





Birth incidence of Prader-Willi syndrome in France

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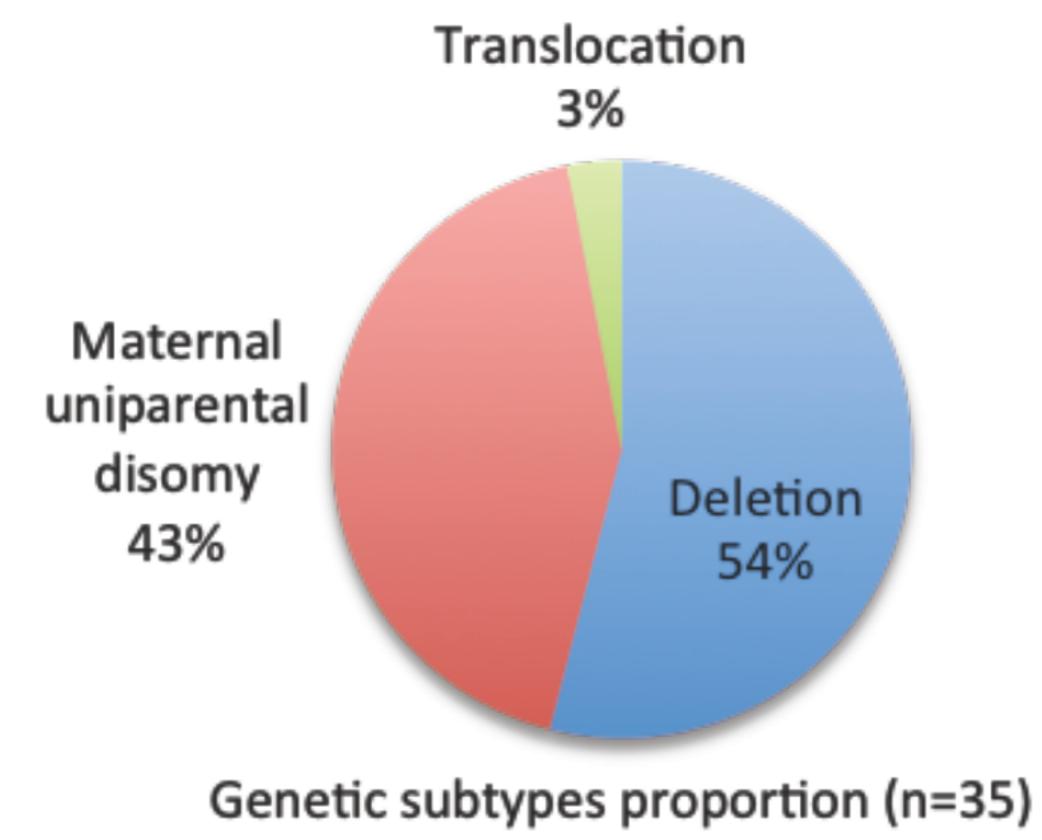
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Background: Prader-Willi syndrome (PWS) is a rare and complex genetic disorder characterized in neonates and infants by hypotonia and feeding problems. French birth incidence has never been reported.

Objective: To evaluate incidence of PWS at birth in France.

Method: Identification of patients with a molecular diagnosis of PWS born between January 1st, 2013 and December 31st, 2013 was obtained by combining various approaches in order to try to reach exhaustive results: e-mailing to members of the Pediatric Endocrinology French Society and Neonatology French Society, to genetics laboratories involved in the molecular prenatal and neonatal molecular diagnosis of PWS and to the French PWS patients association. In addition, number of live births was obtained from the annual report of the National Statistic Institute.

