

When did recombination suppression events occur in bird ZW sex chromosomes?

Deborah Charlesworth



A recent paper found that the sex chromosomes of the crested ibis have more gametolog pairs than many other birds. This Comment discusses that this finding suggests that WZ recombination stopped independently in the sex chromosomes in different bird lineages.

A recent paper¹ describes two interesting findings. The first is support for previous results suggesting that the W chromosome of the crested ibis might carry many of the genes found in its Z chromosome, whereas the Ws of other Neognathous birds so far studied have extremely few such genes. The paper also describes evidence that, like some other birds, this species has a sex chromosome-autosome fusion. A recent fusion could, in principle, explain the first result, but probably cannot explain it fully, leaving an intriguing puzzle with two alternative resolutions (Box 1), either of them with surprising implications about bird sex chromosome evolution. This Comment discusses these possibilities, and the implications for the timing and independent occurrences of recombination suppression events in bird ZW sex chromosomes.

Sex-linked regions in birds

All birds, both the Neognaths and the Palaeognathous groups (including emu, ostrich and tinamous), have ZW sex chromosome systems (as recently reviewed^{2–4}), and the ZW pair is a homologous macrochromosome in all studied species (although sometimes an autosome, usually a small chromosome, has fused to it, creating a slightly larger W or Z, or both, as discussed below). In most Neognathous birds, almost the whole Z chromosome is completely non-recombining in females. The “differential” regions of the homologous W chromosomes lack almost all genes carried on the Z chromosome counterparts; like the highly degenerate mammal Y, the chicken W is about 3 times smaller than its homologue, and only a small “pseudo-autosomal” region (PAR) recombines in the heterogametic sex and has diploid gene coverage in both sexes. Among Palaeognathous birds, the differential region sizes differ greatly⁵, with over half of the ostrich Z chromosome being pseudoautosomal, a bit more in the emu (but only 21% in the related kiwi), about half in some tinamous, and only 3% in another tinamou lineage⁶, implying that expansions of the fully sex-linked regions have evolved (as diagrammed in Fig. 1).

Similar expansions in mammal XY systems were first detected as “evolutionary strata”; the genomic locations of the genes present in the ancestral state (and usually in the X chromosome) revealed contiguous regions, termed strata, carrying Y-X gene pairs (gametologs) with different divergence levels⁷. In therians (the placental or Eutherian mammals and the marsupials), the first Y-linked region, termed S1, appeared after the actively male-determining SRY gene evolved, about

180 million years ago, and new strata subsequently evolved, one in marsupials, and four more independently in Eutherians⁸ after an autosome became fused with the X and Y⁹. In all these strata, very few X chromosome genes have Y-linked alleles¹⁰ demonstrating profound Y chromosome degeneration.

Strata have also been found in at least one fish¹¹, schistosomes¹², and a few plants, indicating that similar sex chromosome changes have repeatedly evolved. Species and lineages in which strata have evolved are important because they indicate that recombination has been suppressed. However, even rough degeneration data, or stratum age information, are scarce. Thus the reason(s) for recombination changes in sex chromosomes, expanding their fully Y or W-linked regions, are not currently understood (reviewed in refs. 13–15). In birds, studies of different species’ Z chromosomes suggest that they carry very similar gene sets (unless a fusion has occurred), but different parts are completely W-linked (Fig. 1). This allows the evolution of sex linkage, and its consequences, to be compared for the same gene sets at different points in the process.

Genetic degeneration in evolutionary strata

Degeneration occurs in both XY systems and species like birds, with female heterogamety (ZW systems), based on observed low proportions of differential region X or Z genes retaining Y- or W-linked copies. Proportions of genes whose sequences are incomplete, or lack start codons or include premature stop codons, or other signs of being pseudogenes, are also informative, but are even scarcer in the published literature than presence/absence data. The number of generations during which genetic degeneration has been evolving in each stratum can be estimated using Y-X or Z-W divergence, given enough genes that retain Y- or W-linked sequences (termed gametolog pairs). Synonymous mutations, or mutations at fourfold degenerate sites, are ideal, as they evolve similarly to neutral variants, for which divergence accumulates linearly over generations¹⁶. If close relatives are available, inter-species divergence can be used to correct for possible rate differences between strata. The use of all site types is problematic, as rates will differ for different strata (which include different proportions of sequences under selective constraints). Failing reliable dating by this approach, the times of events that suppressed recombination can be ordered in a phylogeny based on the accumulation of sex-specific sequence variants, and nodes in the phylogeny can be dated¹⁷. An advantage is that variants of any site type can be used, providing more information than synonymous mutations, though both approaches are limited by gametolog gene pair availability.

