DIAGNOSIS

The diagnosis of the PWS as early as possible is the key for a correct treatment of the conditions and may dramatically improve the quality of life of the patients. The basis is a correct clinical-diagnosis that may be based on age specific signs and symptoms like hyperphagia, failure to thrive in the first years of life, low stature, small head and face, characteristic facies, late in life by the hypogonadism and the neuropsychiatric signs (Table 2). The molecular diagnosis confirms the clinical suspect and can be performed by different techniques (Table 1). Among the tests available the most cost effective is definitely methylation specific PCR. The downside of this technique is that it detects the PWS but not the underlying cause. FISH diagnostic detects only the deletions but not the other causes of PWS. Besides this FISH requires working with cells that once the costs and it is laborious to do for foreign patients. STR studies offer a means to detect the presence of the sole ICP having also the parents DNA. A newer method is methylation specific MLPA that is able not only to confirm the diagnosis of PWS but also to detect deletions on the molecular level.

MS-MLPA TECHNIQUE

MS-MLPA is a MRC-Holland® kit from MRC-Holland® (Figure 2 and 3) allows the simultaneous determination of copy number variation and DNA methylation status in a semiquantitative fashion. It consists of a probe mix that contains 41 probes, 25 of which are specific to the PWS/AS critical region genes. Mix contains also 18 probes not related to the PWS/AS region. Among the probes specific for the PWS/AS region, 15 probes are methylation sensitive (including 1 control probe for methylation mapping on other chromosome) and contained a HhaI restriction site. MS-MLPA assay allows to detect gross deletions, molecular lesions most frequent in patients with PWS and AS. MS-MLPA is the breakdown method of choice for PWS diagnosis considering that it allows the investigation of methylation status of several loci, then reducing the risk of false positives or of false negatives due to SNPs; furthermore, if a probe fails there are other 4 probes to assess the methylation status. MS-MLPA is able to detect IC deletion in case of Prader-Willi patients with imprinting defects.

REHABILITATION AND TREATMENT

Current guidelines suggest a multidisciplinary approach that takes into account the different needs of the subject during different phases of life.

• In the first phase, from 0 to 3 years, the objectives are: reducing hyperphagia, sufficient nutrition, development of normal motricity both by quality and quantity.

• In the second phase, from 3 to 5 years (since the development of hypogonadism) the objectives are: weight control and correction of the daily calory intake, prevention and management of “secondary’’ pathologies, developing and maintaining autonomy.

• In the third phase, from 5 to 13 years (between the ages of 1800 kcal a day) we focus on autonomy, both feeding and playing with toys, facilitating a correct socialization.

• In the fourth phase of life, from 13 to adulthood, the objectives are: continuing the rehabilitation programs with the aim of maintaining the acquired autonomy, leading to the integration of the adult patient in society.

• Other objectives: preventing and treating problems related to the symptoms of PWS, as hypertension, hyperlipidemia, diabetes, dental caries, visual disturbances, endocrinological disturbances (hirsutism, acne), cutaneous and subcutaneous changes.

REHABILITATION CYCLES

B.I.R.D. Rehabilitation cycles

The main target of the rehabilitation cycles is giving PWS families some professional assistance while the PWS patients continue to live in the home. The Italian culture gives great importance to the family and PWS patients are often taken care of at home with one of the family members especially in the early years. PWS patients are followed on the medical and therapeutic as well as on the educational, psychological, social aspects but in the adulthood with the exception of the medical part the patients receive much less attention that is mostly concentrated in occupational therapy in day centers. The main goals are: treatment of obesity, occupational therapy with a medium diet, multiprofessional support, family therapy.

We offer these rehabilitation cycles for the PWS patients that receive much less attention that is mostly concentrated in occupational therapy in day centers. The main goals are: treatment of obesity, occupational therapy with a medium diet, multiprofessional support, family therapy.

The main targets of the rehabilitation cycles for adults are: treatment of obesity, occupational therapy with a medium diet, multiprofessional support, family therapy.

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CONCLUSIONS

The B.I.R.D. Rehabilitation cycles are a multidisciplinary approach, on one side is the diagnosis of the disease, clinical and molecular, on the other it is a “fighting” with the disease, living it with hormonal therapies, diet or education of all the persons that interact with a patient.

PWS demands a strict medical monitoring due to the many known complications and life threatening situations but primarily requires a strict control on their environment, in particular access to food sources.

Our institute discuss its attention in particular to the diagnosis, education of the families and professionals, treatment cycles for PWS adults.

We provide aid in the clinical diagnosis and offer molecular diagnosis that range from a 24 hour floppy infants screening in the first days of life, to MLPA tests, to free of charge methylation tests.

We furthermore offer counselling to families and others regarding the disease and its characteristics.

We also offer therapeutic cycles for adult PWS patients, where we concentrate in a three-four week period a custom diet with active medical and educational activities.

Our goal is simply to be a partner for the families who can have their questions answered and that can find them in all phases of life.