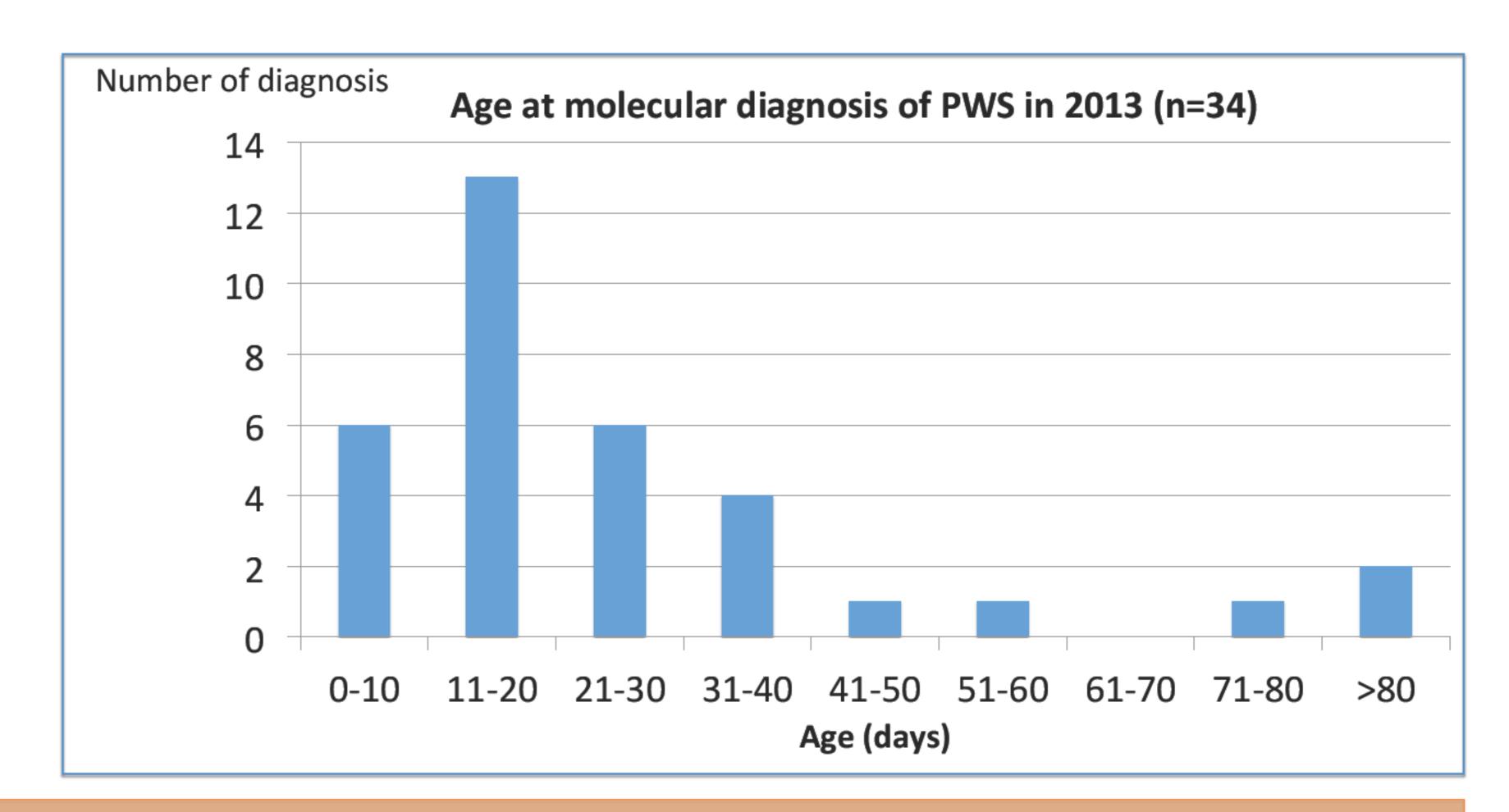
Results: 38 newborns with molecular diagnosis of PWS were identified in France in 2013. Three prenatal diagnoses were made during this period whose outcome was a pregnancy interruption, giving the number of 41 diagnoses per year. There were 781621 live births and 7961 lifeless births in 2013, birth incidence of SPW was therefore 1/19258. Molecular mechanism was known for 35 patients (85%). We found 54% of neonates having deletions, 43% having maternal uniparental disomy (mUPD) and 3% having a translocation.



Age at molecular diagnosis was available for 34 patients. Median age at molecular diagnosis was 17 days (range 3; 521). 91% were diagnosed within the first two months of life. A few missed early diagnoses were identified.



Regional birth incidence of PWS in France in 2013



Conclusion: Birth incidence of Prader-Willi syndrome in France in 2013 was close to 1/20000 live births. We confirmed the increasing proportion of mUDP. Median age at diagnosis was 17 days. Very few remained undiagnosed at this time, and improving care to these patients requires continuous efforts.