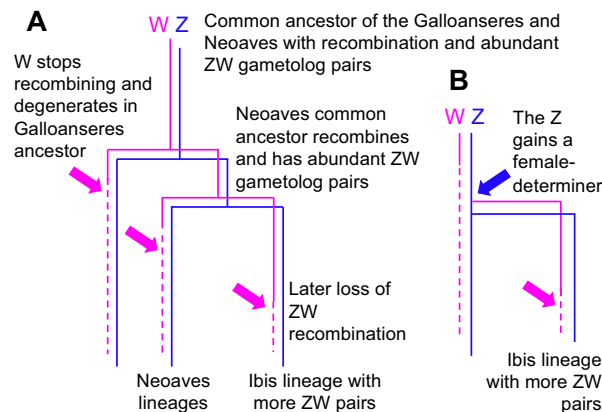
Genome sequences and divergence analyses or analyses to detect W-specific sequence variants, indicate that, as in mammals, new strata have evolved independently in different bird lineages. The oldest stratum in Palaeognaths evolved at about the same time as Eutherian strata S2a and S3⁸. In most Neognaths, profound degeneration leaves few gametolog pairs for reliable divergence estimates in the early-

BOX 1

Two alternative possibilities to explain how the ibis lineage W chromosome could carry more homologues of Z-linked genes, compared with other Neoaves lineages. The ZW pair in the common ancestor of the Galloanseres and Neoaves is thought to have been (largely) recombining, and to have carried many ZW gametolog pairs. Recombination stopped in the Galloanseres (which then lost most ZW gametolog pairs and now have very few pairs), but continued in the ancestor of the Neoaves, as shown in diagram **A**, which shows the first possibility. In this scenario, recombination later stopped independently in different Neoaves lineages, at different times, explaining their different numbers of gametolog pairs. The lineage leading to the ibis continued recombining after the ancestor of the Neoaves split from the Galloanseres and before other Neoaves lineages evolved suppressed recombination, explaining why the crested ibis has even less gene loss, or degeneration, than others. Part **B** shows the second alternative, in which a common Neoaves ancestor had lost W-Z recombination and degenerated, but a turnover event subsequently occurred in the ibis lineage, involving the evolution of a new Y chromosome from an ancestral X, followed by independent loss of recombination and degeneration

A. Different lineages underwent independent events causing loss of recombination between the Z and W chromosomes, as indicated by pink arrows; the W lineages after such events are shown as dashed lines, to indicate loss of W-linked genes (genetic degeneration).

B. After a common Neoaves ancestor had lost W-Z recombination and degenerated, the ibis lineage Z gained a female-determining factor (blue arrow), becoming a non-degenerated “neo-W”, which later stopped recombining with the Z and started degenerating independently.



branching group Galloanseres that includes the chicken and duck¹⁸, though songbirds, and also in the later-branching Neoaves (about 95% of bird species)¹⁹, may have a few more²⁰.

Comparisons of numbers of gametolog pairs in homologous W-linked regions of different species can suggest that recombination stopped in some of them, but continued in those with more gametolog pairs. The great bustard, *Otis tarda*, appears to have evolved a smaller PAR than its relatives, indicating the evolution of a new fully sex-linked region without involving a chromosome fusion²¹. In the reed warbler, phylogenetic analysis detected and dated a low recombination rate region in a former autosome after fusion to the sex chromosomes¹⁷, but failed to detect strata in the ancestral ZW part, possibly because of high divergence.

Two Neoaves species, the crested ibis and white-tailed tropicbird (*Phaethon lepturus*), both in the order Phaethoqornithes¹⁹, have been suggested to have more gametolog pairs than other birds²². The new study of the crested ibis confirms this using long-read sequencing¹. 175 non-redundant gametologous genes were found on the W (representing 19.1% of Z genes; the W total of 414 genes includes many duplicates). Both the Z and W have a micro-chromosome (22) fused to the end with the oldest stratum. This created a second PAR, carrying 154 genes, of which a part now has low coverage in females, indicating

loss of recombination (as found for other such fusions²³) and some degeneration. This neo-Z part carries 53 genes, of which 32 (60%) are detectable on the neo-W. Excluding these 32 neo-W genes, the older ibis W part retains a total of around 140 ancestral Z genes, many more than other Neognathous birds, though only the chicken W-linked region, with 28 genes, is currently based on long-read sequencing³. The gene-rich ibis W, therefore, probably does not simply reflect better ascertainment through long-read sequencing. The difference even affects the two oldest strata; in chicken, these include only 6 W-linked genes, versus 43 in the crested ibis.

