- One gene copy active functional haploidy
- Isulin Growth factor 2 (Igf2) Paternally expressed
- H19 (a non-coding RNA) Maternally expressed
- Regulated by a cis-acting, Differentially Methylated Region (DMR) also called in impringin control region (ICR)
- The same enhancer controls their expression
- Doesn't matter if maternal lgf2 is mutated cos it wont be expressed.



## Pader-Willi Syndrome

- Deletion
- Obesity, short, small hands/feet, unsual facial features, mild mental retardation
- Compulsive overeaters
- Failure to imprint  $\rightarrow$  wrong level of maternal or paternal gene expression Paternal imbalance.

## Angelman Syndrome

- Speech impairment none or minimal use of words
- Receptive and non-verbal communication skills higher that verbal ones
- Movement or balance disorder, usually ataxia of gait