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novel dystrophin-related protein (DYSB)
gene mutation in Italian patients with a
proximal form of congenital muscular
dystrophy. Congenital muscular
dystrophies are a heterogeneous group of
inherited disorders characterized by
impaired muscle function. A severe form of
skeletal muscle weakness and a specific
pattern of inheritance were defined in a
previously reported Italian family in which
a recessive form of congenital muscular
dystrophy was observed. The analysis of
the muscular dystrophy in an affected
patient's sister provided a direct evidence

for a causative role of the genetic defect.

The affected patients showed a unique dystrophic pathology characterized by a lack of connective tissue within the muscle fibers. The molecular pathogenesis of the muscular dystrophy was investigated by genomic and expression analysis in the

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is the most precise known parameter in sex

determination. Indeed, analysis of the full Y chromosome has shown that there is variation in the number of repeats and other features, which complicate precise analysis. As an example, the SRY sequence is found in only half the copies. Analysis of the unique sequence of the gene OCT1 has further proven problematic. As another example, the XIST gene is not located on the sex chromosomes, but instead on the X chromosome. This gene is expressed in females, and has been implicated in the inactivation of the X chromosome in females, yet its role in males has not been clearly defined. Additionally, genes found on the Y chromosome have not been shown to have a role in human sexual development. (Li, Yang et al. (2001) Nature 412:710-716). The Sry gene located on the Y chromosome is responsible for development of the testes in males. Many, but not all, sex-linked traits in humans are not due to mutations in the SRY gene.

Because SRY is a testis-determining factor in the male, the majority of mutations affecting sex determination in humans occur outside the SRY gene and the testis-determining pathway. Given that SRY is a direct determinant of spermatogenesis in males, a number of characteristics of the SRY gene and the Y chromosome suggest that mutations in SRY might cause defects in sperm maturation. Initial evidence for defects in sperm came from the analysis of sex-linked aneuploidy, which is more common in sons of fathers with mutations in the SRY gene. Polymorphisms in the SRY gene have been observed in all human populations studied, indicating a strong evolutionary pressure. In addition, mutations in the SRY gene are a major cause of male infertility and affect 10% of infertile men. These observations have led to the suggestion that SRY function in spermatogenesis may have led to a constraint on its evolution and a

requirement for very tight regulation. (Zhu, Luu et al. (1997) Hum. Mol. Genet. 6:1773-1776). In a similar manner, mutations in the FOXL2 gene on the X chromosome have been shown to lead to malformations of the ovaries, including premature ovarian failure. (WO 99/24455). Additionally, mutations in the WNT4 gene on the X chromosome are associated with sex reversal in humans and mice, and mutations in

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Easy Boiling Apple Pudding with Vanilla Sauce

I like to make this recipe because it's easy to make and takes about one and a half hours. It's one of those recipes that doesn't have too many dishes to do and each portion is enough.

1. If the apples are not perfectly smooth, mash them in a food processor. Once they're smooth, add the sugar, salt and cinnamon and mix until well incorporated. If you're not using a food processor, mash them with your hand.
2. Pour the milk in a large heat-proof bowl, and then arrange the apples in the milk and mix well.
3. In a small bowl, whisk together the flours and baking powder. Pour into a separate bowl, and mix the eggs into the flour.
4. Heat the oven to 180°C (350°F). Grease a medium baking dish with butter.
5. Pour the baking mixture into the baking dish and spread evenly. Sprinkle with sugar.
6. Bake for

50-60 minutes, until a knife inserted comes out clean. 7. To make the vanilla sauce, beat together the butter and sugar in a food processor until mixed well and creamy. Reduce the speed and add the flour and cornflour (this will make the sauce smooth). Add the vanilla extract. If the sauce is too thin, add the milk a little at a time and pulse until the sauce is thick. 8. Remove from the oven and turn the oven down to 160°C (325°F). Spread the sauce on top of the apple pudding. Return to the oven and bake for 5 more minutes. 9. Serve with vanilla ice cream (I used vanilla ice cream). Tip: Once your vanilla sauce is prepared, it can be stored in the fridge for a day or two. This recipe is suitable for diabetic and/or low-cholesterol diets. 1. Field of the Invention The invention is directed to a cam locking device for locking a cam wheel of a rotary friction lock in the open position. 2. Description of the Prior Art Many emergency or escape exits are

covered by cam locks or the like which
require manual operation