Recombination suppression in bird lineages

Once a bird W chromosome region has stopped recombining, gene losses should accumulate irreversibly over evolutionary time, and species with more genes in a W-linked region are unlikely to have evolved from one with fewer. It, therefore, seems possible that recombination in the Galloanseres (with very few ZW gametolog pairs) stopped independently of the later-branching Neoaves (some with more gametolog pairs). If the lineage leading to the ibis continued recombining after the split from the Galloanseres, different Neoaves lineages might then subsequently have evolved suppressed recombination independently, with the lineage that includes the crested ibis

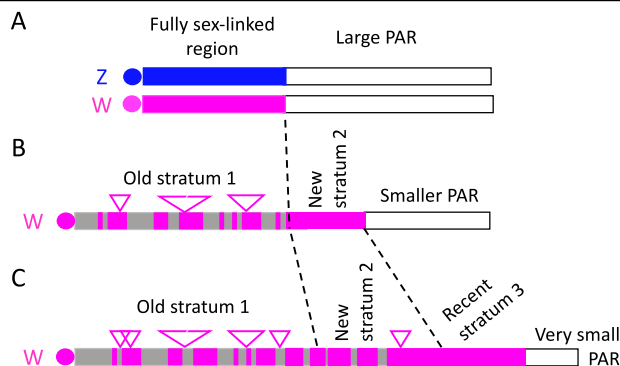


Fig. 1 | Sex chromosome evolution in a lineage with female heterogamety (a ZW system in which the females are ZW heterozygotes), diagramming understanding of bird ZW pairs from several taxa described in refs. 17–22. **A** A young ZW chromosome pair with a centromere at the left-hand end (indicated by a filled circle) and a large pseudo-autosomal region (PAR) in which crossovers occur at the other end. A non-recombining W-linked region has evolved, in which W-specific variants have accumulated (symbolised by pink in the W diagram), but it is not highly diverged, and its size, gene content and arrangement are still similar to those of the Z, though it differs by the presence of a female-determining locus within the region. **B** The homologous W chromosome arm, after a subsequent recombination suppression event, creates a new stratum (the new pink region to the right of the dashed vertical line). The initial W-linked region has degenerated (grey indicates gene losses) and expanded by accumulating repetitive sequences (pink triangles), making the W longer than the Z. The new fully sex-linked region, Stratum 2, has gained some W-specific sequences but has not yet degenerated. This change leaves a smaller PAR at the chromosome end. **C** Another recombination suppression event has occurred across a new region, further shortening the PAR, and creating a 3rd stratum that has not yet degenerated, while stratum 2 has started degenerating and expanding.

having even less gene loss, or degeneration, than others. This scenario is not impossible, as the ordering of the early radiation of Neoaves is not definitively resolved¹⁹. Short branches will make it difficult for any approach to tell whether or not recombination suppression events occurred in a common ancestor or independently in the short evolutionary times after lineages split from a common ancestor before they split again to form further descendant lineages (see diagram A in Box 1). The possibility suggested here was already envisaged, and previous analyses^{4,24} hinted at several independent recombination suppression events within the Neognathae, and even possible ongoing recombination in the brown pelican, a relative of the ibis, and in the white-tailed tropicbird and lesser flamingo.

An alternative possibility is a turnover event like that proposed in the guppy, involving the evolution of a new Y chromosome from an ancestral X²⁵. Such a change might be followed by independent loss of recombination, and degeneration. Although it seems unlikely that a new W chromosome could evolve from a Z in a bird, studies in more Neoaves lineages are needed to test the possibilities.

In conclusion, these new results suggest that the Neognathous birds, not just the Paleognaths, may have undergone several events creating independent strata corresponding to similar, but perhaps not identical, Z chromosome regions. Together with non-recombining regions created by fusions with autosomes in several lineages, including in the crested ibis⁴, birds may have fully sex-linked regions of many different ages and may be ideal for studying the evolution of suppressed recombination, and subsequent genetic degeneration of

the W-linked regions created. Long-read sequencing data should allow ascertainment of more ZW-linked gene pairs in previously studied taxa, enabling the times of that recombination suppression events to be ordered in a phylogeny, by the approaches mentioned above, e.g., ref. 17. This may more clearly establish strata boundaries in birds, whether they differ between species, and how many such events occurred in different lineages.

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Comment

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D.C. wrote the Comment.

Competing interests

The author declares no competing interests.

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