

# Prenatal Diagnosis of Beckwith- Wiedemann/Angelman Syndromes

John Johnson, Linda Beischel,  
Michelle Griffin, Jon Schoof,  
UCSF/CPMC/Genzyme

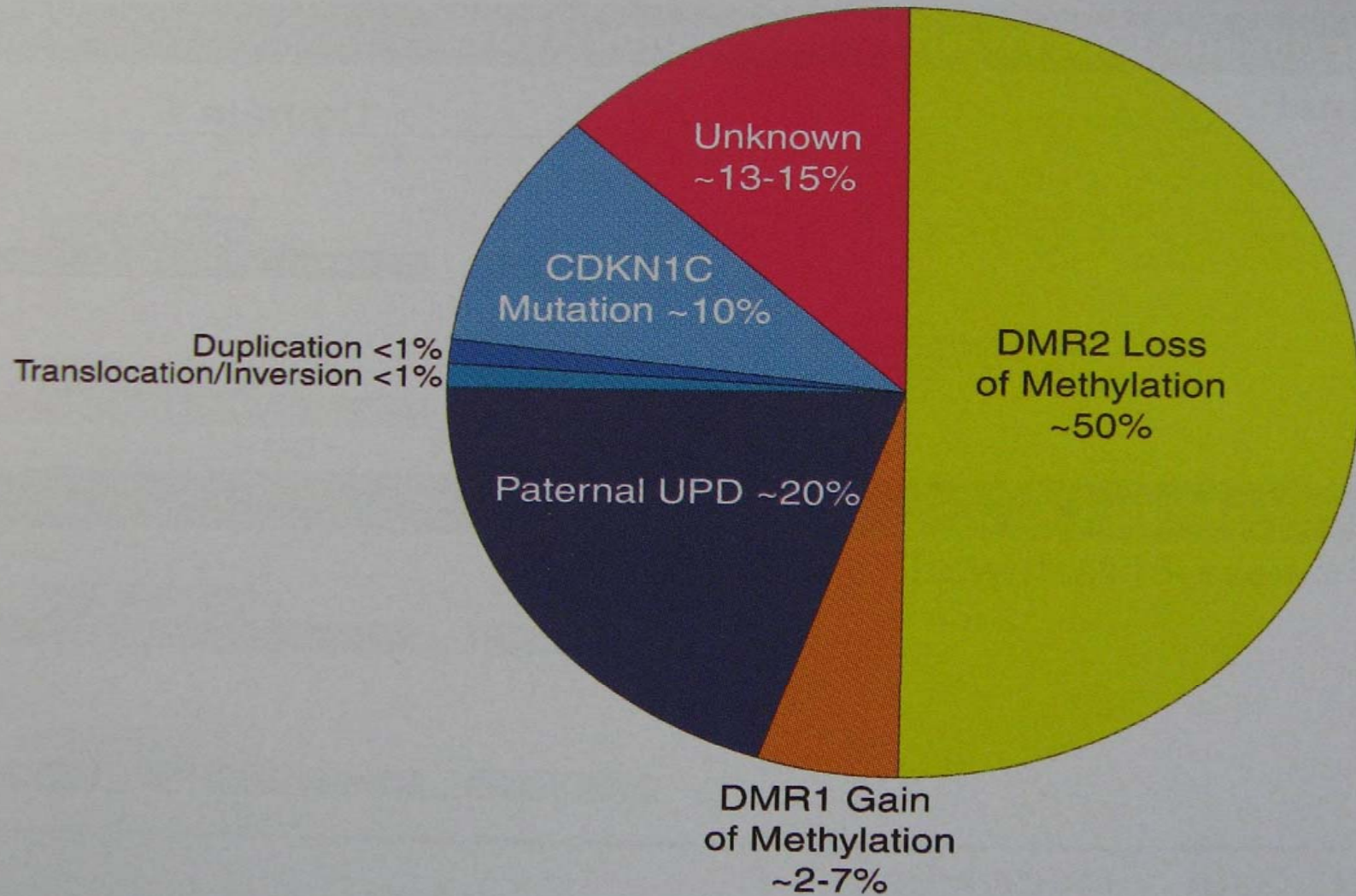


# Beckwith-Wiedemann Syndrome

- 1/10,000-1/15,000
- Overgrowth syndrome
- Cancer syndrome (5-10%)
- Disordered Imprinting



