**Table I-1. Overview of Maternal PKU Collaborative Study (MPKUCS) papers**

| **Author, Year** | | **Title** | **Study Date** | **Countries** | | **# of Women** | **# of Pregnancies** | **Cognitive Outcomes Reported** |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Studies Reporting Cognitive Outcomes** | | | | | | | | |
| Rouse et al., 20041 | Effect of High Maternal Blood Phenylalanine on Offspring Congenital Anomalies and Developmental Outcome at Ages 4 and 6 Years: The Importance of Strict Dietary Control Preconception and Throughout Pregnancy | | NR | | US  Canada | 526  100 matched controls | 576  413 live births | * McCarthy at 4 yrs * WISC-R at 6 yrs |
| Guttler et al., 20032 | Impact of the Phenylalanine Hydroxylase Gene on Maternal Phenylketonuria Outcome | | NR | | US  Canada  Germany | 196 fully evaluated;  236 genotyped | 308 pregnancies;  253 live births;  196 offspring fully evaluated | * WISC-R at 6-7 yrs or later |
| Levy et al., 20033 | Pregnancy Experiences in the Woman With Mild Hyperphenylalaninemia | | NR | | US  Canada  Germany | 48 | 58 | * WISC-R at 2 years * McCarthy Scales of Children’s Abilities * Bayley Scales of Infant Development |
| Waisbren et al., 20034 | Cognitive and Behavioral Development in Maternal Phenylketonuria Offspring | | NR | | US  Canada  Germany | NR | NR;  228 offspring;  70 controls | * WISC-R at 7 years * Test of Language Development, 2nd edition * Peabody Individual Achievement Test-Revised * Stroop Interference * Visual Motor Integration * Child Behavior Checklist |
| Koch et al., 20035 | The Maternal Phenylketonuria International Study: 1984-2002 | | 1984-2002 | | US  Canada  Germany | 382 | 572 | * McCarthy at 4yrs * WISC-R at 7 yrs * WISC-R at 10 yrs |
| Widaman et al., 20036 | Relation of Prenatal Phenylalanine Exposure to Infant and Childhood Cognitive Outcomes: Results From the International Maternal PKU Collaborative Study | | NR | | US  Canada  Germany | NR | 572 pregnancies;  413 offspring evaluated | * Bayley at 1 year * McCarthy at 4 years * TOLD CSLQ at 4 years * WISC-R at 7 years |
| Antshel et al., 20037, 8 | Developmental Timing of Exposure to Elevated Levels of Phenylalanine is Associated with ADHD Symptom Expression | | NR | | US | NR | 15 MPKU offspring;  46 PKU patients;  18 controls | * ADHD symptoms * FSIQ |
| Platt et al., 20009 | The International Study of Pregnancy Outcome in Women with Maternal Phenylketonuria: Report of a 12-year study | | 1984-1999 | | US  Canada  Germany | PKU NR;  101 controls | 576 pregnancies;  414 live births | * Bayley at 2 yrs * McCarthy at 4-5 yrs * WISC-R data presented on children who had reached age 7 |
| Koch et al., 200010 | The international collaborative study of maternal phenylketonuria: status report 1998 | | 1984-1998 | | US  Canada | NR | 572 HPA  99 control | * Congenital anomalies * McCarthy at 4 years |
| Waisbren et al., 200011 | Outcome at Age 4 Years in Offspring of Women With Maternal Phenylketonuria: The Maternal PKU Collaborative Study | | 1984-2000 | | US  Canada | 205  253 offspring received preschool evaluation | 572 pregnancies;  412 offspring | * McCarthy at 3-5 years * Test of language development at 4 years * Achenbach at 4 years * Vineland at 4 years * Bayley at 2 yrs |
| Waisbren, et al., 199812 | Neonatal neurological assessment of offspring in maternal phenylketonuria | | NR | | US | NR | 56 PKU offspring;  45 control offspring | * Dubowitz Neurological Assessment of the Preterm and Full-Term Newborn Infant * Bayley Mental and Motor Scales at 1 year * Receptive-Expressive Emergent Language Scales at 1 year |
| Hanley et al., 199613 | The North American Maternal Phenylketonuria Collaborative Study, developmental assessment of the offspring: preliminary report | | 1984-1994 | | US  Canada | NR | 134 HPA offspring;  58 control offspring | * Bayley at 2 yrs * McCarthy at 4 yrs |
| Cipcic-Schmidt et al., 199614 | German Maternal Phenylketonuria Study | | 1989-1994, worked with North American study since 1992 | | Germany  Austria | 275 | 43 pregnancies;  34 live births | * Bayley tests at 2 yrs |
| Koch et al., 199415 | The international collaborative study of maternal phenylketonuria: status report 1994 | | 1984-1994 (US)  1986-1994 (Canada)  1993-1994 (Germany) | | US  Canada  Germany | NR | 402 pregnancies;  99 control | * Bayley Scales and McCarthy at 3.5 to 4.5 yrs |
| Koch et al., 199316 | The North American Collaborative Study of Maternal Phenylketonuria: Status Report 1993 | | 1984-1992 | | US  Canada | 379 HPA | 318 HPA;  59 control | * Bayley scale at 12-24 mos * McCarthy Scale between 3.5-5yrs |

ADHD=attention deficit hyperactivity disorder; FSIQ=full scale intelligence quotient; MPKU=maternal phenylketonuria; NR=not reported; PKU=phenylketonuria; TOLD CSLQ=Test of Language Development Spoken Language Component; WISC-R=Wechsler Intelligence Scale for Children-Revised