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This thesis describes the prevalence rates of testicular microlithiasis in symptomatic and asymptomatic boys, testicular growth in boys with acquired undescended testis managed with a conservative attitude, and the volume of retractile testes.

In Chapter 1 the prevalence of testicular microlithiasis and its relation with benign and malignant tumours is assessed after reviewing the paediatric literature. Moreover, a guideline is provided for the management of boys with testicular microlithiasis. We found that the prevalence rate of testicular microlithiasis is 4.2% in asymptomatic boys and 1.6% in symptomatic boys. The development of a testicular malignancy is occasionally reported after testicular microlithiasis has been diagnosed. The management of boys with testicular microlithiasis varies widely. Most authors advise regular self-examination and some perform testicular ultrasound and/or screen tumour markers. After reviewing the literature for follow-up, we advise regular self-examination from the age of 15.

Chapter 2 discusses testicular microlithiasis in asymptomatic and symptomatic boys. Firstly, we describe three boys with testicular microlithiasis, two who were free of symptoms and one who suffered from testicular pain. In the second part, the prevalence of testicular microlithiasis is assessed by means of ultrasound in asymptomatic males of 0 – 19 years old. We examined 694 asymptomatic boys of whom 670 participated in the study. The results showed classical testicular microlithiasis to be present in 16 boys (2.4 %) and limited testicular microlithiasis in 12 boys (1.8 %), thus yielding a total prevalence of 4.2 %. We also found that prevalence of testicular microlithiasis increases with age. Testicular malignancies were not found in any patient.

The third part retrospectively describes the prevalence of testicular microlithiasis in males of 0 – 18 years old who had been referred for scrotal ultrasound. After scanning the data of 363 patients, testicular microlithiasis was found in 21 patients (5.8%), the mean ages of the patients with and without testicular microlithiasis were 11.3 and 6.7 years, respectively. Significantly more images were available for patients with testicular microlithiasis and only 8 of the 21 cases (38.1%) had been identified by the consultant radiologists. None of the boys with testicular microlithiasis were found to have a testicular malignancy. The results indicate that the consultant radiologists are advised to be even more vigilant towards testicular microlithiasis and sufficient planes should be scanned and stored.

In the fourth part, we assess the prevalence of testicular microlithiasis by ultrasound in boys and young men with congenital or acquired undescended testis. We performed 181 ultrasounds in 181 patients with congenital undescended testis and 636 ultrasounds
in 320 patients with acquired undescended testis. Testicular microlithiasis was found in 14 patients (2.8%), of whom 11 (2.2%) had classical testicular microlithiasis and 3 (0.6%) had limited testicular microlithiasis. No difference in the prevalence of testicular microlithiasis was found between congenital and acquired undescended testis.

The fifth part describes a prospective ultrasound study in which the prevalence of testicular microlithiasis was determined in boys with Down syndrome of 0 – 18 years old. In addition, we compared testicular size with normative values. Testicular ultrasound was performed in 79 boys with Down syndrome and testicular microlithiasis was present in 22.8% of the boys. It was diagnosed in 21.4% of the boys < 7 years old, in 21.4% of the boys between 7 and 12 years old and in 26.1% of the boys ≥ 12 years old. There was no significant difference in the prevalence of testicular microlithiasis among the three groups. The mean testicular volumes of patients with Down syndrome were significantly smaller than the normative values in boys aged < 7 years old and aged ≥ 12 years old.

The final part of Chapter 2 describes two case reports. The first report involves a 9-year-old boy with pseudoxanthoma elasticum, who already had testicular microlithiasis at the age of two. To our knowledge, this is the youngest patient with pseudoxanthoma elasticum in whom testicular microlithiasis has been documented. The other report describes two cases of testicular microlithiasis in a 9-year-old and a 13-year-old boy who had previously been diagnosed with 18q- syndrome and subtelomeric 11q- deletion, respectively. We think that this is the first documentation of patients having both these chromosomal abnormalities and also testicular microlithiasis.

Chapter 3 describes the testicular volumes of healthy boys and of boys with retractile testis. In the first part we obtained reference data for testicular volume measured by ultrasound in asymptomatic boys of 0.5 – 18 years old. In addition, we assessed the validity of the Prader orchidometer per age group by correlating it with the volume measurement by ultrasound. The results showed a statistically significant correlation in the volume measurements by the Prader orchidometer according to reference curves. Moreover, we showed that the measurements of testicular volumes by the Prader orchidometer had an accurate goodness of fit with measurements by ultrasound.

The second part of the chapter discusses the volume of retractile testes. We used ultrasound to determine the volume of retractile testes in boys, and compared these volumes with normative values. The results showed that the volumes for the boys with retractile testes were significantly smaller than normative values (p < 0.001). Furthermore, the testicular volumes of retractile testes measured in inguinal position were significantly smaller than the testicular volumes measured in scrotal position (p < 0.001). We concluded that volumes of retractile testes are significantly smaller than normative values.
Chapter 4 presents some aspects of acquired undescended testis. In the first part of the chapter the natural history and long-term testicular growth of acquired undescended testis is assessed prospectively after spontaneous descent or pubertal orchidopexy in case of non-descent. A total of 391 boys with 464 acquired undescended testes were included in the study. In accordance with Dutch consensus on non-scrotal testis, spontaneous descent at puberty was awaited; if this did not happen, orchidopexy was performed at puberty. The results show that in 77.5% there was spontaneous descent at puberty; in the other 22.5%, pubertal orchidopexy was performed due to non-descent. Of the 494 testis volume measurements after spontaneous descent, 92.7% were at ≥ 10th centile for age of which 63.0% were ≥ 50th centile, and 21.7% ≥ 90th centile. After pubertal orchidopexy for non-descent, of the 85 measurements 92.9% were at ≥ 10th centile, 62.4% ≥ 50th centile, and 14.1% ≥ 90th centile. In unilateral cases, after spontaneous descent 59.2% of the retained testes were found to be smaller than their counterpart and 30.6% were equal in size. After pubertal orchidopexy in unilateral cases, 78.4% of testes were smaller, and 17.6% were equal in size. We concluded that acquired undescended testis has a 77.5% tendency of spontaneous descent at puberty and in nearly all cases, after spontaneous descent as well as after pubertal orchidopexy, long-term testicular growth is within the normal range.

The second part of Chapter 4 provides an overview of the current views on acquired undescended testis. We speculate that acquired undescended testis is in fact congenital: it is the result of a short funiculus at birth, which allows a low scrotal position early in life. However, as the boy grows, the testis evolves into an undescended state. As a result of gonadal stimulation at puberty, spontaneous descent occurs in three of every four cases.