm