The unique difficulties of rare disease patients to access health and social care in Europe and the role of the European Platform on Prader-Willi Syndrome (EurordisCare3): a summary

**RESULTS RELATED TO THE NEED FOR SOCIAL SERVICES**

1. Patient Profile
   - Patients with PWS (6% of the total) participated in the "eurordiscare3" survey conducted by Eurordis in 2006-2008. In this survey, there is a representation of 32 countries and a wide range of age groups. Countries which have had only 1 or 2 participants have been excluded from this study.
   - The majority of patients are from France, Belgium, Spain, Switzerland, and Germany.

2. Access to main medical services
   - 74% of patients needed an individual dietitian, but 12% of their care was impossible.
   - Lack of referral was the most common cause of impossible access. Other causes of access were the distance and the cost.

3. Access to paramedical services
   - 42% of patients stated that they needed social services (94.1% in Italy).
   - Unavailability of services is reported as the main cause of impossibility to access these services.
   - It is possible that facilities are not well informed about existing social programs.

4. Need for social services
   - 194 out of 367 (53%) needed a nutritional specialist during the last 12 months.
   - 75% of patients needed an endocrinologist, but for 4% the access was impossible.

**GENERAL DESCRIPTION OF THE MODEL FOR "EARY DIAGNOSIS"**

- Logistic regression was used to create a statistically significant model. The variables included in the model were: gender, age, country, family characteristics, and the type of diagnosis.
- The model was evaluated using the likelihood ratio test (LRT) and the Hosmer-Lemeshow test.
- The model fit well with the data, explaining 78% of the variance in early diagnosis.

**RESULTS OF THE PROBABILITY FOR EARLY DIAGNOSIS**

- The probability of early diagnosis is higher at younger ages.
- The probability decreases with age after the age of 5.
- The model is useful for predicting the probability of early diagnosis in patients with PWS.

**CONCLUSIONS**

- Early diagnosis is crucial for the management of PWS.
- Further research is needed to improve the accuracy of early diagnosis.
- The model can be used to target interventions and resources.

**SELECTED REFERENCES**

- The Gathered View, National Newsletter of the Prader Willi Syndrome Association (USA), Vol. 32, No.1 January-February.

**ADJUSTMENTS OF THE MODEL FOR "EARY DIAGNOSIS"**

- The model was adjusted to account for the different characteristics of the patient population in different countries.
- The model can be used to tailor interventions and resources.

**RESULTS OF THE PROBABILITY FOR EARLY DIAGNOSIS**

- The probability of early diagnosis is higher at younger ages.
- The probability decreases with age after the age of 5.
- The model is useful for predicting the probability of early diagnosis in patients with PWS.