

phenotype and the youngest had amenorrhea with no breast development (B1) and pubic hair. Furthermore, according to physical examination, deafness was not mentioned.

**Conclusions** According to results, it seems that clinicians should consider different presentation for pure gonadal dysgenesis with familial pattern and further evaluation is needed in malignant degeneration of the gonadal dysgenesis in the patients with 46, XX PGD.

**PO-0094 ANTHROPOMETRIC CHARACTERISTICS AND BONE MINERAL DENSITY IN PATIENTS WITH PHENYLKETONURIA**

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**Background** Phenylketonuria (PKU) treatment requires a diet restricted from natural proteins and supplemented with phenylalanine (Phe)-free L-amino acid mixtures. Growth impairment and compromised bone mass have been described.

This study aims to evaluate anthropometric characteristics and bone mineral density (BMD) in a cohort of PKU patients.

**Methods** We conducted a retrospective longitudinal study collecting anthropometric characteristics (weight, height, body mass index (BMI) and BMD every 6 months from birth to 12 years of age in 34 patients with diet restrictions.

We compared the data results, expressed as z-scores, with the general population, as well as between patients with Phe <360 mmol/ml (optimal) and patients with >360 mmol/ml.

**Results** Our PKU patients are shorter than the reference population; the sample mean was below z-score=0. Weight was comparable to that of the reference population and BMI had a tendency to be over the population mean.

Growth impairment in PHA-deficiency is not related to plasma Phe concentration at birth but might be related to its levels throughout the follow up; patients with <360 mmol/ml were shorter.

BMD was below the population mean in all cases (52% osteopenia).

**Conclusions** PKU children in our study have a below than average height. Weight is consistent with the population average and BMI tends to be above it.

Height seems more affected among those patients with better metabolic control.

BMD is below the population mean. These data do not vary depending on the levels of Phe at diagnosis, but by the phenylalaninemia during growth.

Further studies are needed to investigate the effect of diet restriction in PKU.

**PO-0095 EARLY PROGRAMMING OF AEROBIC AND NEUROMUSCULAR FITNESS AT PRIMARY SCHOOL AGE. THE ABCD-STUDY**

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**Background and aims** Low birth weight and accelerated postnatal growth are associated with adult cardiovascular disease. In this perspective body composition and obesity can result from a disturbed energy balance due to early reprogramming of energy intake and expenditure. We hypothesise that low birth weight and accelerated growth may predispose (“program”) reduced physical fitness at 8–9 years of age.

**Methods** Aerobic fitness was measured using a 20 metre multi-stage shuttle run test (20m-MSRT) and neuromuscular fitness using the standing broad jump (SBJ) test and handgrip strength test was measured in 194 children (104 boys) of Dutch ethnicity at mean age 8.6 years in a prospective birth cohort.

**Results** Subjects with low birth weight and accelerated infant growth reached mean ( $\pm$ SD) 20m-MSRT levels of 3.9 which was significantly lower than (1) normal birth weight and normal infant growth (2) low birth weight and normal infant growth and (3) normal birth weight and accelerated infant growth groups (all  $p < 0.01$ ). Low birth weight subjects had mean grip strength of 12.3 kg ( $\pm$ 3.0), which was significantly lower than normal weight subjects with no effect of infant growth on this relationship. There was no association of birth weight or infant growth with grip strength or SBJ.

**Conclusions** Low birth weight with accelerated infant weight gain was associated with diminished aerobic fitness. Higher birth weight was associated with increased neuromuscular fitness. These early changes may explain increased susceptibility to obesity and related risk factors in low birth weight and early growth accelerated children.

**PO-0096 CLINICAL EFFECTIVENESS OF IDURSULFASE IN BOYS AGED 0–5 YEARS WITH HUNTER SYNDROME: 3-YEAR DATA FROM THE HUNTER OUTCOME SURVEY**

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**Background and aims** Symptoms of Hunter syndrome typically become apparent at 2–4 years of age. Previous analyses have demonstrated improvements in certain clinical measures in young patients receiving idursulfase (Shire); however, data on long-term idursulfase use in these patients remain limited. This analysis used data available in the Hunter Outcome Survey (HOS), a global, observational registry sponsored by Shire, to investigate long-term effectiveness of idursulfase in boys with Hunter syndrome aged 0–5 years.

**Methods** As of January 2014, 260/564 males followed prospectively in HOS had received  $\geq 1$  idursulfase infusion (excluding those who had received a bone marrow transplant or were enrolled in the TKT018/TKT024 clinical trials), were aged 0–5 years at treatment initiation and were included in the analysis. Median age at first treatment was 3.5 years; median treatment duration was 41.6 months. Clinical measures recorded in HOS at annual timepoints over 3 years were compared with baseline values.

**Results** Median urinary glycosaminoglycan (uGAG) levels, liver size and left ventricular mass index had improved at all yearly