Evolution of the French database of children with Prader-Willi Syndrome

Sandy Faye[1,2], Cathy Brochado[1,2], Catherine Molinas[1,2,3], Marie Dupuy[1], Julie Cortadellas[1,2], Dibia Liz Pacoricona Alfaro[1,4], Marion Valette[1,2,4], Mélanie Glattard[1], Christelle Noual[1], Sophie Çabal Berthoumieu[1], Maithé Tauber[1,2,3] & Gwenaelle Diene[1,4]
& All the Physicians of the 20 French PWS Competence Centres

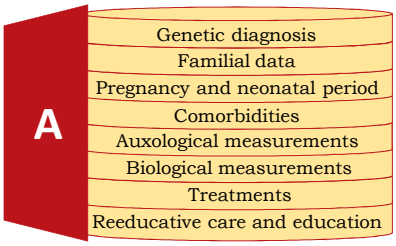
1: Centre de Référence du syndrome de Prader-Willi, Hôpital des Enfants, Toulouse, France; 2: Axe Pédiatrique du CIC 9302/INSERM, Hôpital des Enfants, Toulouse, France; 3: INSERM U1043, Centre de Physiopathologie Toulouse Purpan, Université Paul Sabatier, Toulouse, France; 4: INSERM, UMR 1027- Université Toulouse III Hôpital Paule de Viguier, Toulouse, France

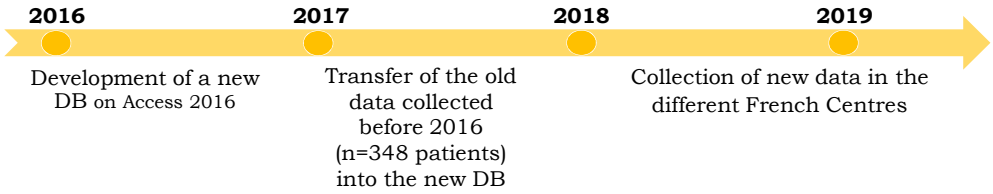
BACKGROUND

The French Reference Centre (RC) for Prader-Willi Syndrome (PWS) labelled by the French Ministry of Health in 2004 began a national register of children with PWS in 2005. In 2008, a cohort study in the different competence centres has been implemented in order to create a national database (DB). This DB includes medical, socio-demographic and familial data of children and adolescents with PWS.

METHODS

New French Database: > 400 variables





RESULTS

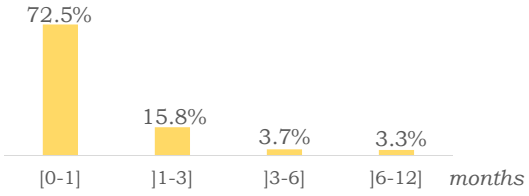
First data analyses of the **534 children** (age less than 18 years) among the 1372 identified patients

Sex ratio: **51.9% of boys**

Born between 2001 and 2018

Median age: 9.9 years (0.5 to 18 yrs)

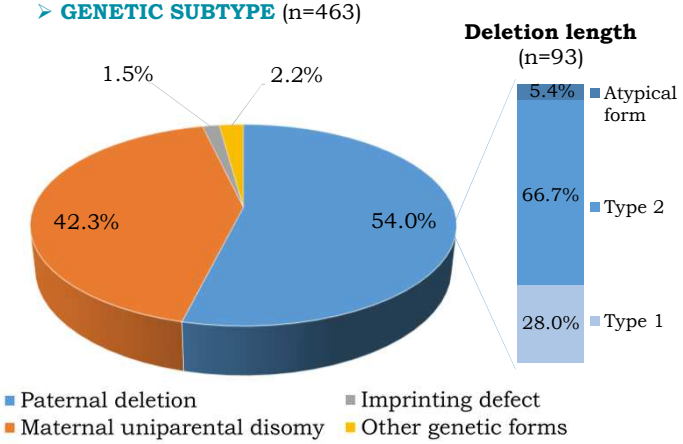
➢ AGE AT DIAGNOSIS (n=512)



Patients diagnosed in the first year of life: 95.3%
- during the **first month of life** (0.1 to 1 month): **72.5%**
- between 1 and 12 months: 22.8% with median age of 2 months

Children **born since 2014** (n=125):
80.8% diagnosed during the first month of life
51.4% deletion *vs* 43.1% disomy

➢ GENETIC SUBTYPE (n=463)



Deletion length (n=93)

➢ GROWTH HORMONE TREATMENT (n=507)

Patients **treated with GH: 95.9%**

Median age to introduce GH treatment (n=473):**16.2 months**

Evolution of median age at start of GH
- Children born 2001-2014 (n=366): 19.0 months (3.0 months to 14.7 yrs)
- Children born 2014-2018 (n=107): **11.3 months** (2.7 to 33.6 months)

>> Earlier start of GH treatment over time

CONCLUSION **This big database, covering different aspects of PWS clinical and social profiles (>400 variables), is a powerful tool for retrospective and prospective studies. The first analyses show an earlier diagnosis and start of GH treatment over time.**

Special thanks to Pfizer Laboratory and the French Prader-Willi Syndrome Association