# Causes of BWS



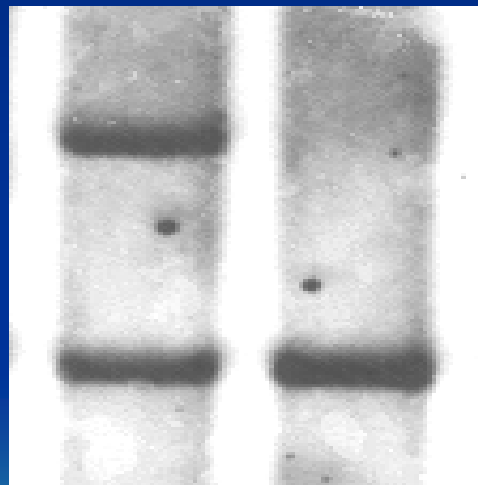
# LOI by Southern analysis

Probe for KCNQ1OT1

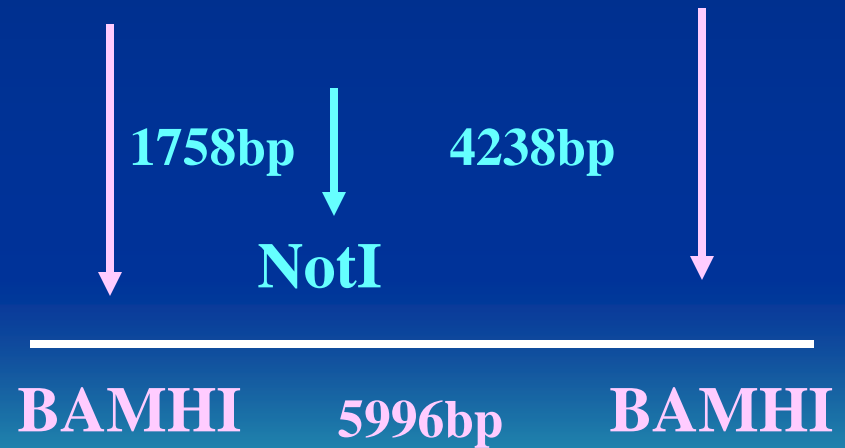
RE **NotI**- will not cut a methylated area

Mat 6.0

Pat 4.2



NL BWS



# IVF and IMPRINTING

- Sheep and cattle: IVF and LOS
- Olivenness 2001 1/73 IVF/ICSI births BWS
- Cox 2002 2 AS after ICSI, not UPD or 15q11 del
- Orstavik 2003 AS after ICSI, not UPD or 15q11 del
- DeBaun 2003 BWS registry, 4.6% ART (.8%)
  - 5/6 DMR2, 1 DMR1
- Gicquel 2003 6/149 BWS born after ART, all DMR2
- Chang 2005 10 BWS/IVF, 5 by ICSI, 1 IUI
- Shuman 2006 2 of 8 HH/LOI DMR2 were IVF

# Shodair IVF Experience

- First referrals 2003 from CPMC, Dr Main

- As of 3/31/09:

<b>BWS</b>	<b>Amnio</b>	<b>CVS</b>	
<b>IVF</b>	<b>464</b>	<b>166</b>	<b>1 POS (CVS)</b>
<b>ICSI</b>	<b>86</b>	<b>12</b>	
<b>Omphalocele</b>	<b>90</b>	<b>27</b>	<b>6 POS AM, 2 IVF 3 POS CVS, 2 IVF</b>
<b>AS</b>	<b>Amnio</b>	<b>CVS</b>	
<b>IVF</b>	<b>421</b>	<b>178</b>	<b>2 POS (AM), 1 IVF 1 AMA UPD 15</b>
<b>ICSI</b>	<b>79</b>	<b>11</b>	